ENGAGING WITH CHARCOT-MARIE-TOOTH DISEASE: A GROUNDED THEORY APPROACH

by

NICOLAAS WILLEM ALBERTS

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ABSTRACT

This qualitative study focuses on the experiences of adults with Charcot-Marie-Tooth disease (CMT), a neuromuscular condition, and explores what living with this disease encompasses. The study is structured around two fundamental research questions that amount to people's experiences regarding how (in which areas) the disease affects them, and how they continuously deal with it.

In order to address the research questions, data gathered from participants was qualitatively analysed, using grounded theory methodology. The study culminated in the formulation of a substantive grounded theory as to how affected people manage the disease's manifestations in order to optimise their continuous adaptation and well-being. A tripartite of concerns comprised the core concern, whereas the basic social psychological process of *engaging with CMT* emerged as the core strategy used by affected people to deal with the concerns. The core's three sub processes constituted three mostly sequential stages that CMT-affected people pass through in their adaptation to the disease. The identified theory and existing stage models of adaptation to chronic illnesses and disabilities were juxtaposed and discussed. The three stages were compared to and integrated with the relevant literature. These actions revealed that there are a number of new formulations and processes contained in all three stages, and that the first and last stages (*orientating* and *optimising*) are themselves unique. It emerged that there is no theoretical end-point to the adaptation process, but that a relative saturation point amounted to a variant of an outcome, called qualified wellness. For most, the core strategy was successful in resolving the main concern. A few, however, still experienced fear and agony about inheritance and dependency issues.

This study contributes, via the route of knowledge and insight empowerment, to the well-being of people with CMT, including those who are struggling but do not know that they have this disease. Broadening of insight may also benefit medical help professionals and streamline service delivery.

Key words: Charcot-Marie-Tooth disease, grounded theory, adaptation to chronic illnesses and disabilities, coping, management, stages, well-being.

Dedicated to the memory of my beloved

Mother, Kitta (8.4.22 - 6.8.2003)

To my fellow CMT-affected individuals:

"*It is not what you have lost that counts, but what you have left"*
(Anon)

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"Our life is what our thoughts make it"

(Marcus Aurelius)

ABBREVIATIONS

BDI	Beck Depression Inventory
CID	Chronic illness and disability
CMT	Charcot-Marie-Tooth disease
CNS	Central nervous system
DSD	Dejerine-Sottas disease
GT	Grounded theory
GTM	Grounded theory methods
HMSN	Hereditary Motor and Sensory Neuropathy
HRQOL	Health related quality of life
MD	Muscular dystrophy
MS	Multiple sclerosis
NCV	Nerve conduction velocity
PGD	Preimplantation genetic diagnosis
PN	Peripheral neuropathy
PNS	Peripheral nervous system
PSS	Perceived Stress scale
QOL	Quality of life
RLS	Restless legs syndrome
RSE	Rosenberg Self-esteem scale
SF-36	Medical Outcome Study Short Form-36
SWB	Subjective well-being
WHO	World Health Organisation
WHOQOL-BREF	World Health Organisation Quality of Life Assessment - Abbreviated Version

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CHAPTER 1

INTRODUCTION

This study focuses on people with a rather unfamiliar disease called Charcot-Marie-Tooth neuropathy (CMT). CMT may be described as an inherited, lifelong neuromuscular disease that is rarely fatal, but that impairs affected people's quality of life to various degrees (Northern, 2000).

Throughout this document, people who have this disease will be referred to as CMT-affected people (or a person). Since the individual with the disease usually functions within a family or similar system, it would be technically correct to reason that the disease also indirectly affects his or her family members. However, this study focuses on the individual adults themselves, that is to say, those who are physically affected by the disease. "Affected people" or "CMT-affected people" therefore refer to these individuals only, unless otherwise specified.

In order to investigate the experiences and behaviour of CMT-affected people in the context of living with the disease, a qualitative study was embarked upon. In this qualitative study I, who have been diagnosed with CMT and who have lived with it for my entire life, have also participated. In this study, I therefore occupy the dual role of researcher and participant.

The enquiry into the disease-related experiences and actions of affected people will be structured around the following two research questions:

1. *In what areas, and how, does CMT affect the lives of those who have the disease, according to their own frame of reference?*
2. *How do people who have CMT manage the manifestations, effects, implications and challenges that stem from this disease, in order to augment their well-being?*

The basic nature of CMT disease, including its salient manifestations and effects, will be briefly introduced in this chapter, whilst a more in-depth elucidation of the neuropathy will be undertaken in Chapter 2.

CHARCOT-MARIE-TOOTH DISEASE: A NEED FOR RESEARCH

Charcot-Marie-Tooth disease is an inherited neuropathy that embraces a heterogeneous group of neuromuscular conditions. Notwithstanding underlying physiological differences between many of the disorders in this group, all types produce remarkably similar symptoms and clinical presentations (CMT United Kingdom, 2004a; Kedlaya, 2007). The disease was named after the three neurologists who first systematically described it in 1886, namely Jean-Martin-Charcot, Pierre Marie and Howard Henry Tooth (Chudley, 2000). The disease is also known by another often-encountered name, Hereditary Motor and Sensory Neuropathy (HMSN), and by a now largely defunct name, Peroneal Muscular Atrophy (PMA) (CMT United Kingdom, 2004a).

Physiologically speaking, CMT refers to a group of slowly progressive disorders that primarily affect the body's peripheral nerves. The peripheral nerves connect the central nervous system (the brain and spinal cord) to muscles, joints and sensory organs, including the skin. Both the motor nerves (that carry messages from the brain and spinal cord to muscles and other end points) and the sensory nerves (that transport signals from the skin and other sensory organs to the spinal cord and brain) are affected by CMT and typically deteriorate slowly over many years, rather than abruptly (CMT United Kingdom, 2004 a&b; Kedlaya, 2007).

Fundamentally, the peripheral nerves comprise bundles of nerve fibres, which, metaphorically, resemble a network of electrical communication cables that branch throughout the entire body. Each nerve fibre consists of a cell body and extensions (fibres) of varying lengths. The axon is usually the long fibre that extends from the cell body, with the longest axons being up to 1 metre in length (the nerves that serve the feet muscles). In peripheral neuropathy, including CMT, these fibres are usually the primary location of the pathology. It is here where deterioration takes place over time. Typically, it is either the insulating

layer around the axon, or the axon itself that is at fault. In most cases, the muscles and other end point structures that are dependent upon the nerves, gradually deteriorate to various degrees (Guillain-Barré Syndrome [GBS] Support Group of the UK, 1999).

In CMT, the muscles that are served by the deteriorating (peripheral) motor nerves receive inadequate signals, causing them to atrophy. In this process, the muscles furthest away from the spinal cord/brain, such as those in the extremities like the feet, ankles, calves, forearms and hands, are usually affected first, and in most cases they are also affected the most profoundly. Deterioration in the **sensory nerves** typically results in feelings of numbness and lack of sensation (Chance,2001; GBS Support Group of the UK, 1999).

These physiological abnormalities lead to a vast array of symptoms and manifestations in CMT-affected people. Typical manifestations and effects of the disease include muscle wastage, physical weakness, chronic fatigue, walking and running difficulties, thin lower legs, high arched feet, absence of certain reflexes such as the knee jerk, poor balance, frequent falls to the ground, a characteristic high stepping gait and difficulties executing fine motoric tasks (Crabtree, 2000; Kedlaya, 2007).

There are rather specific underlying physiological processes and mechanisms involved in the various types of CMT, such as the ways in which the peripheral nerves are affected and the different ways in which they are inherited or genetically transmitted by means of the abnormal gene(s) that are responsible for the disease. These, as well as the classification of the different types of the disease and available treatments, will be expounded in Chapter 2. Even though CMT mostly does not affect life expectancy, the quality of life of people who have the disease are with very rare exception adversely affected by it, and in many cases profoundly so (Young & Suter, 2003).

The symptoms vary greatly, even between affected members of the same family. For some, CMT amounts to rather less debilitating symptoms, which causes only

moderate disruption in their lives. For many, various degrees of disability result from CMT. This may seriously impair quality of life in many areas, including activities of daily living (Padua et al., 2006). Actually, the disease is capable of, and in some cases does, cause severe disablement, leaving the affected person wheelchair bound and largely dependent on others. Reilly (1998, p.8) states that 20% of all CMT-affected people are seriously handicapped. Life expectancy may indeed be implicated in cases of phrenic nerve involvement (that serves the diaphragm muscle), in other words, where breathing is impaired. Some affected individuals may experience pain to various degrees (CMT United Kingdom, 2004b; Crabtree, 2001; Young & Suter, 2003).

It is deemed necessary to make a distinction between CMT and muscular dystrophy (MD) because many confuse the two and erroneously regard CMT as a form of MD. In muscular dystrophy, the defect is within the muscles itself, whereas in CMT the defect is in the peripheral nerves that supply the muscles. The muscles of CMT-affected people are technically "normal"; they atrophy because they do not receive adequate messages via the defective nerves.

In most forms of muscular dystrophy, the fundamental defect is in the membrane around muscle fibres. A protein called dystrophin, which plays a role in retaining the structure of the membrane, is either absent or abnormal. This results in muscle substances, particularly enzymes, leaking out, resulting in atrophy (Emery, 1994). In the most common type of CMT (type 1), the insulating layer around nerve fibres, called the myelin sheath, deteriorates, resulting in impaired conduction of signals. In certain types of CMT however, the nerve fibre itself (the axon), rather than the myelin sheath that surrounds it, is at fault (Grandis & Shy, 2005). Both CMT and MD can only be acquired via inheritance, and both are incurable (Emery, 1994; Grandis & Shy, 2005).

CMT also differs from multiple sclerosis (MS). In both CMT type 1 and MS, the protective shield around the nerves, the myelin sheath, degenerates, in MS however, this takes place in the central nervous system (brain and spinal cord), whereas in CMT it occurs in the peripheral nerves. The only known cause of

CMT is inheritance, whereas the cause of MS is not clear. It is suspected that a virus or allergy has the rather bizarre effect of causing the body to attack its own myelin (Northern, 2000).

Charcot-Marie-Tooth neuropathy is not a well-known disease, in fact, most general practitioners are not very familiar with the neuropathy and large sections of the population have never heard of it (Beyer & Daino, 1990). However, the disease is not rare (in the applicable context); its prevalence is generally estimated at one in 2500, in other words, one CMT-affected individual for every 2500 of the population (Carter, England & Chance, 2004; Chance, 2001; Crabtree, 1997; Grandis & Shy, 2005; Tanaka & Hirokawa, 2002; Young & Suter, 2003).

According to a number of researchers (Arnold, McEntagart & Younger, 2005; Chetlin, Gutman, Tarnopolsky, Ullrich & Yeater, 2004; Crabtree, 1997; Kedlaya, 2007; Northern, 2000; Padua et al., 2006; Shapiro & Goldfarb, 1990), CMT is the **most common** inherited neurological disorder in the world. Crabtree (1997), for example, states explicitly that CMT is "… the most common inherited neuromuscular disorder known …" (p.293). By comparison, Emery (1994) estimates the prevalence of severely affected people with muscular dystrophy as one in 5000. However, according to a more recent source (Wenneberg, Gunnarsson & Ahlström, 2004b) the prevalence of the two diseases are very similar.

The fact that CMT is not a well-known disease creates problems for those affected by it, eventually hampering their coping efforts. Beyer and Daino (1990) highlight a few important dilemmas, most of which actually encapsulate various *motivations for undertaking this study*. They write: "Those with CMT have additional suffering. Very little is known about this hereditary disease and it often takes years to get a correct diagnosis. There is no cure… it generates little research. Even when they are properly and timely diagnosed, people often feel isolated, as most of them do not know anyone else with this disease, or for that matter, anyone who has ever heard of it. Even its name creates problems in that it implies a dental rather than a neuromuscular affliction. Almost nothing is known

about how CMT affects individuals' lives or how it affects families. Even less is known about how people deal with it" (p.143-144).

Even though the dental affliction remark, which refers to Howard H. Tooth, one of the founders of the disease, may very well contain an element of humour I, as a CMT- affected individual, can confirm that people have indeed asked me on more than one occasion if the disease affects ones teeth!

Humour aside, if one applies the ratio of 1 CMT case for every 2500 people, as reported above, to South Africa's population of 48,7 million (Statistics South Africa, 31st July, 2008), there could at present be approximately 19480 people in this country who have CMT. These people are confronted on a daily basis by the myriad of stressors engendered by living with the disease, creating pressure to continuously having to adapt.

Even though brochures/leaflets containing hints and guidelines on living and coping with CMT may be acquired from most CMT and muscular dystrophy organisations worldwide, there is a paucity of scientific research that delineates these processes on both theoretical and practical levels (Arnold et al., 2005). However, the assertion by Beyer and Daino (1990) that the disease generates little research (see above) has since changed dramatically regarding the physiological aspects, particularly on the genetics frontier (Crabtree, 2001; Grandis & Shy, 2005). Unfortunately, the dearth of research regarding the psychosocial aspects, particularly how people experience the disease, how they deal with it and adapt to it, has not improved much, if at all.

The availability of brochures and fact sheets mean little if affected people are not aware that they have this disease, which, in view of the limited public knowledge about CMT, is very likely (Crabtree, 2000). CMT is not limited to any particular ethnic group; it affects all ethnic and racial groups equally across the world (CMT United Kingdom, 2004a; Isitt, 2005; Kedlaya, 2007). In view of the composition of South Africa's population, [Africans comprise 79,2% of the total population, Whites 9,2%, Coloureds 9% and Indians/ Asians 2.6% (Statistics South Africa, 31

st July 2008)], a large proportion of the theoretically estimated 19480 people with CMT may belong to previously disadvantaged communities. Often, these communities struggle on many frontiers and they typically reflect environmental conditions in which disabled people face a particularly steep uphill battle in order to cope (Disabled People South Africa [DPSA], 2001; Swartz, 2004).

It has already been pointed out that the disease causes varying degrees of disruption in the lives of affected people, impairing their quality of life. As the above quotation from Beyer and Daino (1990) illustrates, people with CMT may even face additional obstacles to the myriad others that confront disabled people in general. The generic obstacles faced by the disabled, and with which the CMT-affected also grapple, are highlighted by Dr. Bill Albert of the British Council of Disabled People. According to him, one of the main problems that confront the disabled is stigmatisation, which amounts to disabled people being seen by society as lesser beings, including their functioning in various contexts. Furthermore, many disabled people internalise this and see themselves as unworthy. Dr. Albert points out that, worldwide, disabled people are being discriminated against on myriad levels and that they face numerous battles in order to obtain adequate health, social support and education (Rodgers, 2005).

Disabled People South Africa (2001, p.42) feels so strong about the effects of all this that they state: "The consequence is that disabled people are oppressed and discriminated against in all aspects of life, resulting in shorter life-spans, poverty and dependence…" Swartz (2004) also calls for more empathy towards disabled people; for what it means to loose ones place in society because one is different. He strongly feels that: "the biggest myth about disability is that it is something that an individual 'suffers from' and that it is unchanging" (p.23). Before elaborating further upon the ways in which CMT influences the lives of affected members, a brief look at the concept quality of life (QOL).

Quality of life

Despite the popularity of quality of life as an outcome measure in health research, there is little consensus about the definition of the concept, as well as the dimensions it embraces (Skevington, 1999). According to Huang, Wu and Frangakis (2006) perusal of the relevant research reveals that terms such as quality of life, health-related quality of life (HRQOL), health status and subjective well-being (SWB) are very often used interchangeably. Gill and Feinstein (1994), cited by Huang et al. (2006), analysed 75 articles that dealt with quality of life measuring instruments and found that a mere 15% of these articles attempted to define what quality of life encompasses. Neither did the researchers motivate their choices of particular measuring instruments. Not one of the articles distinguished between QOL, HRQOL and SWB.

For the purpose of this study, the World Health Organisation's (WHO) rather lengthy definition of quality of life, quoted by Skevington (1999), is regarded as the most suitable, because, amongst others, it is broad and all-inclusive. Besides, a measuring instrument called the WHOQOL-BREF, which, according to Taylor, Myers, Simpson, McPherson and Weatherall (2004), was based on the conceptual framework of the WHO, was applied in this study.

The World Health Organisation defines quality of life as: "…an individual's perceptions of their position in life, in the context of the culture and value systems in which they live, and in relation to their goals, expectations, standards and concerns. It is a broad ranging concept, affected in a complex way by the person's physical health, psychological state, level of independence, social relationships and their relationship to the salient features of their environment" Skevington (1999, p.451). Skevington (1999) continues by stating that a sixth domain, that addresses the religious aspect, was later added to the list contained in the definition. In the context of a person's health, Nätterlund (2001, p.8) states: "When physical status and functional abilities are the focus of research this is often referred to as health related quality of life". Functional status includes one's ability to perform the usual daily activities necessary to meet one's basic needs in

the various domains of life, as well as to fulfil the various functional roles in this regard (Abresh, Jensen & Carter, 2001).

Moving the magnifying glass to CMT and its relationship to quality of life, Redmond and Ouvrier (2001) administered the Medical Outcome Study Short Form-36 (SF-36), a generic instrument used to assess health-related quality of life, to about 350 CMT-affected individuals in order to establish how the disease affected them. This instrument assesses quality of life in eight domains, that is to say, degrees of limitation in each domain caused by the disease. The eight domains are *physical functioning, role limitations (physical), bodily pain, general health, vitality, social functioning, role limitations (emotional) and mental health.*

The results revealed that people with CMT scored lower than the general population on all eight domains, the lowest being on the physical dimensions. The SF-36 scores in each domain range from one to 100. The physical functioning score was approximately 25 points lower than that for the general population, with all other physical scores, including bodily pain, at least 10 points or more lower than the general population. Mental health was the least affected by CMT.

The eight subscales of the SF-36 can be combined to yield two global scores, namely Physical Function and Mental Function. Vinci and his research team (2005), in Shy and Rose (2005), found that both these global scores were significantly lower for their CMT subjects than for the Italian population as a whole. Another study that reports markedly diminished SF-36 scores (all scores, but especially the physical aspects) in CMT affected subjects compared to the general population was done by Padua et al. (2006). They included about 200 subjects from different centres in their study.

Various researchers, for example Bird (2006); Carter et al. (1998); Northern (2000); Padua et al. (2006) and Pfeiffer, Wicklein, Ratusinski, Schmitt and Kunze (2001) have reported that pain is a factor in CMT and that it impairs the quality of life of affected people. The latter research group compared CMT participants

with stroke patients. They hypothesized that emotional stress would be higher in the stroke patients because "they have less time to develop successful coping" (p.550), (compared to CMT with which one is born), but instead found the degree of stress to be similar in the two conditions. Pfeiffer et al. (2001) also exposed a variety of quality of life related variables that were affected by CMT, for example, manual dexterity was impaired, pastime activities were restricted and professional life was interfered with in more than 50% of the subjects. They established that the disease forced early retirement or fundamental retraining in as much as 36% of their subjects.

A rather unique problem that aggrieves CMT-affected people in the context of quality of life is the fact that many of us overtly appear to be coping, yet internally we are really struggling to keep up and are exerting much, much more energy than non-affected people exert in the process. Beyer and Daino (1990) put it as follows: "Many people with CMT, even those with braces and canes, look and act very normal. This normal appearance is one of the reasons physicians (and others) tend to dismiss their complaints" (p.145, parenthesis added). This research duo continue by pointing out that, in reality, the limitations imposed by CMT more often than not cause anxiety, embarrassment, anger, frustration and feelings of depression for the affected people.

As this introduction to the effects of CMT on quality of life indicates, the disease may disrupt and threaten the lives of affected people to various degrees. It is one of the fundamental goals of this study to closer investigate the ways in which the disease affects the lives of affected people. Shy and Rose (2005) makes the important statement that, even though it may very well be expected that quality of life will be impaired by a serious disease such as CMT, it nevertheless remains meaningful to establish **how** it is affected and to establish **which factors** contribute to it. This can be done in various ways. Arguably, there are two main routes to pursue, namely the application of *quantitative methodology* via standardised quality of life and other questionnaires on the one hand, and *qualitative research techniques*, such as interviews, observations and self-reports by categorisations of data, on the other.

Aside from the already discussed matter of confusion regarding the definition of quality of life, the more structured route of using existing QOL questionnaires have definite drawbacks. Henning (2004) describes a major one as follows: "We do not want to place this understanding within the boundaries of an instrument that is designed beforehand because this will limit the data to those very boundaries" (p.4).

Shy and Rose (2005) expose other, perhaps more indirect, problems. They, for example, point out that quality of life does not necessarily correlate with objective disease severity, largely because quality of life has a strong, if not decisive, subjective component. Hence, many studies rather unexpectedly find high questionnaire-assessed quality of life in chronically ill and disabled groups, a phenomenon that is referred to as the disability paradox. Quality of life is also a dynamic concept that is continuously changing during the course of the disease. Scores on tests may vary over time due to different influences, for example depression or other psychosocial factors that are not necessarily related to disability stemming from neuropathy.

Quantitative methods are generally not regarded as the most suitable methodology for exploring the subjective experiential worlds of people; qualitative techniques are, also in general, considered better suited for this purpose (Berg, 2004; Sarantakos, 1997). By asking people in an open-ended interview how they experience the effects of the disease, and how they cope with it, as will be done in this research, their subjective views may expose richer information than most, if not all, questionnaires can. Furthermore, in the interview situation matters such as the disability paradox may be investigated in more detail because one is not restricted to the questions in a questionnaire. Themes and other information may be followed up, potentially revealing even more information.

Even though it is conceded that qualitative methodology may not entirely solve all the above problems mentioned by Shy and Rose (2005), it is nevertheless considered the most suitable route for this study, which, fundamentally, will

explore the experiential world of people with CMT. Qualitative and quantitative research methodology will be discussed at length in Chapter 3. For now, it suffices to conclude this section with a quote. Anthropologist Robert Murphy, quoted by Beyer and Daino (1990, p.143) "has lamented that one of the questions a person with a disability or illness is never asked by physicians is, 'what is it like'". In this exploratory study, participants will be asked exactly this and similar questions in order to establish their subjective views on how CMT affects them and their quality of life.

The above bird's-eye view of the effects of CMT on affected people's quality of life links to the first of the dyad of this study's research questions, which was formulated as follows in the beginning of this chapter: *In what areas, and how, does CMT affect the lives of those who have the disease, according to their own frame of reference?* The second leg of the research problem, which will be introduced and contextualised in the remainder of this chapter, sprouts from the first and comprises the largest component of this study:

How do people who have CMT manage the manifestations, effects, implications and challenges that stem from this disease, in order to augment their well-being?

The latter question, namely how affected people *deal* with the disease's manifestations in order to optimise their adaptation, takes us into the realm of coping research, specifically coping with illnesses and disabilities.

COPING WITH CHRONIC ILLNESSES AND DISABILITIES: MORE NEED FOR RESEARCH

Coping with chronic illnesses and disabilities (CID) has generated much research in recent decades, especially during the latter half of the 1900's. For example, a literature search done by De Ridder and Schreurs (1996) discovered more than 700 empirical studies on this topic in the eight years between 1985 and 1993. The focus of research has been on the relationship between various coping strategies, or styles, that people adopt in response to their medical conditions, and various psychosocial outcomes, such as quality of life (Livneh, Lott & Antonak, 2004). Naturally, coping is not limited to CID, but instead applies to all problems,

situations and threats that necessitate coping responses. In fact, in the general context, coping research was so popular that Coyne and Gottlieb wrote in 1996: "… such studies (on coping) currently represent one of the most active research programmes in **all** of psychology" (p.960, parenthesis and accentuation added).

In the study of coping, at least two basic approaches, or distinctions, are prominent. One views coping as a dynamic process and holds that coping choices and behaviour, such as which coping strategy to adopt, depend on the nature of the situation that confronts an individual. Context is therefore an integral part of the coping process. The other approach allocates rather low prominence to situational or contextual factors. Instead, coping is seen as a stable enduring personality trait or style phenomenon (Lazarus & Folkman, 1984; Stone, Greenberg, Kennedy-Moore & Newman, 1991).

Arguably, the second mentioned approach is the oldest and dates back to work of Sigmund Freud, in the early 1930s (Endler & Parker, 1990). Hence, historically, coping processes were studied from the perspective of psychoanalytic ego psychology. The focus was on the ego's role in the regulation of anxiety via various mechanisms, which included both mature, adaptive ego processes, as well as various defence mechanisms, which were largely unconscious. To access these processes, a trained clinician, using tools such as projective techniques, was required (De Ridder, 1997; Folkman & Moskowitz, 2000). The ego psychology model contributed to coping being viewed as an enduring, stable style or trait phenomenon, rather than as a variable process that is continuously influenced by, and that varies in response to, the demands of each situation. In this (the former) approach, assessment of coping amounted to trait or style measurement (Lazarus & Folkman, 1984).

The style/trait approach to coping contains various nuances. One is that people's stable ways of coping stem from their personality characteristics, that is to say, "certain personality characteristics predispose people to cope in certain ways when they confront adversity" (Carver, Scheier & Weintraub, 1989, p.270). Another refers to people having a "preferred set of coping strategies that remains

relatively fixed across time and circumstances" (Carver et al., 1989, p.270).

Although the stable coping approach, embracing enduring traits, uniform styles and habitual ways of coping, has remained popular, there are serious deficiencies that need to be highlighted. The most prominent of these amounts to the fact that the focus is not on situational variability in the coping process. "Because trait measures characterise an individual's typical responses to stressors, situation-specific effects are lost" (Stone et al., 1991, p.648). Said differently, the situational context in which the coping takes place is not given the necessary prominence. The large role allocated to unconscious processes is also problematic; a need evolved for the creation of instruments that can assess conscious coping efforts (De Ridder, 1997; Lazarus & Folkman, 1984).

An example of the stable coping approach continuing is the work by Carver et al. (1989), who had incorporated its measurement in their COPE questionnaire. Actually, they developed the COPE in such a way that it has both dispositional and situational formats. The situational approach will be discussed next.

The situational approach, the other salient approach in coping research, is represented by the *transactional model* (Lazarus & Folkman, 1984), which was developed in response to the stable-coping approach. This model offers an alternative to the traditional models and sees coping as a dynamic process that changes between situations, indissolubly connected to stress. Lazarus and Folkman (1984, p.141) define coping as: "Constantly changing cognitive and behavioral efforts to manage specific external and/or internal demands that are appraised as taxing or exceeding the resources of the person". De Ridder (1997, p.418) states that, of the more than 30 definitions of coping that she had identified, the above definition has had the most important impact on the way in which coping is conceptualised.

In order to be able to assess behavioural coping in specific situations rather than enduring traits, the transactional view required the development of new measurement instruments; this subsequently gave rise to various coping

questionnaires. One of the most frequently used questionnaires is the Ways of Coping scale, developed by Lazarus and Folkman in 1980 and revised by them in 1985 (Stone et al., 1991).

In their clarification of the above-mentioned definition of coping, Lazarus and Folkman (1984) state: "… the problem of confounding coping with outcome is addressed by defining coping as efforts to manage, which permits coping to include anything that the person does or thinks, regardless of how well or badly it works" (p.142). They continue that, by using the word manage, they avoid the pitfall of equating coping with mastery. In addition to mastery of the environment, management also includes minimising, avoiding, tolerating and accepting the stressful conditions.

These views of coping and management largely reflect the spirit in which these concepts are conceptualised in the present project. Managing CMT, however, is seen as even broader still than this description, including both reactive (responding to disease-related stressors) and proactive (a future orientation, including making provision for the uncertain disease course) strategies. Management additionally includes an ex post facto element, that is to say, strategies to deal with the effects of disease-related traumas from the distant past, especially childhood. In this study, the terms "management" and "coping" will be used synonymously; as will the words "dealing with" and "handling". The reason for this is mainly that focusing on similarities and differences between these concepts will contribute little, if anything, to the answering of the research questions.

The approach of Lazarus and Folkman's (1984) transactional model, in the context of coping with medical conditions, holds that people with chronic illnesses and disabilities are confronted on a regular basis by the manifestations and effects of their diseases and conditions, which amount to an array of adaptive tasks, or stressors, that they face. In order to deal with, or regulate, the resultant stress, people engage in the coping process. Simplified, the coping process encompasses: 1) primary appraisal, which is a cognitive evaluation of the potential

impact or significance of the stressor (the threat) to the particular individual, 2) secondary appraisal, which amounts to an evaluation of the coping options available to deal with the stressor and, 3) the actual coping responses or strategies, which can be either behavioural and/or cognitive/emotional. There is also a step to evaluate the effectiveness of the coping (De Ridder & Schreurs, 1996).

There are two fundamental types of coping strategies, also referred to as the functions or foci of coping, namely *problem-focused coping* and *emotion-focused coping*. The former is directed at problem solving concerning the stressor as such, which does something about the situation that causes the stress. The second, emotion-focused coping, embraces cognitive processes directed at managing the emotional distress that results from the stressful encounter (Carver et al., 1989; Lazarus & Folkman, 1984).

Another prominent differentiation encountered in the literature is between *avoidance and approach* forms of coping, also referred to as passive and active forms of coping (Billings & Moos, 1981; De Ridder, 1997). Lazarus and Folkman (1984) provide the directive that, in general, emotion-focused coping is more likely in situations where the individual appraises that nothing can be done about the problem (stressor). Problem-focused coping will most likely be employed if the individual appraises that he or she can indeed do something about the problem that causes the stress. The former includes strategies such as avoidance, minimisation, distancing and other cognitive strategies to reduce emotional distress. Examples of problem-focused coping are defining the problem, generating, weighing and choosing between alternatives, and acting. Searching for information about one's disease and altering your home to enable streamlined living with a medical condition, are typical examples of problem-focused strategies (Groomes & Leahy, 2002; Lazarus & Folkman, 1984). Felton, Revenson and Hinrichsen (1984) argue that many illness-related stressors, specifically those that are out of the individual's control and management efforts, may best be dealt with via emotion coping strategies.

Livneh et al., (2004, p.413) pithily list trends and patterns in the literature

regarding different outcomes of coping with CID. With reference to coping with *chronic conditions*, under which CMT resorts (Shy & Rose, 2005), three salient findings pertaining to general trends are: 1) engagement type coping strategies, which include problem focused coping and relying on own internal resources such as optimism and internal locus of control, are generally related to lower levels of distress and higher well-being, 2) disengagement type coping strategies, for instance wishful thinking, using alcohol or blaming self/others, relate to higher levels of distress, and 3) avoidant, emotion focused type of coping tends to be associated with rather lower levels of quality of life.

Exceptions to these general trends are by no means rare. Avoidance, distancing and even cognitive distortion of the situation, for example, under certain circumstances may be adaptive rather than maladaptive (Felton et al. 1984). Exceptions to the general trends may be understandable especially in view of the fact that effective coping with any CID depends on a myriad of factors. A few are the nature of the disease, resources available to the individual and at what stage in the adjustment process (to the disease) the person is busy coping with. Regarding the latter, people with certain diseases are inclined to employ problem-focused strategies immediately after diagnosis, whereas at later stages the regulation of emotion becomes more prominent (De Ridder & Schreurs, 1996; De Ridder, Schreurs & Bensing, 1998).

Like the stable trait approach, the transactional approach has both *strengths and weaknesses*. A crucial advantage is that context comprises an integral part of the model. Said differently, individuals and their environments are inseparable and are in a continuous relationship with each other. This is not the case in other coping approaches, which are inclined to view people and their environment as separate entities.

Various researchers, including Coyne and Gottlieb (1996), De Ridder (1997), De Ridder et al. (1998), as well as Folkman and Moskowitz (2000), have highlighted weaknesses in the transactional model. In many cases, these criticisms are intertwined with criticisms regarding coping assessment instruments, most

noticeably the Ways of Coping Questionnaire (WCQ), which was developed for use in the transactional model. Many fundamental reasons for undertaking the present study are actually embedded in the following and formerly discussed criticisms against extant coping research.

Coyne and Gottlieb (1996) assert that the questionnaires themselves are not actually the essence of problems with the transactional approach; instead, the approach itself is at fault. They point out that Lazarus and Folkman (1984) state that they regard coping as being limited to: "conditions of psychological stress, which requires mobilization and excludes automatized behaviors and thoughts that do not require effort" (p.142). Coyne and Gottlieb (1996) maintain that the exclusion of *habitual* coping is a serious omission. They point out that many patients with chronic diseases such as multiple sclerosis (and probably CMT!) over many years have refined and routinized many coping strategies that work and have proven themselves. They continue that one criterion for effective coping with recurrent stressors actually amount to the existence and extent of these strategies!

Another problem is that the model does not allow for *anticipatory* coping. This type of coping takes place when people anticipate an approaching stressor and then act to avoid it. De Ridder (1997) directs our attention to the fact that the temporal aspect of coping efforts is also not addressed; the model does not specify whether coping applies only to immediate responses or whether it can persist over longer periods. She also refers to an assertion by Moos and Schaeffer (1993) that, because coping is restricted to being a reaction to stressors, many other determinants of coping, such as one's goals and commitments, are ignored. There is also no agreement on the number of dimensions that comprise adequate coping behaviour. This ranges from a few broad dimensions to the extreme number of 28 ways of coping in McCrae's (1984) instrument.

Folkman and Moskowitz (2000) add to the list of deficiencies by pointing out that coping has historically focused on the regulation of distress, and that the role of positive affect in stressful events has been neglected. Earlier research by

themselves on stress in caregivers indicated that, in 99, 5% of the 1794 interviews that they conducted, participants reported at least one substantial positive event in the stressful encounter.

Even though, as already mentioned, Lazarus and Folkman (1984) do not equate coping with effectiveness in terms of outcomes, the coping literature is peppered with direct and indirect referrals to the contribution (effectiveness) of various types of coping, and even coping per se, in improving affected people's well-being. To what extent then, is coping helpful in assisting people in their adjustment to their medical conditions? Felton et al. (1984) investigated exactly this question, aiming to establish the relationship between coping and psychological adjustment to various diseases, including rheumatoid arthritis and cancer. The four outcome measures for adjustment that they used were acceptance of illness, self-esteem, positive affect and negative affect. Overall, they found that coping had rather limited effect on people's adjustment to their diseases.

According to De Ridder and Schreurs (1996) there are other studies that found similar disappointing results; only a small proportion of the variation in patients' well-being (determined by assessing various outcomes such as depression) can be attributed to coping per se. Petticrew, Bell and Hunter (2002) analysed 37 studies to investigate the association between coping styles/types and survival, as well as recurrence, in cancer. They found little evidence that even well known coping styles such as problem focused coping, stoic acceptance and denial, have had any significant effect on the mentioned outcomes.

Possible explanations for the ostensibly limited contribution of the coping process is summarised by De Ridder and Schreurs (1996). One argument favouring coping with a CID, is the view that coping may be better conceptualised in terms of its *prevention* of more serious manifestations occurring, rather than as enhancing well-being as an outcome. The dangers of not acting upon stressors are another nuance of this argument (Mishel & Sorensen, 1991). In assessing the effectiveness of coping, the wrong choice of outcome measures may also give a

false picture. An example, taken by De Ridder and Schreurs (1996) from Folkman (1992), is the situation where a person is awaiting the result of a biopsy. Here, anxiety reduction may be a more appropriate outcome measure than, for example, experiencing depressed mood.

Clearly, despite well thought out reasons being furnished for findings that question the effectiveness of coping, more work is needed to arrive at a position where the contribution of coping to one's adaptation to a CID manifests clearer. In addition: "… a greater differentiation regarding the nature and phase of the disease, and its accompanying adaptive tasks (will) provide greater insight into the real effects of coping" (De Ridder & Schreurs, 1996, p.76, parenthesis added).

In order to address the shortcomings in extant coping research, Folkman and Moskowitz (2000) recommend, amongst others, the increased utilisation of qualitative research methodology. One can hardly express the advantages of this route better than they do. They write: "Although quantitative methods have the advantage of facilitating comparisons within and between individuals across stressful events and require little labour to score, they usually provide only a superficial description of actual coping processes. A great deal more can be learned about coping that helps support positive affect by asking people to provide narratives about stressful events, including what happened, the emotions they experienced, and what they thought and did as the situation unfolded" (p.652).

The above reasoning, together with the already introduced deficiencies in the extant quantitative coping literature, encapsulate the reasoning why a qualitative study was decided upon in order to investigate the phenomena of continuously adapting to, and engaging with, CMT, which includes coping with the neuropathy's manifestations.

As discussed, there is a paucity of research on the psychosocial aspects of managing the effects and manifestations of CMT, and all other congenital neuromuscular diseases. This study is therefore also regarded as a fruitful endeavour from a need perspective. This paucity of research may be a

considerable stressor for people seeking and subsequently dealing with a diagnosis of CMT. In the words of Julie Etchingham (2000, p.4), patron of the then CMT International UK: "Coping with any medical condition for life is a daunting prospect, but it is far more difficult if there is little information about it and even doctors themselves find it hard to diagnose. This is a problem faced by many people with Charcot-Marie-Tooth Disease. It may be the most common inherited neurological disorder, but learning to deal with it often means a long search for answers and help".

As indicated, there may potentially be more than 19000 people with CMT at present in South Africa. In view of the scarcity of public knowledge about the disease, it would not perhaps be unreasonable to expect that most are not aware of what disease they have. These individuals struggle and "suffer in silence and ignorance" to cope with the effects of CMT. Amongst those who have already been diagnosed, some may also struggle to obtain quality information about the disease.

It is the overall aim of this study to add value for these individuals, as well as for everybody with CMT. By providing knowledge of how affected people experience having the disease, and by delineating their management strategies and experiences, the quality of life of a number of CMT-affected people may improve, not only because enhanced knowledge may result in better coping, but also because they may practically learn from others with the same disease. As formulated earlier, these two facets, namely the disease related experiences and the management strategies employed, encompass the two research questions.

The qualitative methodology chosen as the most suitable vehicle in order to satisfy the research questions is the grounded theory approach (Glaser, 1965, 1978, 1992, 1998; Glaser & Strauss, 1967). This methodology allows for the formulation of a substantive grounded theory in the area(s) under investigation.

Following the present introduction chapter, Charcot-Marie-Tooth disease will be elucidated in Chapter 2, including certain basic underlying concepts. In Chapter

3, the magnifying glass focuses on the research design and method of this study, which comprise three interdependent sections: mainly theory, praxis and triangulation. In the first, various research paradigms, designs and methods will be looked at, with grounded theory as the main focus. The second focuses on the practical implementation of the methods, and the third elucidates measures taken to triangulate the qualitative findings. Next, in Chapter 4, the results of the research will be presented. The greater part of the chapter focuses on the grounded theory that has emerged, but a section on triangulation is also included. Lastly, a discussion and conclusion (Chapter 5) follows. Here, the findings are contextualised within the extant literature. There are three sections. In the first, the formulated stages are compared to a synthesised stage model of adaptation to CID. In the second, the stages and their contents are integrated with the extant literature on adaptation to CID, lastly followed by a conclusion.

CHAPTER 2

CHARCOT-MARIE-TOOTH DISEASE

INTRODUCTION

In the previous chapter Charcot-Marie-Tooth disease and its effects on affected people's quality of life were introduced. Basic issues related to coping and living with chronic illness and disability (CID) were also discussed, and the research questions were formulated. This chapter focuses on Charcot-Marie-Tooth disease and aims to delineate the basic nature of the disease and its impact on the lives of affected individuals. However, before we focus on CMT as such, two fundamental issues need to be elaborated in this chapter. First, CMT is a peripheral neuropathy: the peripheral nervous system is the primary location of the disease, even though deterioration in body parts served by the affected nerves, for example muscles, may be more directly observable. A brief introduction to the peripheral nervous system is therefore deemed necessary. Second, CMT is one of a family of peripheral neuropathies that includes both inherited and acquired conditions. A short discussion of these general peripheral neuropathies will be undertaken in order to show where and how CMT fits in.

We then proceed with the depiction of Charcot-Marie-Tooth disease. After an introduction and a short history, basic genetic concepts and causes of the disease are delineated. This is followed by a brief discussion of a number of salient CMT types. We then turn to the symptoms and general impact of the neuropathy and how the disease is diagnosed. Lastly, the matter of available treatments receives attention.

THE PERIPHERAL NERVOUS SYSTEM

The human nervous system includes various specialised structures and systems that form an integrated communication network throughout the body. CMT United Kingdom (2004b) metaphorically compares the human nervous system to a

network of electric wires. The central nervous system (CNS) comprises mainly the brain and spinal cord. The particular division that is the most relevant to this study however, is the peripheral nervous system (PNS). It comprises all the nerves that connect the CNS to the muscles and joints, as well to the peripheral sense organs, for example pain receptors in the skin, muscles and glands. The nerves that carry signals from the sense organs to the CNS are called sensory, or afferent, nerves. Motor, or efferent, nerves are the opposite; they take signals from the CNS to the muscles and organs. The peripheral nerves and its innumerable minor branchings, reach almost all parts of the body (Meyer, van Papendorp, Meij & Viljoen, 2002).

According to Meyer et al. (2002), nerve tissue as such consists of: (1) nerve cells or neurons, which transmit the signals and (2) glial cells or neuroglia which, amongst others, protect and support the neurons. The neuron has three parts: (1) a cell body (2) nerve fibres called dendrites, which receive signals and (3) an axon, which is a nerve fibre of varying length whose function is to transfer the nerve signal to its destination. Many axons, depending on the type, are surrounded by a white insulating layer called the myelin sheath, in principle similar to the insulating material around electric cables. These myelinated nerve fibres bundle together and constitute the peripheral nerves. The myelin itself is made up of lipoprotein material and, in the PNS, it is formed by glial cells called Schwann cells (Meyer et al., 2002). Once again, it is similar to an electric cable, where the wire in the centre represents the nerve fibre and the insulating material around the outside the myelin sheath. Damage to either the fibres or the myelin sheath will result in diminished or impaired conduction of impulses in that nerve (CMT United Kingdom, 2004b).

PERIPHERAL NEUROPATHY (PN)

Peripheral neuropathy (PN) is a broad category that envelops a complex of disorders in the peripheral nervous system that are essentially due to damage to either the protective coating around the nerves or to the nerve fibres itself. It is a very common disorder - so common in fact that it affects more people than the better-known disease of rheumatoid arthritis (Senneff, 1999). PN affects people

of all social and cultural backgrounds. The estimated prevalence is 3 to 4% of the general population. All ages are affected, but people in middle and late adulthood more so. In terms of etiology, or causes, a common distinction encountered in the literature is between acquired and inherited peripheral neuropathy. CMT is an example of the latter. About 50% of peripheral neuropathies are the result of complications that stem from diabetes mellitus. HIV infection is also associated with PN. About 10 to 25% of PN are idiopathic, meaning no definite cause can be found. Amazingly, it is said that there are more than a hundred causes of peripheral neuropathy (Senneff, 1999; Wolfe, 1999).

Conditions that may cause peripheral neuropathy include:

1. Direct trauma to the nerve (e.g. injuries).
2. Hereditary disorders (e.g. Charcot-Marie-Tooth disease).
3. Metabolic disorders (e.g. diabetes mellitus and vitamin B deficiencies).
4. Systemic effects of malignancies and their treatment (e.g. cancer, leukaemia).
5. Infectious or inflammatory conditions (e.g. AIDS, HIV without AIDS, Syphilis, rheumatoid arthritis, leprosy).
6. Exposure to toxic compounds (e.g. alcohol, solvents, glues and heavy metals)
7. Neuropathy related to the use/abuse of certain drugs.
8. Miscellaneous causes (e.g. exposure to cold temperature, ischemia)

(Abresh et al., 2001, pp 467-468)

In many cases, peripheral neuropathy appears to occur initially at the extremities of the longest nerves, that is, farthest from the spinal cord and brain. Symptoms vary according to the nature of the damage. Sensory neuropathies typically induce symptoms such as diminished sensitivity, numbness, pain and sensations of tingling and burning. Motor neuropathies typically result in weakness of the limb extremities, the feet, lower legs, hands and wrists. Lastly, autonomic neuropathies may, for example, lead to diarrhoea, light- headedness and sexual dysfunction (Senneff, 1999).

The discussion will now turn from general peripheral neuropathy to a specific subtype thereof, namely Charcot-Marie-Tooth disease, the subject of this study.

CHARCOT-MARIE-TOOTH DISEASE

Introduction

As was depicted in Chapter 1, Charcot-Marie-Tooth syndrome is a heterogeneous group of inherited diseases of the peripheral nerves, which is characterised by a chronic motor and sensory polyneuropathy. Muscles typically atrophy because they receive inadequate signals from the pathologic peripheral nerves that serve them. The disease affects both children and adults of all ethnic groups worldwide and may cause significant neuromuscular impairment (Chance, 2001; CMT United Kingdom, 2004a).

Great inroads have been made in recent decades with the identification of the mutant genes that are responsible for the disease. To some extent, these advances have forced existing classification systems to be adapted from time to time in order to incorporate these developments (Reilly, 1998). This has contributed to a lack of uniformity regarding classification of the many types and sub- types of the disease.

Not enough progress has unfortunately been made regarding standardisation of the many synonymous and often confusing names for this disease. Actually, since the very early beginnings, the name appears to have been an issue. Certain researchers originally named the disease after H. Eichhorst who did some research on progressive muscular atrophy in 1873. In the second decade of 1900, the disease was called Hoffman disease and later Charcot-Marie-Tooth-Hoffman disease (Kedlaya, 2007). Eventually it was decided to name the disease after Charcot, Marie and Tooth, the first who systematically described it in 1886.

The disease had since early times also been referred to as Peroneal muscular atrophy (PMA) because the peroneal muscle in the shin is amongst the first to be affected, and this name is still used in some circles. The most recent name,

Hereditary Motor and Sensory Neuropathy (HMSN), is a very accurate description of the nature of the disease. *Hereditary* refers to the cause of the disease, it affects the *motor* and *sensory* functions and it is a *neuropathy*, a disease of the peripheral nervous system. As if there are not enough names already, CMT United Kingdom (2004b) states that the disease is also known as hereditary hypertrophic neuropathy and in some circles as Dejerine-Sottas disease. The former term is encountered rather frequently in the literature: it refers to the actual underlying pathology that takes place in the nerves in the case of certain types of CMT, for example, CMT type 1.

According to organisations such as CMT United Kingdom (2004a) and South Africa's Muscular Dystrophy Foundation (MDF fact sheet # 2, 2002) HMSN is now the preferred term. No such clear pattern of preference between CMT and HMSN, the two main options, could however be detected in the literature. Many sources tend to use the two terms interchangeably, as synonyms. Reilly (1998, p.6) writes: "The classification of the hereditary motor and sensory neuropathies (HMSNs) is particularly difficult as the term 'HMSN' tends to be used in the clinical literature and the term 'Charcot-Marie-Tooth disease (CMT)' in the genetic literature. The underlying genetic defect has not been found for all of the HMSNs, so classifications using only CMT do not include all of the clinical syndromes".

Crabtree (2001) also holds that CMT is part of the broader category HMSN, of which more than 20 have so far been described. Carter, Abresh, Fowler, Johnson, Kilmer and McDonald (1995) state that there are at least 8 known forms of HMSN, and that only the first three (types I, II and III) represent the Charcot-Marie-Tooth syndrome. Carter et al. (2004, p.151) add type IV to the list and conclude that CMT accounts for as much as 90 percent of HMSNs.

After much reflection about the name, I have decided in principle to follow Linda Crabtree, founder of CMT International, who has CMT herself, who writes: "If we were to lean on the side of research these days, I'd say the other name for CMT, Hereditary Motor and Sensory Neuropathy (HMSN) would be more correct, but,

because so much research is going on, the entire classification of these diseases may change, so we'll stick with CMT for the time being" (Crabtree, 2001, para.7). In addition, all the participants in this study have been diagnosed with Charcot-Marie-Tooth disease and not other variants of HMSN. Before a few important types and subtypes of CMT receive attention, some interesting historical facts.

Early historical developments

Jean Martin Charcot (1825-1893), Pierre Marie (1853-1940) and Howard Henry Tooth (1856-1926) were the first to describe the full clinical syndrome of CMT in 1886. Fragmentary descriptions of the disease did exist before 1886, for example by Virchow in 1855 and Eichhorst in 1873. Eichhorst's work in particular was comprehensive and influential because he described affected persons in six generations (Dyck & Lambert, 1968a).

In 1886, Charcot was 61 years old and a professor of neurology at the Salpêtrière, a rather famous hospital on the bank of the river Seine in Paris, where he had achieved an international reputation. Interesting enough, the hospital had originally been the arsenal and gunpowder store of King Louis XIII. Pierre Marie was 33 years old and was Charcot's student and secretary. Charcot was a tenacious investigator and a prolific writer, credited with many achievements. His contributions were in fact so substantial that a postage stamp was issued in 1960 to commemorate him and his work (Chudley, 2000; Smith, 2001).

Charcot was a dramatic, entertaining teacher who attracted many students who later became famous in their own right, for example Joseph Babinski, Giles de la Tourette and Jules Dejerine. His most well known acolyte was Sigmund Freud, who studied hysteria and hypnoses with Charcot in 1885 for about five months. Charcot founded the well-known journal "Archives of Neurology" and is regarded as the father of modern clinical neurology. He was also the first to distinguish between neurosis and psychosis, as well as amongst the first to use hypnosis as a clinical tool. No wonder then that he is regarded as one of the founders of psychiatry. Charcot's name is also associated with amyotrophic lateral sclerosis,

as well as with a type of osteoartopathy called Charcot's joint (Chudley, 2000; Smith, 2001).

In 1886, in conjunction with his student, Pierre Marie, he described five children with progressive muscular atrophy and weakness, especially of the peroneal muscles, that were mainly familial in nature. They referred to this as peroneal muscular atrophy, a name that is still used today in some circles. Across the English channel, Howard Henry Tooth independently and using his own cases, described the same condition in his doctoral thesis at Cambridge University. These three neurologist's clinical description of uncomplicated CMT was so thorough that there has been no real need to modify it, though later developments, for example histopathological findings, naturally had to be incorporated. Interestingly, Charcot and Marie, despite the brilliant description, were not certain about the pathologic localization of the disorder, hypothesising a spinal cord lesion as the most probable localisation. Tooth, however, hypothesised correctly that the disorder is primarily caused by peripheral nerve dysfunction (Chudley, 2000; Kedlaya, 2007; Smith, 2001).

For many decades after Charcot, Marie, and Tooth described the disorder, confusion abounded as to what should be included in the disease and what not (Smith, 2001). In 1893, Dejerine and Sottas described a syndrome with very severe symptoms that were similar to those described by Eichhorst, Charcot, Marie and Tooth. Roussy and Lévi's syndrome, described in 1926, also had similar symptoms, but differed because affected people had a static tremor of the hands. Confusion gradually began to clear since more or less the mid- 1950s, with the groundbreaking work of Dyck and Lambert in the 1960s clearing up much ambiguity. They concluded that Dejerine-Sottas-disease was indeed a form of CMT, but a very severe variant. Roussy and Lévi's syndrome was also classified as CMT, but with superimposed tremor (Dyck & Lambert, 1968a).

Heredity and genetics are crucial concepts in CMT. The disease can only be acquired by way of inheritance, from either ones parents or, much more rarely, by way of new gene mutations. Many of the CMT types are inherited in different

ways. In view of their importance, a few basic genetic and inheritance concepts will be discussed in order to create a framework, or context, for the content to follow. The latter will include various distinctions between the different types and variants of the disease, including the clinical profiles.

Basic genetic concepts

Human life starts off as a single cell, the fertilized ovum, in which is contained all the genetic material, called the human genome, of a particular person. Through countless divisions (duplications) and differentiations, all our body cells develop, with each cell also containing the exact same genetic information. Each cell has a nucleus wherein almost all the genetic material is located. This encompasses 46 thread-like bodies, the chromosomes, of which 23 are inherited from the father and 23 from the mother. They exist in pairs, one from each parent, hence 23 pairs in total. Twenty- two of these pairs are called autosomes and are designated XX: they are unrelated to the biological sex of the individual. The 23rd pair is called the sex chromosome because they determine the person's genetic sex: females have two X chromosomes (XX), but males have one X and a smaller Y (XY) (Emery, 1994; Youngson, 1995).

The chromosomes carry the genes, which may be thought of as units of information that are hereditary. Genes have the crucial function to direct or code for the production of proteins, the basic building blocks of life, of which there are two types. Structural proteins help build the intracellular structures, for example cell membranes, while functional proteins perform functions that are important for normal cell functioning, for example the activation of chemical compounds (MDF fact sheet # 10, 2001). There are tens of thousands of genes in each nucleus. The exact same genes are present in all of our cells, but only certain ones are active in particular settings. The genes that code for the making of muscle protein, for example, are only active in muscle tissue and the ones for making myelin only in the nerve cells, and so forth (Emery, 1994).

Genes (and chromosomes) are composed of DNA (deoxyribonucleic acid). There are four building blocks that make up DNA, called DNA bases or nucleotides.

They are adenine, cytosine, guanine and thymine. These bases only pair with each other in certain ways and innumerable variations are possible. "A gene is therefore a specific order of DNA bases that carries a specific message" (MDF fact sheet # 10, 2001, p.7). DNA molecules are long and complex polymers, which, microscopically, resemble a double ladder that is twisted into a spiral - the so-called "double Helix". All this in turn is tightly coiled up to form the chromosome.

Chromosomes display distinct bands when stained with the applicable dye. The location of a particular gene on a chromosome is achieved by identifying the band in which it is situated. It is thus possible for geneticists to establish the exact location of a particular gene on a chromosome. Chromosomes resemble the letter X and the point where they are joined is called the centromere. The centromere is not in the centre, leaving us with a short top side of the X, the "p" side, and a longer bottom portion called the "q" side. A gene position designated as 22p10, for example, means that the gene is located on the short leg (p) of chromosome 22 in position number 10 (Emery, 1994; Youngson, 1995).

Etiology

Technically, Charcot-Marie-Tooth disease is caused by a genetic disorder, that is to say, there is a genetic defect present in the body. This genetic defect in turn results from inherited genetic material. The disease is mostly passed from parents to offspring, but new (de Novo) mutations in genes may also cause the disease (Northern, 2000). Charcot-Marie-Tooth neuropathy cannot be acquired in any other way than by inheritance, which includes spontaneous mutations (CMT United Kingdom, 2004 a&b).

Modes of CMT inheritance

Each of our genetic traits is determined by a pair of genes, one from the mother and one from the father. If the two genes are the same, the person is homozygous for that gene. When, for example both genes are for red hair, the person is homozygous for the red hair gene. Being heterogeneous means that the two genes are different; in the case of CMT the latter means that one gene is normal whilst

the other is the disease producing, mutant gene. This may result in one of two main scenarios. In the first scenario, the abnormal gene is "suppressed" by the normal one, in which case the mutant trait is said to be **recessive.** In other words, if both genes of a pair need to be abnormal to produce the disease, it is called *recessive inheritance.* Another scenario is where the one mutant gene is not dampened by the healthy one and is expressed: the mutant trait in this instance is **dominant.** Put differently, if only one gene of the pair needs to be abnormal to produce the disease, it is a case of *dominant inheritance* (Emery, 1994).

From the above information on dominant and recessive inheritance, as well as autosomal and X-linked inheritance, it is clear that various combinations of disease inheritance are possible. There are mainly three ways in which CMT is inherited: autosomal dominant, autosomal recessive and X-linked (MDF fact sheet # 2, 2002). Each will now be discussed. The diagrams below (Figures 2.1 – 2.3 b) are used with permission from CMT United Kingdom.

AUTOSOMAL DOMINANT INHERITANCE

This is the most common form of CMT inheritance. The affected person has one normal and one abnormal gene on the particular chromosome pair (heterogeneous). As can be seen in Figure 2.1, each child of an affected parent has a 50% chance of inheriting the disease. Both sexes are equally affected.

If a child is fortunate to inherit the healthy gene of the pair from an affected parent, it is the end of the

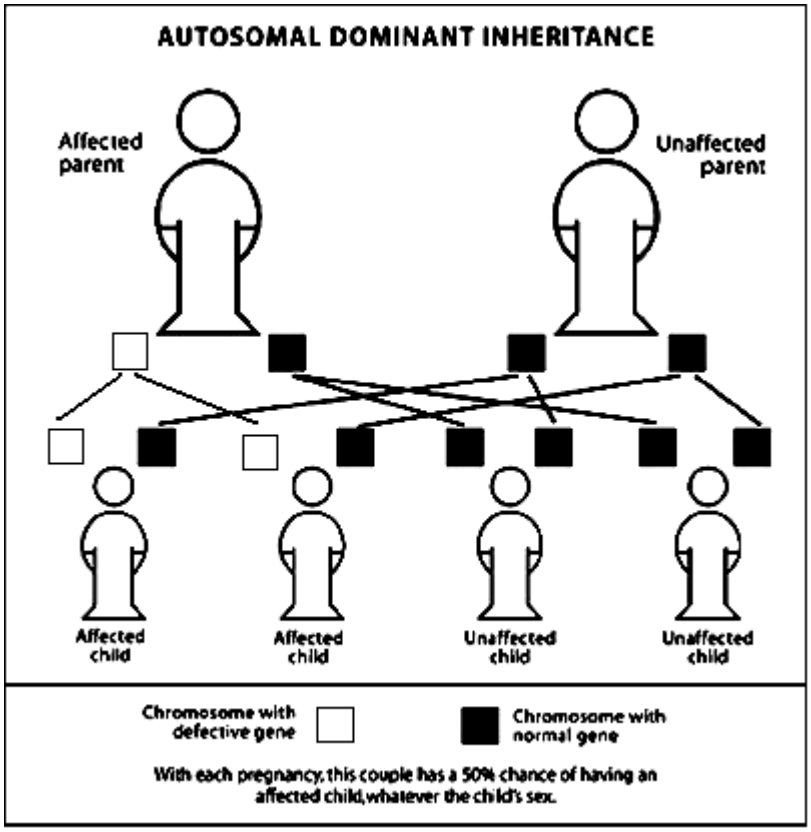

Figure 2.1: Autosomal Dominant Inheritance

line; that child cannot pass on the disease further. Unfortunately, it is possible for CMT to develop spontaneously by means of a new mutation, that is, when unaffected parents have a child. For further generations it is then the same scenario of, in this case, autosomal dominant inheritance (Emery, 1994; Northern, 2000).

AUTOSOMAL RECESSIVE INHERITANCE

Lately, the various forms of CMT that can be inherited in this fashion have mainly been grouped under CMT type 4 (Reilly, 1998). It is much rarer than autosomal dominant inheritance because the chances of two carriers ending up with each other to conceive children are small. How does this work? In autosomal recessive inheritance, both parents are carriers: they have the faulty gene but are more or less symptom free.

The faulty gene is suppressed by the other healthy one of the pair. They will probably not even be aware that they have the mutant gene until they mate with someone who by chance also carries the same mutant gene and they have an affected child. There is a 25% chance of the child being affected. Figure 2.2 depicts how this comes about. Since both parents must carry the rare deviant gene and be heterogeneous, the chance of being blood relatives exists, for example first or second degree cousins, who got the gene from a common ancestor (Emery, 1994, p.88).

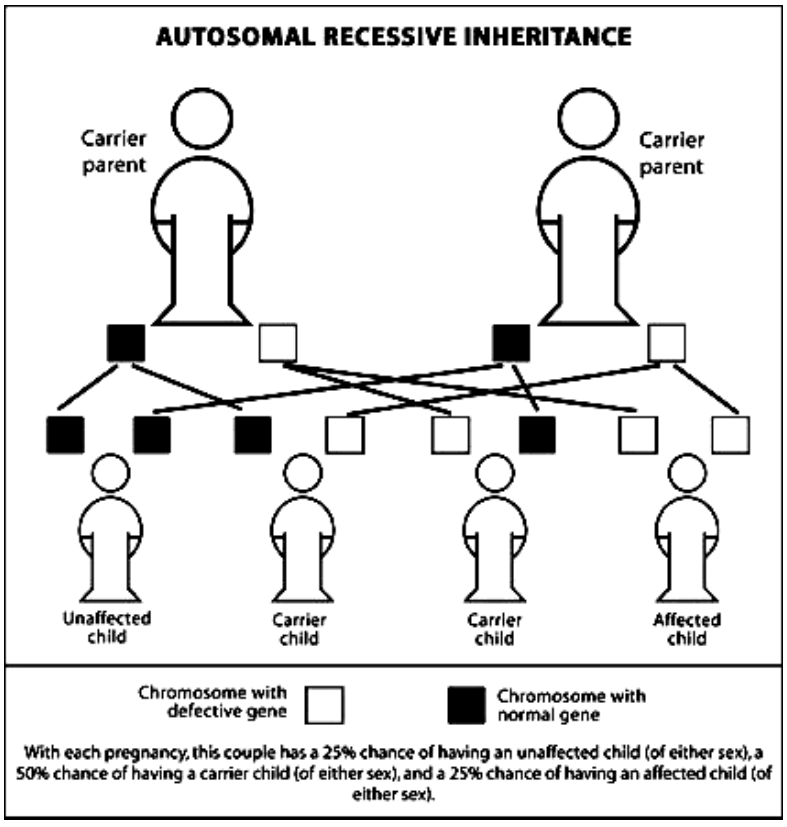

Figure 2.2: Autosomal Recessive Inheritance

X-LINKED INHERITANCE

Technically, X-linked CMT inheritance is also a form of recessive inheritance, but in this case, the defective gene is on the X chromosome of the pair of sex chromosomes. If the mother is a carrier, she is usually symptom-free or is very mildly affected. Recall that she has two X's and thus another normal gene (on the unaffected X chromosome) that suppresses the effects of the mutant one. As can be seen in Figure 2.3 (a), each of her sons has a 50% chance of inheriting CMT, and each daughter a 50% chance of being a carrier. In a scenario where the father is affected, the faulty gene is on his X- chromosome and, because he passes the Y-chromosome on to his male offspring, he cannot transmit the disease to his sons. All his daughters will however be carriers (Emery, 1994). This is depicted in Figure 2.3 (b).

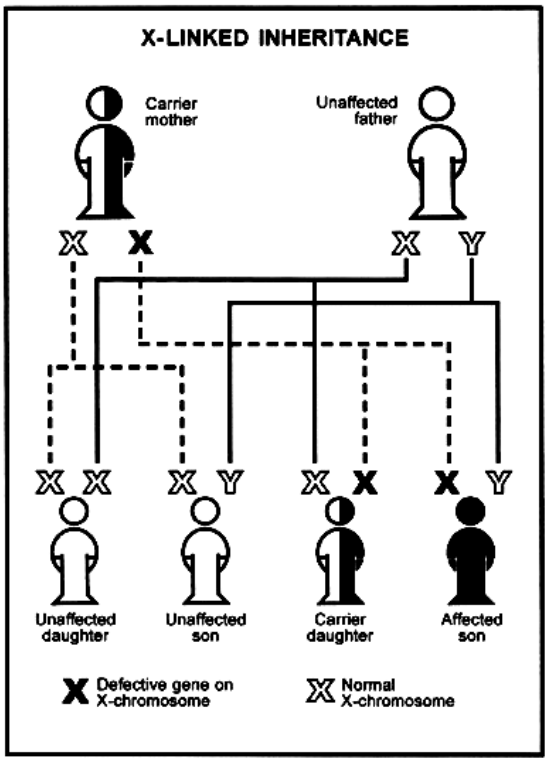

Figure 2.3 (a): X-linked inheritance

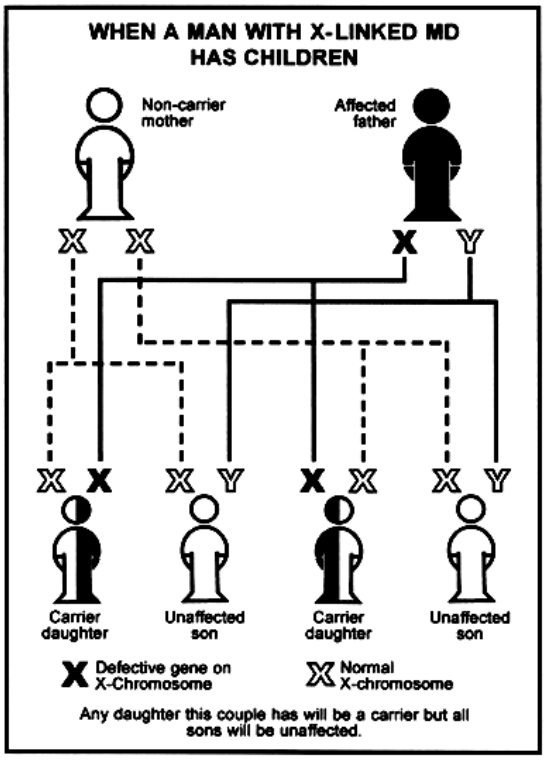

Figure 2.3 (b): X-linked inheritance

It is important to note that, with all types of CMT, it is possible for an individual to be symptom-free despite having the defective gene in his or her DNA. This happens in approximately 25% of cases (Chudley, 2000). The condition may then

appear to skip a generation in some families. The genetic risk, however, remains the same for subsequent generations (Northern, 2000).

Having elucidated basic genetic concepts and ways of inheritance, a depiction of a number of salient CMT types and sub-types, which includes classification on molecular genetic grounds, will follow.

Types of CMT

It is clear from discussions up to this point that there are many types and subtypes of CMT, and even more if all the HMSN's are brought into the equation. It is considered beyond the scope of this study to delineate all the known types and subtypes, many of which are extremely rare and have not been clearly sorted out and described, especially in terms of molecular genetics. Nevertheless, an introduction to a few salient types of CMT is deemed necessary. Amongst others, differentiation between the types is crucial from the perspective of genetic counselling, including the possibility of pre-natal strategies. (Arnold et al., 2005, p.308; Chance, 2001). Similarly, it will not be practical to attempt to elucidate all the applicable physiological intricacies, even of the well-known types. Hence, only a few of the most prevalent/well-known Charcot-Marie-Tooth types and subtypes will be introduced, with CMT type 1 being the most detailed. The important information as to how the various types are differentiated will be described next.

The classification of this group of disorders is continuously changing due to rapid progress in identifying the genetic defects that underlie the different hereditary neuropathies. This has been especially the case in recent times (Grandis & Shy, 2005; Reilly, 1998). Traditionally, the broad types of CMT were distinguished on the grounds of whether the individual has reduced *nerve conduction velocities* (NCVs) or not. The speed and strength of the nerve impulse as it travels down the nerve fibre is measured using electrophysiological techniques. This is still regarded as the most useful way of distinguishing between the different main types, but a combination of traditional and genetic techniques is increasingly

being used. The variants or subtypes of the main CMT types are mainly classified using molecular genetics (Reilly, 1998; Warby, 2001).

Based on electrophysiological data (see above), usually in conjunction with some combination of clinical, neurophysiologic, and histopathologic findings, CMT is subdivided into two fundamental types: 1) **demyelating** (most common example is CMT type 1, or just CMT 1), and 2) **axonal** (mainly CMT type 2 /CMT 2). As the name suggests, in CMT 1 it is primarily the myelin sheath that is affected. In CMT 2, the axon itself is the prime site of the neuropathy. In the case of CMT 1, the conduction speed of impulses in the nerves is considerably slower than normal, whereas in CMT 2 the speed is normal or near normal (Young & Suter, 2003), but the strength of the signal (amplitude) in the latter case is reduced (Chance, 2001; Northern, 2000). Traditionally, a NCV of 38 meters per second (usually, in the median motor nerve) is regarded as the "cut-off" between the two main types; below 38m/sec. is demyelating and above is axonal (Bird, 2006; Padua et al., 2006; Vinci, 2003). Dyck and Lambert (1968a) compared the conduction speeds in three important nerves of CMT1-affected people with those of normal subjects. The results are depicted in **Table 2.1**. Note: NCVs are expressed in meters per second.

Table 2.1: Nerve conduction velocities in CMT1-affected and unaffected subjects (Adapted from Dyck & Lambert, 1968a, p. 609)

	Mean age (years)	**Conduction velocity** (meters per second)		
		Ulnar nerve	*Median nerve*	*Peroneal nerve*
Affected	33, 8	23, 5	24	20, 3
Unaffected	27, 1	61, 6	56, 6	50, 4

Although the electrical test usually clearly distinguishes between types 1 and 2, exceptions do occur and these then are referred to as intermediate CMT (CMT United Kingdom, 2004b).

To reiterate, the subtypes of the main CMT types are differentiated based on the underlying genetic defects. Since the early 90s up to the year 2005, more than 30 genes that cause CMT have been identified, as well as at least 10 other loci (sites on chromosomes discovered, but not yet the actual gene itself) (Grandis & Shy, 2005).

A bird's eye view of the most important types now follows:

CMT 1

CMT 1 is characterised by slow conduction of impulses in the nerves, traditionally a NCV speed of less than 38 meters per second. Nerve biopsy, that is, removing a piece of nerve tissue and viewing it under the microscope, reveals segmental demyelation and remyelation, resulting in layers that are called onion bulb formation. For ease of explanation, this results because of an "overreaction" by the Schwann cells in response to demyelation, in effect an attempt to rectify the deviancy. This underlying pathology is called hypertrophic neuropathy (Chudley, 2000; Tanaka & Hirokawa, 2002; Young and Suter, 2003) and results in current leakage, causing impaired impulse conduction. Even though CMT 1 is primarily demyelating, there is, over time, almost always secondary axonal loss (Carter et al., 2004).

Onset of the CMT 1's is usually in the first or second decades of life. "Classical" symptoms such as slowly progressive distal (far away from the centre of the body) weakness and muscle wasting, sensory loss and foot deformities are hallmarks. The disease varies in severity. About 25% of carriers are asymptomatic (Chudley, 2000). There are at least six to seven variants of CMT1, distinguished on the grounds of the underlying genetic defect.

CMT 1A

This is the most prevalent variant of CMT disease (Dubourg et al., 2001). Between 70% and 80% of people with CMT 1 have type 1A, which is detectable by a blood test. Those affected display the classical demyelating phenotype

(observable characteristics) as described under CMT 1: hypertrophic neurology, onion bulb formation, very low nerve conduction speed and distal muscle atrophy. In the vast majority of cases, CMT 1A is inherited in an autosomal dominant fashion (Chance, 2001; Northern, 2000).

Type 1A is caused by an abnormal gene, PMP 22, on the short arm of chromosome 17 (position 17p11.2). The nature of the abnormality is a duplication; those affected have an extra copy of this gene resulting in a trisomy of PMP-22. The task of gene PMP-22 is to code for a protein, namely peripheral myelin protein, that plays a crucial role in the development and maintenance of compact myelin (via Schwann cells). The trisomy results in over expression, hence causing the pathology (Tanaka & Hirokawa, 2002; Young & Suter, 2003).

Interestingly, in a related condition, hereditary neuropathy with liability to pressure palsies (HNNP), a PMP22 deletion, rather than duplication, is responsible for the disease (Schröder, 2001).

CMT 1B

The defective gene in this sub-type is situated on the long leg of chromosome 1 and it is thought that it is responsible for the protein that makes the layers of myelin adhere to each other (Reilly, 1998). Approximately 5 to 10% of CMT1 cases are type 1B. Most cases are clinically identical to type 1A and they are inherited autosomal dominantly (Bird, 2006). Type 1B is also detectable by means of a blood test (Northern, 2000).

CMT 1C, D, E and F are further subtypes, which are very rare, and for the sake of brevity, will not be elaborated on.

CMT 1X /X-linked CMT

This is the second commonest form of CMT 1, accounting for 10 to 15% of type 1 cases. The faulty gene is located on the X chromosome of chromosome 23, the sex chromosome. Recall that the female's sex chromosome is designated XX and

the male is XY. The mutation is in a gene located at Xq 13.1, which codes for a membrane protein that is probably involved in the formation of gap junctions between layers of myelin (Carter et al., 2004).

In CMT 1X, males are significantly affected more severely than females, and females much more regularly display no symptoms at all (Dubourg et al., 2001). Females have another healthy X chromosome on chromosome 23 (they are XX) that may suppress the deviant one, whereas males have an XY configuration. The clinical manifestations for males are the same as in CMT 1 A (Dubourg et al., 2001; Northern, 2000; Young & Suter, 2003).

CMT 2

CMT 2 is primarily an axonal neuropathy, that is, the axon itself degenerates and not the myelin. Changes however also take place in the cell body (Carter et al., 2004). Nerve conduction velocities are normal or near normal, but action potential amplitudes are diminished. This is a less common type than CMT 1. The clinical picture is similar, but more variable. Dyck and Lambert (1968b) found that hand muscles were less severely affected than in the hypertrophic types, but that the ankle muscles were more severely affected. Onset of CMT 2 was also much later, even up to middle age or even later. CMT 2 is inherited in an autosomal dominant way, although recessive cases are occasionally encountered. Advances in molecular genetics led to the identification of an increasing number of subcategories (Warby, 2001), which, according to Bird (2006), range from CMT 2A to CMT 2L.

Dejerine-Sottas disease (DSD) /HMSN III/ CMT 3

This is a severe demyelating neuropathy with extremely low NCV's [for example, the five DSD participants in Dyk and Lambert's (1968a) study had NCV's of 3, 4, 5, 22 and 42 meter/sec. respectively in the ulnar nerve - please refer to **Table 2.1** for comparison], as well as an *early childhood onset*. In the past, it was thought to be inherited in an autosomal recessive way. Later research disclosed however, that DSD is usually inherited autosomal dominantly and that

the defect occurs in the same genes that cause CMT 1. Many cases, however, are the result of new mutations in the gene (Chance, 2001; CMT United Kingdom, 2004b).

CMT 4

An outstanding feature of CMT 4 is that it is inherited in an autosomal **recessive** fashion, often occurring in families with parental consanguinity. It can be either axonal or demyelating (Northern, 2000). According to Bird (2006), there are no less than seven variants of CMT 4. CMT 4A neuropathy has serious effects and even proximal (closer to the centre of the body) muscles can become weak. Vocal cord paresis is a frequent symptom.

Dyck's (1993) classification lists two further CMT types (he refers to them as HMSN types): CMT 5, which is characterized by additional spastic features and Type 6 (type Vizioli), where optic atrophy is present (Schröder, 2001). Both are rare variants. Interestingly, a participant in this study claims to have been diagnosed with type 6.

In conclusion, Reilly (1998) envisages that, in due course, when the underlying genetic defects of all the CMT's are known, the classification system will primarily be based on these defects. By way of example: Peripheral myelin protein 22 (PMP-22) - related disorders, human myelin protein zero (Po)-related disorders, and so forth. It may very well be reasoned by some that the current neurophysiologic - based classification system is becoming outdated, considering the advances in molecular genetics. According to Young and Suter (2003, p.2548), the replacement of this traditional system with one based on a genetic classification is however not likely, mainly due to the clinical and pathological variability of CMT, even when the same gene is involved, as well as the fact that mutations in very different genes often manifest in similar phenotypes with almost identical symptoms. The useful and logical distinction between demyelating and axonal forms in particular, may very well be with us for many years to come.

Symptoms, clinical features and impact

Charcot-Marie-Tooth disease has been described as a heterogeneous group of disorders, a polyneuropathy, which displays remarkable similarities in terms of symptoms and clinical picture, though certain differences are obviously encountered. Whereas it would be far-fetched to state that a definite generic CMT phenotype with 100% uniform clinical features exists, the symptoms overlap to such a large extent that the phenomenon of a typical CMT phenotype are regularly encountered in the literature (Bird, 2006; Crabtree, 2001; MDF fact sheet # 2, 2002; Northern, 2000).

As far as the two fundamental "watershed" types, namely type 1 (demyelating form) and type 2 (axonal form) are concerned, there is an extensive clinical overlap between the two types (CMT United Kingdom, 2004a; Dyck, Chance, Lobo & Carney, 1993, cited by Carter et al., 1995; Reilly, 1998). Because CMT 1A is by far the most common form, accounting for approximately 70 to 80% of CMT1 cases (Chance, 2001), symptom descriptions of a more general, generic nature tends to originate from this subtype.

Although symptoms of the different types of CMT overlap in general, there is a wide variation in individual clinical presentation of the disease. Even between affected members within the same family, considerable variation in severity, age of onset and clinical signs are encountered (Chance, 2001; Chudley, 2000). It is estimated that 10 to 20% of affected people display no symptoms at all, despite electric nerve conduction tests indicating the presence of the disease (MDF fact sheet # 2, 2002). In addition to genetic factors, variations in the clinical profile also stem from the reality that each person experiences and responds to CMT differently. Lifestyle, as well as intrapsychic, environmental and other contextual variables such as habits, diet, exercise, support systems and stress plays a definite role (Crabtree, 2000; 2001).

The onset of CMT is generally during the first or second decade of life; the first signs are usually noted by parents or similar caregivers between ages 5 and 15. Symptoms tend to become particularly noticeable during adolescence when there

is a sudden increase in growth. It should be reiterated that there is considerable variation. In some cases, most notably CMT 2, the onset is much later, frequently as late as middle age (Dyck & Lambert, 1968b; MDF fact sheet # 2, 2002; Northern, 2000).

The first symptoms to appear are typically related to the feet and legs, resulting in problems with walking and running. Parents may notice that the child has problems picking up his or her feet. The early pioneers, Charcot, Marie and Tooth, had already observed that the peroneal muscle, which runs down the front of the shin and that is responsible for lifting the foot (pulling the toe side upwards), is one of the very first muscles to atrophy (Smith, 2001). This then results in "drop – foot" and, because this causes the individual to trip easily, the knees are lifted high, resulting in the characteristic high stepping gait. There is also a loss of calf muscles, which adds to the gait and balance problems that affected people experience (Crabtree, 2000). Even though muscle weakness can first be detected in the peroneal muscles, weakness of the small foot muscles may have preceded it, but tests of the strength of these muscles are not reliable. Ankle dorsiflexor weakness clearly manifests in the person's inability to walk on his or her heels (Dyck & Lambert, 1968a). Vinci (2003) also mentions that the intrinsic foot muscles are affected very early in the process; the atrophying then spreads to other leg muscles.

Due to the atrophying in distal (far away from the centre of the body) muscles, which in turn results from the degenerative peripheral nerves, atrophying in the feet results in high arched feet, also called "pes cavus", and hammertoes, especially in CMT 1. This more often than not results in affected people struggling to get proper fitting shoes. Occasionally however, flat feet, the opposite, are encountered. The legs take on a characteristic shape known as "inverted champagne bottle legs" or "stork legs ": very thin lower legs because of muscle wastage and normal thigh muscle bulk. As can be expected, twisted ankles and sprains are very common, as is cramping (Northern, 2000; Tanaka & Hirokawa, 2002).

A very prominent feature of the disease is absent or diminished reflexes: the leg, for example, does not reflectively "kick" when hit with the appropriate instrument below the knee. The sequence of diminished tendon reflexes during the course of the disease was found by Dyck & Lambert (1968a) to be Achilles, quadriceps and upper extremity. The sequences in which selected CMT symptoms manifest at different ages are depicted in **Table 2. 2.**

Table 2.2: Neuropathy (hypertrophic) in Charcot-Marie-Tooth disease
(Adapted from Dyck & Lambert, 1968a, p. 608)

			Muscle weakness			Tendon reflexes			
Age (Years)	Number affected people	Pes Cavus (%)	Peroneal (%)	Gastrocnemius Soleus (%)	Intrinsic Hand (%)	Ankle (%)	Knee (%)	Arm (%)	Enlarged Nerves (%)
0 - 10	9	22	11	0	0	22	0	11	11
11 - 30	27	67	59	15	12	78	59	52	41
30 +	31	58	61	26	28	81	48	47	16

Restless legs syndrome, an irresistible urge to move the legs especially when sitting or lying down, is also a clinical feature in many cases. Interesting enough, Gemignani, Marbini, Di Giovanni, Salih and Terzano (1999) found restless legs syndrome to be more prevalent in CMT2 than CMT 1. To reiterate, it is rare for people with CMT to lose the ability to walk altogether, but aids such as walking sticks are frequently used as the disease progresses. However, severe cases may also make use of a wheelchair, though not necessarily full-time. (CMT United Kingdom, 2004b).

In terms of the upper extremities, weakness of the hands present much later, usually after the age of 20. Vinci (2003) is of the opinion that, although their hands are weaker, most affected people do not have such a severe disability in this area. Muscle wastage because of atrophy results in, what Northern (2000, p. 14) calls "a classic CMT hand". There are hollows where a muscle tissue should have been, for example between the tendons at the back of the hand and at the base of the thumb. Atrophying of muscles at the back of the hand allows the stronger

palm muscles to pull the fingers inwards, hence the hand takes on the characteristic claw shape. Finger, handgrip and wrist weakness are present in varying degrees. As can be expected, these changes typically result in difficulties with the execution of fine motoric tasks, for example buttoning, pulling zippers, writing and manipulation of small objects. No wonder then that affected people very often complain that they drop everything. In a variant of CMT 1, called Roussy- Levy syndrome, the individual's hands display tremor (Crabtree, 2000; Northern, 2000).

With nerve and muscle pathology present to the extent as described, it is to be expected that chronic fatigue in varying degrees will be present, and this is indeed one of the prime symptoms of CMT that more or less everyone experiences. Fatigue is exacerbated by a lack of pacing and moderation in physical and related lifestyle activities (Crabtree, 2000).

Sensory involvement in CMT had traditionally been thought of as playing a smaller role than motor involvement, a view also held by Charcot and Marie (1886), cited by Smith, (2001). Later research (for example Carter et al., 1998; Teunissen, Notermans, Franssen, van Engelen, Baas & Wokke, 2004) indicates that sensory functioning may be affected to a much larger extent than originally conceived. Teunissen et al. (2004) divide sensory symptoms in CMT into positive symptoms (tingling sensations), negative symptoms (numbness) and, lastly, pain. Typically, numbness due to sensory loss is present in both the hands and feet. In severe cases, this may result in the person unknowingly injuring him or herself, for example burns from hot cups, blisters because of poor fitting shoes and so forth. Cold hands and feet are common, and poor circulation plays a big role in this (CMT United Kingdom, 2004b). Experiencing "pins and needles" is very prevalent; more so in the lower limbs than in the upper limbs (Redmond & Ouvier, 2001).

The presence and extent of pain, a sensory modality, in CMT appears to be rather controversial. Bird, (2006) and CMT United Kingdom (2004b) are amongst a few sources that claim that pain is not a common feature in CMT. If present, it is

suspected that it may be due to secondary effects on the joints and/or muscles: there is abnormal impact and stress on joints because of weak muscles not doing their job properly, resulting in pain. Bird (2006) nevertheless recognizes that pain could indeed be a factor. Findings contradictory to these by, for example, Carter et al., (1998), Gemignani, Melli, Alfieri, Ingles and Marbini (2004); Northern, (2000) and Padua et al. (2006) however, indicate that pain is indeed a factor in the disease, in many cases even a substantial one. Carter et al. (1998), who studied the extent and nature of pain in CMT, found that 71% of CMT subjects (N= 617) reported the presence of pain. Thirty-nine percent said that pain interfered with their activities of daily living and more than 50% used some form of pain medication. Subjects who reported pain were asked to list the sites of the most severe pain. Unexpectedly to some, the lower back (70%) was top of the list, followed by the knees (53%), ankles (50%), toes (46%), feet (44%), neck (15%), shoulders (12%), hands (7%) and other parts (16 %). Carter et al. (1998) labelled the pain as neuropathic, meaning that the pain is due to injury/disease of the nervous system.

Northern (2000) also identifies the first two mentioned above, the lower back and knees, as major problem areas. He argues that this results from a distorted standing position when the body is bent forward at the hips and the knees are pushed backwards to maintain balance. This unnatural position puts additional strain on the lower back and knees. Gemignani et al. (2004) found, however, that nociceptive pain (pain that originates from somatic or visceral lesions) is more frequent in CMT than neuropathic pain. They conclude that sensory manifestations in CMT are much more frequent than previously thought, especially in CMT2. Further sensations that affected people may experience, are feelings of nerve or muscle "crawling" called formication, and burning sensations, mainly on the hand palms and soles of the feet (Crabtree, 2000). Scoliosis, or curvature of the spine, may be present in more severe cases (Chudley, 2000).

In addition to damage to the peripheral motor and sensory nerves, some loss of autonomic nervous function may also be encountered in CMT, although on a more limited scale. This means that automatically regulated functions, such as

breathing, heart rate, sweating, peristaltic movement of food, the working of stomach sphincters and sexual response might also be negatively affected (Shapiro & Goldfarb, 1990).

Although the central nervous system (brain and spinal cord) is not involved in CMT to a large extent, it nevertheless occurs - these forms are sometimes referred to as CMT plus (Vinci, 2003, p.763). Panas, Karadima, Kalfakis, Floroskufi and Vassilopoulos (2004) found definite CNS involvement in a father and daughter with CMT 1A, even though this is not commonly encountered in type 1. Specifically, they found problems in central motor conduction time, brainstem auditory evoked responses and visual evoked potentials. Evidence of demyelation was also found in the brain of the daughter, but not the father. Carter et al. (1995), on a more applied level, found that abnormal cognitive, intellectual and memory functions are not characteristic of CMT. Shapiro and Goldfarb (1990) found that higher education in CMT affected people has not been limited by the disease and conclude that intelligence is not affected. This is good news for us who are affected with the disease, because it means that our mental faculties remain intact.

In view of the extent of the motor, sensory, autonomic and, to a lesser extent central nervous system degeneration, it is no wonder that a myriad of diverse symptoms, not necessarily encountered, are reported in the literature. A few important ones, taken from Crabtree (2000; 2001) are: digestive and elimination problems, chewing and swallowing difficulties, hearing and eyesight problems, vocal cord paralysis and speech difficulties, poor balance, headache, sleep problems and breathing problems (because the nerve that serves the diaphragm is affected). Problems may also be encountered with proprioception, that is, the ability to tell where one's body and/or body parts are in space. Charcot and Marie (1886) cited by Smith (2001) mention a symptom that affects me personally, namely frequent muscle cramps, more particularly leg cramps. Another one of the outstanding symptoms that manifested in my childhood is toe walking, which is also described in a number of sources, for instance Crabtree (2001) and Vinci (2003).

A useful survey that aimed, amongst others, specifically to evaluate the impact of CMT on the quality of life of affected participants, was done in Australia in 2001. The number of completed and returned surveys amounted to 324. In this study, Redmond and Ouvrier (2001) found that more than 80% of participants were affected by muscle weakness, especially in the lower limbs. As can be expected, there was a strong association between muscle weakness and quality of life. Redmond and Ouvrier (2001) also found that more than 75% of participants reported leg cramps as a major problem. There was a strong relationship between prevalence of cramps and weakness; severity of cramps was also a significant factor in models predicting quality of life. About the same number of participants reported difficulty with balance, be it static standing or walking, especially on uneven surfaces.

Rather unexpectedly, about three quarters of the people in the Australian survey in 2001 also reported restless legs. Scoliosis, or curvature of the spine, although reported more common than in the general population, was nevertheless rare. Forty-seven percent reported tremors, more than in previous studies where the severity of tremors was assessed by doctors. On the sensory side, shooting pains were a problem for 75% of respondents, as were sensitivity to cold in both the upper and lower limbs. Hearing and vision problems were more than in the general population, but unfortunately, causes other than CMT were not ruled out. Alarming from the perspective of injury is that 25% of respondents reported frequent falls to the ground.

In another, significant study on the impact of Charcot-Marie-Tooth disease, Carter et al. (1995) collected data prospectively from 86 CMT types 1 and 2 participants over a 10-year period in order to compile an impairment and disability profile for the disease. They found a slow, progressive decline in muscle strength over the assessment period, with ankle muscles deteriorating the most. On average CMT-affected people produced 20 to 40% less force than normal controls using quantitative isometric and isokinetic measures. Pulmonary problems were not present to a large extent: only one person had severe restrictive lung disease, and 12 (14%) had a history of significant respiratory complications. Thirty percent of

participants had abnormal electrocardiograms, though only 7% had a history of cardiovascular symptoms, which is rather low. More than half the subjects (55%) had problems walking or climbing stairs unassisted.

As can be clearly seen in the foregoing exposition, Charcot-Marie-Tooth neuropathy is capable of profoundly affecting the lives of people with the disease. The physiological effects in particular, being largely not controllable by the individual, have the potential to disrupt affected people's lives and diminish their quality of life. However, to reiterate, the way people deal with the physiological realities plays a role in the eventual outcome of these effects. The way people deal with these physiological realities is the main focus of this research and will receive attention in later chapters when the research findings are discussed.

Diagnosis

Since the different CMT types and subtypes are not inherited in the same way, effective genetic counselling is dependent upon an accurate diagnosis of the type of CMT. It is often very difficult to distinguish clinically between the different types; therefore, confirmation of the clinical diagnosis by means of genetic diagnostic techniques is optimal (Honiball, 2003). However, diagnostic genetic testing, the best way to accomplish this, is not yet readily available for all the types and variants of CMT, if at all. Northern (2000) states that these tests are only readily available for types 1A, 1B and X linked. More types could however be tested for on a limited scale at selected laboratories, especially for research purposes (Bird, 2006). Testing is usually performed by drawing and analysing a blood sample. In South Africa, testing is only readily available for PMP-22 (mainly type 1 A) (Dr Janine Heckman, Groote Schuur Hospital, personal e-mail communication, 2006/04/03).

Other than genetic analysis, a diagnosis is based on a thorough neurological examination and a detailed analysis of the family history. The former includes tests of muscle function by means of an electromyogram (EMG), electrical nerve conduction studies (NCV) and tests of sensory response. In some instances a nerve and/or muscle biopsy, where the tissue is analysed for underlying

pathology, may be done to aid or confirm the diagnosis (Northern, 2000). It should be noted that electrical studies remain the most important diagnostic tool in most cases, especially during the initial diagnostic phases. Children affected with CMT 1A show the typical electrical abnormality from approximately age five (CMT United Kingdom, 2004b).

By way of example of what the diagnostic process practically entails, I have been subjected to the diagnostic process at least three times by different neurologists. Each session was extremely thorough and indeed a long process. Important steps that I remember are a vibrating tuning fork being held to my limbs to establish if I detect the vibration, pricks with a pin on my feet and hands and moving upwards to establish at what point I notice it, my totally absent knee and other reflexes, my muscle and hand grip strength being assessed via resistance, observing my walking, establishing if I can walk on my heels and my rather comical, uncontrolled leg jerking because of the electrical stimulation during the NCV-test. Beforehand, my limbs were submerged in lukewarm water. I do not know if it was supposed to be so, but the nerve conduction tests were rather painful, and even more so during the last test at a State Hospital, where, in my opinion, the technician too abruptly and insensitively turned the strength of the machine to maximum strength.

Getting back to the genetic aspect, problems may occur in the case of predictive testing, especially in the case of the rarer forms of CMT. This happens when symptom-free members of an affected family want to undergo genetic testing in order to establish their chances of developing the disease at a later stage in their lives. Information is not always available whether the presence of the abnormal gene will necessarily result in the disease (CMT United Kingdom, 2004b).

Certain types of CMT, notably type 1A, may be detected prenatally after plus minus 10 weeks of pregnancy. This is done by means of a technique called chorionic villus sampling. The chorionic villi are the early formations of the placenta and have exactly the same chromosomes as the embryo. A small sample is taken either through the neck of the womb or through the abdominal wall. The

procedure carries a one in 50, or even higher, risk of causing a miscarriage. How serious the baby will be affected in later life can unfortunately not be detected by the test (Northern, 2000; Youngson, 1995). This technique, as well as the better-known amniocentesis test, almost inevitably adds additional stressors on parents. Honiball (2003, p.27) aptly asks: "What should they do with a positive result? Abortion?" Their dilemma can clearly be envisaged, more so in view of the uncertain course of the disease.

Another preventative orientated technique that is being developed, and that may even be seen as a better option by some, is preimplantation genetic diagnosis (PGD). This encompasses in vitro fertilization outside the body and subsequent testing for specific genetic dysfunctions before implantation into the woman's body. The embryo develops in the laboratory until the 8-10 cell stadium is reached. A cell from it is then removed and tested for the genetic defect. Only embryos free from the dysfunction are implanted into the womb.

This technique was developed to assist those who have a genetic dysfunction but do not want to pass it on to their offspring. It is already being used to detect more than 30 dysfunctions in embryos, for example Down syndrome, spina bifida, cystic fibrosis, Huntington's disease and certain genetic cancers (Honiball, 2003). Crabtree (2001) specifically mentions that passing CMT to a child may be prevented because of PGD.

In South Africa, this technique is still in its infancy. For sure, there are ethical questions, typically, for example, who decides which dysfunctions are acceptable and which are not? The grave danger of the technique being misused to predetermine the appearance, intelligence, sex and so forth of a child always looms in the background. With well-informed, professional people handling the matter, and the risks controlled for by means of strict policies and procedures, perhaps the potential advantages of this technique outweigh the negatives? Perhaps there will always be disagreement, and similar to so many thorny issues concerning children, the final decision will eventually have to be left in the hands

of (confused or enlightened?) parents. Now, a brief look at available medical treatment and management by professional service providers in the field.

Treatment

Given the characteristic course of this disease, namely gradual, progressive muscle weakening over time that may result in disability of varying degrees, the question may rightfully be asked if this downward spiral necessarily implies a hopeless scenario for people with CMT. Can the disease be treated? Even cured?

The answer to these questions embrace a comprehensive field, ranging from almost apparent corrective steps, such as wearing sturdy boots or using a walking stick, to complicated surgical procedures and even cutting edge technologies, such as the possible use of stem cells. It is considered beyond the scope of this project to attempt an in-depth discussion of this very comprehensive field. A number of treatment options, such as surgery, helping aids and pharmacological interventions, will however be delineated in Chapter 5 as part of the discussion of how the people who are themselves affected by CMT live with and manage the disease on an individual basis.

What needs to be pointed out at this stage is that, despite formidable progress since the early 90s on the genetic frontier concerning etiology, no cure for CMT disease has yet been found. This is not to say that the scenario is hopeless for people with CMT. Various symptomatic treatments exist that can bring relief and improve affected people's quality of life (Grandis & Shy, 2005, p.23). These symptomatic treatments for CMT in many cases encompass management by a multi-disciplinary team consisting of, for example, a neurologist, an orthopaedic surgeon, a physical therapist and an occupational therapist (Bird, 2006; Carter, 1997). Naturally, factors such as degree of seriousness, nature of symptoms, individual choice and so forth play a role in determining the composition of the team, if any.

Vinci (2003, p.762) posits that, because a drug or gene therapy to cure CMT is still unavailable, the only treatment to improve the patient's functional abilities

and quality of life is rehabilitation. The goals of rehabilitation in neuromuscular disease are: "to maximize functional capacities, prolong and maintain independent function and locomotion, inhibit or prevent physical deformity and to provide access to full integration into society with good quality of life" (Carter, 1997, pp. 69-70).

This chapter provided background information about CMT neuropathy and related underpinnings. The latter included basic genetic concepts and general acquired (not congenital) peripheral neuropathy. The physiological aspects of CMT disease are complex and were dealt with only cursory. However, these aspects do not reflect the main goal of this research, which amounts to how people subjectively experience the effects of the disease, and how they deal with it in order to augment their well-being. In order to investigate these two fundamental research problems, the route that will be followed and the vehicle(s) that will be used need to be looked at. The discussion now proceeds to the methodology of this study.

CHAPTER 3

METHOD

The overarching aim of this chapter is to elucidate the designated research method, including the implementation thereof. The rationale for selecting it amongst many alternatives will also be addressed. For explanatory purposes, the contents of the chapter will be sorted into three overlapping sections or parts.

Part 1, called "mainly theory" (Ekins, 1997, p.3) focuses largely on theoretical issues pertaining to the area of research methodology, as is applicable to this study. The section includes visiting important groundwork issues, such as the various research paradigms and philosophical perspectives in which all research is rooted (Neuman, 2000; Ponterotto, 2005), as well as the salient research methods and approaches of each. The main feature of Part 1 is a delineation of grounded theory, which is the chosen methodology. Part 1 may be seen as being a theoretical context for the practical component in Part 2.

Part 2, is called "praxis" (Fassinger, 2005, p.157), and encompasses the study's operational component. It explains how the research was conducted, in other words, how the selected method (grounded theory) was applied. Amongst others, it deals with the research procedure, the sampling, and the specific methods employed.

Part 3 focuses on the triangulation of the qualitative data, and comprises a description of various psychometric and other questionnaires.

PART ONE: MAINLY THEORY

INTRODUCTION

The discussion in this section focuses on theoretical issues, including the underlying philosophical framework and specific methodologies utilised in the

present research, as well as the reasons for selecting them. A logical way to proceed may be to begin by revisiting the research aims and questions, since these represent both our point of departure and the destination we are heading for. The content of this chapter may be conceptualised as an investigation of the different routes to get to the destination, eventually concentrating on depicting the best way.

The two **research questions** of this study, which were also stated in Chapter 1, are as follows:
(a) In what areas, and how, does CMT affect the lives of those who have the disease, according to their own frame of reference?
(b) How do they manage the manifestations, effects, implications and challenges that stem from having this disease, in order to augment their well-being?

Embedded in the abovementioned research **aims** (that is, to satisfy the stipulated research questions), but not an explicit research aim per se, is the motive to obtain a deep understanding of what it means to be living with CMT. By addressing these aspects, the knowledge base about CMT may be broadened, which is considered a meaningful endeavour in view of the fact that the disease is rather unknown (Crabtree, 2000), as well as the high estimated prevalence worldwide, including South Africa.

An almost natural progression following the statement of the research aims/questions is the question of how to proceed in order to optimally investigate them. This amounts to deciding on the most suitable research design and method (or combination of methods), an undertaking that necessitates closer investigation of research methodology that is considered relevant to the present research. The discussion in this regard will broadly proceed from general issues, such as the various theoretical frameworks or research paradigms, to the specific strategy that was implemented. A few fundamental requirements and matters need to receive attention before we begin this discussion.

In order to establish what method(s) will be able to optimally address the research aims, various factors had to be taken into consideration:

(a) As already discussed in Chapter 1, there is a paucity of research regarding the psychosocial aspects of living with CMT. This includes the illness experiences of CMT affected people, as well as how they manage the effects of the neuropathy and their lives in general in order to cope with it. No standardised disease specific measuring instrument that assesses quality of life in people with CMT could be traced. In fact, Shy and Rose (2005) call for the development of such a CMT-specific quality of life instrument, amongst others to assess the outcomes of new treatment possibilities. The only CMT- specific instrument, the CMT Neuropathy Score (CMTNS), which had been developed recently, only assesses the levels of physical disability in CMT.

Generic quality of life instruments have only been used on a limited scale in CMT studies, and when used, have focused almost exclusively on the health-related quality of life phenomena. Pfeiffer et al. (2001), for example, used the Sickness Impact Profile (SIP) to assess the health-related quality of life of subjects with CMT, while Padua et al. (2006) employed the Medical Outcome Study Short Form 36 (SF-36) for the same purpose.

Overall, psychometric instrument based quantitative studies have to date generated limited knowledge on what living with CMT encompasses. The paucity of research on CMT is hardly better regarding qualitative research, where only a limited number of exploring and descriptive studies, focusing more on genetic and family issues, could be traced. Ideally then, a suitable method will have to allow for the exploration of even the most fundamental aspects of living with CMT. This entails engaging the people themselves by asking them how they subjectively experience the effects of the disease and how they are dealing with it.

(b) I, the researcher, am myself affected by CMT and will be a participant in this study. Even though various researchers, such as Tenni, Smythe and Boucher (2003), highlight the reality that, when analysing one's own case, the researcher's perspective should be incorporated in the data, the application of a rigorous method, as well as the use of clear prescribed guidelines, steps and procedures, are advisable to minimise researcher bias as far as possible. Bias is here understood as

what Henning (2004) describes as biasing or distorting the findings to mean what the researcher wants them to mean (even though not necessarily consciously and deliberately).

(c) The chosen method will have to provide suitable procedures and techniques that enable the thorough and orderly analysis, as well as the description, of concepts.

(d) In view of the limited public knowledge and inadequate infrastructure regarding CMT (see Chapter 1), difficulties in locating research participants are anticipated; the suitable method therefore should be able to accommodate working with a rather small group (small N).

(e) Finally, the elected method should ideally allow for the formulation of a theory or, more directionally stated, a process orientated theoretical model that addresses the above-mentioned requirements. Put differently, this study aims for the research output of a theory on how adults with CMT manage the symptoms, effects and manifestations of the disease themselves in order to optimise their adaptation to it and, in so doing, their well-being. This includes the management of current *and* anticipated effects of the disease and refers to both the less pleasant, negative side of the disease, as well as the more positive aspect in the sense of still attempting to live life to the full.

From the above, it was apparent that quantitative research methods on their own were not a viable option. There were far too few participants to meet the necessary quantitative requirements of sample size and representativeness (Berg, 2004), as well as a lack of disease specific measuring instruments. Furthermore, as was pointed out in Chapter 1, quantitative methods are also not the most suitable method to explore the subjective world of participants.

These factors led me to qualitative research methods, of which there are a myriad in existence. Since the overriding purpose of this chapter is the elucidation of the chosen research method, as well as how the research was carried out (praxis), it

follows that the accent will be on grounded theory methods. An in-depth discussion of all the qualitative methods that was decided against will therefore not be undertaken. Instead, a selection of the salient ones, including various underlying issues pertaining to qualitative research, will be introduced. Interwoven in this discussion, will be the reasons why particular choices were made, culminating in the motivation in favour of grounded theory, as well as the specific version of grounded theory. However, for the discussion to meaningfully proceed to the elucidation of qualitative research, the foundation in which all research is anchored, namely the different research paradigms, or philosophical perspectives, needs to be clarified.

RESEARCH PARADIGMS

All research, including qualitative research, is rooted in broad, underlying theoretical perspectives or scientific paradigms. According to Patton (1990, p.37): "A paradigm is a worldview, a general perspective, a way of breaking down the complexity of the real world", and "paradigms are really about epistemology and philosophy of science" (p.39). Guba and Lincoln (1994) define a paradigm as: "the basic belief system or worldview that guides the investigator, not only in choices of method but in ontologically and epistemologically fundamental ways" (p.105).

These broad approaches, or sets of basic beliefs, are in effect frameworks within which researchers conduct research. They set the context for studies (Ponterotto, 2005). The basic beliefs and assumptions that are inherent to them guide the research process along (Neuman, 2000). Paradigms are largely based on (and typically differ regarding) their ontological, epistemological and methodological assumptions. *Ontology* concerns itself with the question what the nature and form of reality comprises, as well as what can be known about it. *Epistemology* focuses on the study and acquisition of knowledge, including the relationship between the knower (research participant) and the would-be knower (researcher). *Methodology* amounts to the procedures and processes of research, including the specific ways and methods of inquiry (Guba & Lincoln, 1994; Myers, 1997; Ponterotto, 2005). Ponterotto (2005, p. 132) mentions two other fundamental assumptions on which the paradigms differ. They are *axiology* (the role of researcher values in the

research process) and *rhetorical structure* (the type of language used in presenting the results and procedure of the research).

Guba and Lincoln (1994) and Neuman (2000) maintain that three broad approaches have crystallised over the years, namely positivist, interpretivist and critical. Although this threefold classification system is popular, different naming, finer distinctions and elaborations are common. Denzin and Lincoln (1994), for example, substitute interpretivism with constructivism. Furthermore, there is a lack of uniformity as to how and where to position phenomena; participant observation, for example, is regarded by some as a core research approach, whilst others define it as merely a data collection technique (Caelli , Ray & Mill, 2003).

Ponterotto (2005) indicates that above-mentioned threefold classification is not rigid and that various combinations are possible. As far as practically selecting specific methods for one's research project is concerned, Patton (1990) states that paradigms and their link to method choices are by no means absolute and that "all kinds of variations, combinations, and adaptations are available for creative and practical situational responsiveness" (p. 39).

Positivist research

Positivist research is the oldest branch and is essentially the approach of the natural sciences. This is the world of quantitative techniques. Ontologically speaking, positivists believe that there is an objective reality out there to be discovered. This reality can be accessed, measured and described by an outside researcher. In other words, epistemologically speaking, the researcher and the subject are separate entities, and the former must ensure objectivity by not allowing the parties to influence each other. Where influencing is suspected, it is seen as a threat to validity and various strategies are followed to minimise it. Hypothesis testing, the formulation of causal laws, controlling of variables and statistical methods are hallmarks of this approach.

Post positivists fundamentally share the same positivist beliefs, but concede that reality can never be fully described; describing as much as possible becomes the

aim (Denzin & Lincoln, 1994; Neuman, 2000). The ontological position of positivists is referred to as *naive realism* and that of post positivists as *critical realism* (Ponterotto, 2005). Positivism is the fundamental paradigm that underlies quantitative research.

According to Black (1994, p. 425): "...health services research is dominated by quantitative methods". In the specific area of chronic illness and disability, research (CID), where CMT will be located (Shy & Rose, 2005), the positivist approach is very popular. An example is the use of a vast array of standardised coping and quality of life scales in order to investigate coping with disease/disability, as well as the impact on well-being.

It needs to be mentioned, however, that positivist research is not at all without criticism. Especially in recent times, many followers of other schools of thought such as constructivism, critical theory and postmodernism typically allege that: "…. (Positivist) criteria reproduce only a certain kind of science, a science that silences too many voices", and that these methods "are but one way of telling stories about society" (Denzin & Lincoln, 2005, p.12). Other typical critique against the positivists, who are obsessed with numbers, is that their myriad formulas diverge from the practical life world of people, and that they are in danger of merely reducing people to numbers (Neuman, 2000).

Constructivist/interpretivist research

Even though there are differences in accent between these two concepts I, for the sake of brevity, will follow Ponterotto (2005, p.129) by discussing them together. Contrary to positivism, the constructivist/interpretivist perspective holds that there is no objective reality "out there" which can be readily accessed for research purposes, but instead that reality exists in the minds of people. It is constructed socially, in other words, when people interact with each other; reality is based on shared meanings, and is subjective rather than objective. This ontological position is referred to as *relativism*. In sharp contrast to positivism, the researcher and participant are not regarded as separate entities; they, in fact, can hardly be because they are continuously co-creating reality inter-subjectively. The epistemological

position is therefore transactional and subjectivist (Guba & Lincoln, 1994; Ponterotto, 2005; Schwandt, 1994).

The purpose of interpretivist research is therefore not to discover an objective reality, if it exists, but instead to understand and *interpret* people's motivations for their actions, how they construct their lives, the meanings they attach to it and the *context* in which the drama is acted out. Science is not neutral; value free research is not possible. Important subdivisions of this paradigm include symbolic interactionism, phenomenology, hermeneutics and ethnography (Sarantakos, 1997).

An example of the constructivist approach in CID- research is the work of Kathy Charmaz (1983, 1990, 2000), who investigated identity and loss of self in the chronically ill using constructivist grounded theory. The present research may also be positioned in this paradigm since the roots of grounded theory, the chosen methodology that will be delineated later, is largely considered to be symbolic interactionism (Corbin & Strauss, 1990; Coyne & Cowley, 2006; Kendall, 1999; Kools, McCarthy, Durham & Robrecht, 1996).

Critical research

The theoretical basis of the critical perspective is a combination of conflict theory, critical sociology, Marxism and feminism. The main task of research, according to this approach, is social critique in order to expose restrictive practices, injustices, social domination and power imbalances, as it exists in the status quo, with a view of eliminating it. Whereas, basically, positivists regard reality as objective and interpretivist view it as being subjective in nature, the critical perspective is somewhere in between. Both subjective meanings and objective relations are important (Myers, 1997; Sarantakos, 1997).

The ontological position, according to Guba and Lincoln (1994) is *historical realism*. A "virtual reality" (Guba & Lincoln, 1994. p.110) has crystallised over time in the context of values relating to, amongst others, political, economic, ethnic and gender inequalities. Critical research's epistemological view is similar to that

of constructivism, namely a transactional/subjectivist position, but values (both researcher's and participant's) play a more important role here (Ponterotto, 2005).

With regard to chronic illness and disability research, it can be concluded that the critical paradigm is involved implicitly or explicitly in studies that focus on discrimination against and stereotyping of the ill and disabled, including poor access to amenities.

As has been expounded, the three research paradigms are fundamental to *all* research forms and types, including qualitative research, which will be pursued in this study. An elucidation of qualitative research follows.

QUALITATIVE RESEARCH

Qualitative research is difficult to define and has no distinct paradigm and set of methods that is entirely its own. It encompasses various approaches based on different philosophical perspectives and even has a generic variant that either claims no theoretical viewpoint at all or combines several approaches (Caelli et al., 2003; Denzin & Lincoln, 2005).

Denzin and Lincoln (1994, p.2) describe qualitative research as follows: "Qualitative research is multi method in focus, involving an interpretive, naturalistic approach to its subject matter. This means that qualitative researchers are studying phenomena in its natural settings, attempting to make sense of, or to interpret, these phenomena in terms of the meanings people bring to them". According to them, the history of qualitative research has progressed through various phases or moments (they identify five such moments) since the early 1900s. Although different methodological foci tended to come to the fore more prominently during each of the historical phases, Denzin and Lincoln (1998) state that: "Qualitative research, as a set of interpretive practices, privileges no single methodology over any other" (p.5). They additionally quote Nelson et al. (1992) who write: "No specific method or practice can be privileged over any other, and none can be eliminated out of hand" (p.5).

In terms of methods as such, qualitative researchers use a large variety of these, for example narrative, archival and discourse analysis, as well as semiotics and grounded theory. They may even use statistics (Denzin & Lincoln, 1998).

A very useful and often encountered way to introduce qualitative research is to contrast it with quantitative research. The latter, encompassing the hypothetico-deductive method, is almost entirely rooted in the positivist and post-positivist paradigms. Verifying of hypotheses in a tightly controlled experimental situation and application of statistical techniques are hallmarks of quantitative research (Ponterotto, 2005). The differences between quantitative and qualitative research are frequently depicted in tabular form and I will follow suit: in this instance, the data depicted in **Table 3.1** is adapted from Schurink (1998, p. 242-243), who in turn based his table on Denzin & Lincoln (1994) and Neuman (1994). Since the differences between qualitative and quantitative research are legion, the table does not purport to be all-inclusive, and furthermore contains generalisations.

Table 3.1: Selected comparisons between qualitative and quantitative research
(Adapted from Schurink, 1998, pp.242-243)

QUALITATIVE RESEARCH	QUANTITATIVE RESEARCH
Uses an inductive form of reasoning: develops concepts, insights and understanding from patterns in the data.	Uses a deductive form of reasoning: collects data to assess preconceived models, hypotheses and theories.
Uses an emic perspective of inquiry: derives meaning from the subject's perspective.	Uses an etic perspective: the meaning is determined by the researcher.
Is ideographic: thus aims to understand the meaning that people attach to everyday life.	Is nomothetic: aims to objectively measure the social world, to test hypotheses, and to predict and control human behaviour.
Regards reality as subjective.	Regards reality as objective.
Captures and discovers meaning once the researcher becomes immersed in the data.	Tests hypotheses that the researcher starts with.
Concepts are in the form of themes, motives and categories.	Concepts are in the form of distinct variables.

Observations are determined by information richness of settings, and types of observations used are modified to enrich understanding.	Observations are systematically undertaken in a standardised manner.
Data are presented in the form of words, documents and transcripts.	Data are presented by means of exact figures gained from precise measurement.
The research design is flexible and unique and evolves throughout the research process. Cannot be exactly replicated.	The research design is standardised according to a fixed procedure and can be replicated.
Data are analysed by extracting themes or categories.	Data analysis is undertaken by means of standardised statistical procedures.
The unit of analysis is holistic, concentrating on the relationships between elements, context, etc. The whole is always more than the sum.	The unit of analysis is variables, which are anatomistic (elements that form part of the whole).

Ambert, Adler, Adler and Detzner (1995) point out that qualitative research in general focuses on the "how and why" of human interaction, including how people create mutual understanding, insight and meaning. The *context* in which human behaviour occurs is regarded as crucial. They furthermore state that: "… qualitative research frequently falls within the context of discovery rather than verification" (p.880). Qualitative research is thus concerned with the life worlds of people; their feelings, motivations and the use of symbols such as language and gestures (especially the different levels of meanings embedded in it) (Berg, 2004; Henning, 2004). Berg (2004) makes the valid point that many human experiences cannot be meaningfully expressed in numbers, for instance recollections of the nature of smell, sight and sound qualities.

Over the years, relations between the qualitative and quantitative research positions have proved not to be overly harmonious, with both "camps" criticising the other's alleged weaknesses (Kelle, 2006). For me, from a pragmatic viewpoint, there is little to be gained by discrediting either qualitative or quantitative research. Kelle (2006) pointedly asserts that both approaches have strengths and weaknesses.

Arguments over many years between the qualitative and quantitative camps have not been particularly productive, if at all; proponents of each position tend to merely emphasise weaknesses of the other tradition instead of clarifying their own. Therefore, it amounts to which approach is best suited to the project at hand. Kvale (1996, p.69) puts it as follows: "In conclusion, qualitative and quantitative methods are tools, and their utility depends on the power to bear upon the research questions asked". I chose qualitative research as the main methodology for this study because it allows me to better investigate the feelings, interpretations and meanings of people who are living with CMT, and thus reach my research objectives.

Regarding qualitative methodology in health research: "... the rapid proliferation of qualitative approaches in the health sciences began in the 1980s" (Thorne, Joachim, Paterson & Canam, 2002, p.2). The types of illnesses studied are diverse. In a meta study where they reviewed almost 300 qualitative studies done between 1980 and 1996 on some aspect of living with a chronic disease, Thorne et al. (2002) found that certain diseases were more popular as areas of focus than others were. Breast cancer, diabetes, asthma and cardiovascular diseases were the most often studied, whereas Chronic Obstructive Pulmonary Disease and hypertension were much less popular. Rheumatoid arthritis, prostate cancer, multiple sclerosis, epilepsy, pain and psoriases are further examples of conditions studied by means of qualitative methodology. Charcot-Marie-Tooth disease, the focus of this study, does not appear on the list; neither does any other disease in this group of neuropathies.

In terms of the qualitative method/strategy that is the most frequently encountered in CID research, Thorne et al. (2002) found in their meta study that, of the studies that explicitly reported their methodological direction, 29% were grounded theory studies, 25% were phenomenological, 13% ethnographical and 33% a range of diverse, less prominent methodologies.

Before continuing with the discussion of rigour in qualitative research, a fundamental phenomenon that influenced many choices pertaining to methodology

(and related issues) needs to be briefly looked at. It concerns my role as a researcher.

The role of the researcher

In qualitative research, the researcher is mostly regarded an integral part of the process of meaning construction; in fact, he or she is the co-constructor of meaning. This personal involvement and concomitant subjectivity is however not seen as something to be avoided, but rather as an asset in the sense of utilising the researcher's experiential knowledge (Meulenberg-Buskens, 1997). Henning (2004) states that the researcher may in fact be regarded as the main instrument of research. Following from this, it can be deduced that it will be an impossible task to fully control for or abolish researcher bias in qualitative research (Cutcliffe, 2000; Caelli et al., 2003; Denzin & Lincoln, 1994; Mantzoukas, 2004; Patton, 1990).

A qualitative report should therefore include information about the researcher's position - in the words of Greenhalgh and Taylor (1997, p.5): ….." the most that can be required of the researchers are that they describe in detail where they are coming from so that that the results can be interpreted accordingly." It is important that any personal and/or professional information that may have affected data collection, analysis or interpretation be reported (Patton, 1990). Mantzoukas (2004) extends this idea by stating that researchers should also report the motivations for their decisions and provide an audit trail "of his or her decision-making pathways" (p.1003).

In this study, I occupy the dual position of researcher and participant. Ambert et al. (1995) summarise research on this position as reflecting two main viewpoints. On the one hand, there is the advantage of the researcher, as a member of the group being studied, being able to provide fresh, unique insight and information. In addition, it can also assist in reducing the distance created by the hierarchical relationship between the researcher and the other. However, the danger is always there that the researcher may be blinded to nuances and idiosyncrasies, as well as being biased in some way. Getting very close to the people they study, although

not limited to researchers as participants, may result in researchers "… abandoning the researcher role altogether. Or close friendship may blind the researcher to unpleasant facts" (Ambert et al., 1995, p.887).

Having been diagnosed with CMT and having had this condition for many, many years, the possibility of undesirable bias on my side exists. Being aware of this fact, however, puts me in the position to concentrate on minimizing the potential negative effects. Henning (2004) briefly discusses a number of ways how this can be achieved, for example when the findings are presented, they should be backed up by enough empirical evidence, complemented with a strong theoretical base and presented in a coherent, logical and convincing argument.

One of the reasons why I chose grounded theory as the main methodology for this study is because the method is structured and encompasses a fair amount of guidelines and procedures that, if followed, should increase rigour and ultimately trustworthiness (Charmaz, 2000; Fassinger, 2005). In addition, as will be shown under the next heading, a few basic verification measures is deemed necessary and would be employed in order to combat bias, increase rigour and enhance the credibility of my findings.

Notwithstanding concerns regarding the possibility of bias, I feel strongly that, ultimately, my having this disease constitutes an asset rather than a liability for the study. Not only can my illness experiences add value in terms of delineating the intricacies of the condition, but also in the sense of better insight and understanding of others who are affected by CMT. Patton's words will however always be in the back of my mind: "Any research strategy ultimately needs credibility to be useful. No credible research strategy advocates biased distortion of data to serve the researcher's vested interests and prejudices" (1990, p.55).

Rigour and verification in qualitative research

Unfortunately, certain tensions between quantitative and qualitative research appear not to have languished. One of the positivist's main arguments is that verification is lacking in qualitative research, that "these researchers have no way

of verifying their truth statements" (Denzin & Lincoln, 2005, p.8). It should be mentioned that people who use the standards of quantitative research to judge the qualitative alternative, are often disappointed (and vice versa!). The two approaches encompass contrasting orientations and it is best to appreciate the strengths that each offers. Quantitative research is almost exclusively rooted in the positivist paradigm: the world of quantifiable, measurable variables, statistics, hypothesis testing and a linear research path. In contrast, by its very nature, qualitative research is non-standard, unconfined, non-linear and dependent on the subjective experience of both researcher and the researched. So called "soft data", for instance words, utterances and symbols, rather than numbers, are used in qualitative research (Greenhalgh & Taylor, 1997; Neuman, 2000).

The strength of the quantitative approach lies primarily in its reliability. This is also known as repeatability, meaning the same measurements should yield the same results over time. The strength of qualitative research on the other hand lies in its validity, or closeness to the truth (Greenhalgh & Taylor, 1997).

Even though it may be more productive to appreciate and concentrate on the strengths of both approaches, value judgments and critical comparisons, at times comprehensive and even debilitating, are frequently made (Kelle, 2006). Berg (2004, p.3) comments as follows on this matter: "Unfortunately, because qualitative research tends to assess the quality of things using words, images and descriptions, whereas quantitative relies chiefly on numbers, many people erroneously regard quantitative strategies as more scientific than those employed in qualitative research". He then pointedly asserts: "this is not to suggest that qualitative methods are without methodological rigour. In fact good qualitative research can be very rigorous" (p.3). The processes involved in knowledge development, the latter regarded by many as the central aim of research, should be followed meticulously and conscientiously in order to ensure that this knowledge is not flawed and subsequently of little use (Caelli et al., 2003). Morse, Barrett, Mayan, Olson and Spiers (2002), as well as Patton (1990), assert that rigour is always a desired goal in research, irrespective of whether quantitative or qualitative methodologies are used.

Morse et al. (2002) highlight two main approaches that are used to enhance quality in qualitative research, namely post hoc evaluation strategies and in-process verification strategies. Post hoc evaluation strategies encompass evaluation of the research after the completion thereof and the criteria used often deviate from conventional quantitative criteria of reliability and validity. The problem is that reliability and validity criteria stem from the quantitative paradigm and are not pertinent to the qualitative approach. In response, Lincoln and Guba (1985) formulated the concept of trustworthiness as a substitute for reliability and validity, specifically for use in the evaluation of qualitative studies. Trustworthiness encompasses four subcategories, namely credibility, transferability, dependability and confirmability. They furthermore emphasised the crucial aspect of researcher integrity.

Notwithstanding the contribution of Lincoln and Guba (1985) as described above, Morse et al. (2002) maintains that ad-hoc strategies, no matter how good they are, primarily aim to *evaluate* rigour, but they do not *ensure* rigour as such because errors cannot be corrected timeously. They maintain that it is better to build the verification strategies into the research process as such and then use them during the inquiry to identify and correct errors timeously, ensuring rigour and trustworthiness. Strategies that are used include investigator responsiveness, methodological coherence and the concurrent collecting and analysis of data.

In this research, trustworthiness, credibility and rigour will be addressed by (1) taking transcriptions, analysis and theory pro forma back to the participants for evaluation and feedback purposes (in-process strategy); (2) adhering to the procedures and methods of grounded theory as far as possible (in-process strategy); (3) applying a selection of Lincoln and Guba's (1985) criteria of trustworthiness, which includes triangulation (in-process and ad hoc strategies).

A practical method to enhance validity, which will be employed in this study, is triangulation. Basically, triangulation encompasses the principle of looking at something from different angles to enhance understanding and ultimately improve quality (Berg, 2004; Greenhalgh & Taylor, 1997; Neuman, 2000). The types of

triangulation that will be employed are methods-triangulation and researcher triangulation. Regarding the former, five psychometric instruments and a biographical questionnaire will be administered in addition to unstructured interviews, and the supervisor of this study will co- analyse at least one interview concerning the latter form of triangulation. Another researcher will also check the interview transcripts and coding.

Qualitative methods

In view of the many different types or genres of qualitative research, it is to be expected that there will also be a myriad of matching research strategies, designs and methods. Hybrid designs in various configurations are also possible (Henning, 2004). A comprehensive delineation of the myriad research strategies, which include different designs, genres and methods, will not be embarked upon. Instead, six of the more prominent strategies/approaches will be introduced, eventually concentrating in more detail on grounded theory, the chosen method of this study.

Case study research

This entails the focused investigation of a phenomenon, which is defined as a single clear unit of analysis with identifiable boundaries. The unit need not be an individual, but could be a group, a program, an organization and so forth (Henning, 2004).

Phenomenology

The phenomenological position got momentum in the early 60s with the philosophical perspectives of Husserl and Heidegger. In the late 60s, Schutz focused on advancing the methodological aspects of the approach (Goulding, 2005). The focus in phenomenology is on the nature and structure of experiencing of phenomena (for example emotions, a job, a marriage etc) in order to make sense of the world. The experiential world, namely how individuals experience events, is what matters. Lived experience is a key concept. The analytic process used is characteristic, including steps such as Epoché, bracketing, theme identification and portrayal, and structural synthesis (Patton, 1990).

Ethnography

Essentially, ethnographic research aims to find answers to the question of what the culture of a group of people encompasses. This includes settings such as organizations and communities, as well as smaller groupings. Participant observation, where the researcher is placed in the midst of the research setting, is the prime method used. Intensive fieldwork in natural settings is distinctive. The view that culture is simply a matter of shared beliefs and practices, has been expanded to account for concepts like unequal access to power and other concepts of critical ethnography. Ethnography has evolved and today may be applied to most social research in everyday settings, using several methods and focusing on the meanings and explanations of people's actions rather than quantification (Savage, 2000; Patton, 1990).

According to Goulding (2005), data analysis in ethnography typically utilises techniques such as content analysis. The process begins with identifying and labelling of concepts in the data, followed by grouping them into themes or categories. Often, ethnographic research suffices with "thick description", where the researcher comprehensively describes the observed behaviour, ways, events, and meanings reflected in the data, focusing on the emic perspective. The conceptual level of analysis, however, varies and may focus increasingly on syntheses, "where the data is pooled and the constructed categories are linked" (Goulding, 2005, p.300).

Ethnomethodology

Ethnomethodology aims to answer the question of how people make sense of their everyday activities; to investigate the norms, understandings and assumptions that are so deeply rooted that they are taken for granted (Patton, 1990).

Action research

Henning (2004) holds that the impetus for this method is usually a sense of social action. The aim is emancipation of the participants and the research is undertaken in collaboration with them, which can result in unfortunate ethical dilemmas if care is not exercised (Myers, 1997).

Narrative inquiry

Chase (2005) sees this interesting qualitative approach as a dynamic field still in the making. It encompasses a wide range of interdisciplinary approaches, methods and analytic lenses "all revolving around an interest in biographical particulars as narrated by the one who lives them" (p.651). In the field of CID research, narrative analysis has been applied to HIV/AIDS, multiple sclerosis and cancer research, to name a few.

As already indicated, grounded theory is the research strategy selected for this research. The depiction thereof that follows commences with a discussion of the reasons why this particular research method was judged to be more suitable than other qualitative methods.

GROUNDED THEORY (GT)

Rationale for choosing grounded theory

In the quest for a suitable research method, above-mentioned and other qualitative strategies were considered, but none of them was regarded entirely suitable to satisfy the research questions. This does not at all imply that the methods are inherently inferior (quite the contrary!); in fact, it was practical reasons that were more often than not responsible for ruling them out.

By way of example, an ethnographic study would hardly have been attainable, mainly because prolonged direct contact with participants in their natural settings, a hallmark of ethnography (Goulding, 2005), was not possible. The main reasons for this amount to difficulty in locating (recruiting) participants, as well as the vast geographical distribution amongst those that were successfully recruited. This aspect will be elaborated on in *Part 2* (praxis). Not being able to fully address the research questions was another fundamental reason for discounting certain methods, such as phenomenology. The latter, arguably focusing more on developing a deep understanding for the way in which people experience their life worlds (Goulding, 2005), appears not to adequately address the spirit of the second research question, which amounts to the action taking component (in other words,

the actions that the people are taking in order to deal with their problems). Grounded theory specifically accentuates processes (action) as being the focus of the analysis (Glaser, 1978).

Many qualitative methods also tend to focus on thick description. In due course, however, I became uneasy thinking whether description alone really goes far enough for what I had in mind. The supervisor of this study played no minor role in these developments as she prompted me on various occasions to think of ways to contribute something that will directly or indirectly help this group of people.

Even though, in the words of La Rossa (2005, p.855), "Qualitative research can be descriptive and spectacular", my uneasiness with description being the only focus of my study, is shared by researchers such as Cutcliffe and McKenna (2004), quoted as follows by Cutcliffe (2005, p.423): " While there is nothing inherently wrong with descriptive level studies we, as a scientific community, need to do much more with our current research than describe. Indeed, the most illuminating qualitative findings go further than description: they interpret, they explain; they solve problems". Cutcliffe (2005, p. 423) continue in this context that qualitative research may run the risk of failing the "so what?" test, in other words, it should rather aim to have more utility in the "practical" world.

These considerations led me to grounded theory. This method, at least as originally conceived, specifically aims to formulate a theory in the area of interest, whilst also allowing for varying degrees of description. Grounded theory encompasses a clear set of procedures and techniques that, if correctly applied, will be of immense value, not only in the practical execution of the research, but also in ensuring rigour. Furthermore, it is a comprehensive methodology. Consider what Glaser (1998, p.14) says: "Grounded theory is to my knowledge the only methodology that guides the researcher from the moment he enters the field to a final, publishable draft". By using data collection techniques such as unstructured interviews, exploration of the subjective experiential world of participants, as it is constructed via language, may be possible, given that views on this matter may differ depending on one’s philosophical perspective.

Charmaz (2000, p.522) aptly summarises the strengths of grounded theory as lying in: "(a) strategies that guide the research step-by-step through an analytic process, (b) the self-correcting nature of the data collection process, (c) the methods' inherent bent toward theory… and (d) the emphasis on comparative methods".

Grounded theory is therefore able to meet requirements (a) to (e), stated in the introduction to *Part 1* of this chapter. They may be summarised as follows:

a) In view of the paucity of research regarding the psychosocial aspects of living with CMT, the suitable method will have to allow for the exploration of fundamental aspects such as the illness experiences of CMT-affected people, as well as how they manage the effects of the neuropathy in order to cope with it.
b) Because I occupy the dual role of researcher and participant, the application of a rigorous method, as well as the use of clear prescribed guidelines, steps and procedures, are advisable to minimise researcher bias as far as possible.
c) The chosen method will have to provide suitable procedures and techniques that enable the thorough and orderly analysis, as well as the description, of concepts.
d) Difficulties in locating research participants are anticipated; the suitable method therefore should be able to accommodate working with a rather small group (small N).
e) Finally, the elected method should ideally allow for the formulation of a theory on how adults with CMT manage the symptoms, effects and manifestations of the disease themselves in order to optimise their well-being.

Glaser's (1965, 1978, 1992, 1998) more classical version of grounded theory, pursued in this research, is considered particularly suitable for satisfying requirements (a) and (e).

Overview and historical development

Grounded theory was developed in the 1960s by two sociologists, Barney Glaser and Anselm Strauss. It evolved out of the methodology that the two of them developed when they were studying the phenomenon of dying in various hospitals (Glaser, 1998). Eventually, in 1967, the approach culminated in their book" The

discovery of grounded theory". This publication has since established itself as a seminal work in the qualitative tradition (Cutcliffe, 2005). However, according to La Rossa (2005, p.840), Glaser had already developed the central tenants of grounded theory earlier, namely in a 1965 article (Glaser, 1965).

At that stage, and even today, grounded theory entailed fundamentally different ways and methods compared to the then prevalent logical-deductive research model, and in effect constituted an alternative way of doing social research. The derivation of hypothesis from pre-existing theories and then subjecting them to testing and analysis for verification purposes, were hallmarks of logical-deductive research. In contrast, Glaser and Strauss (1967, p.1) describe their method as: "the discovery of theory from data", meaning that the theory is inductively derived from and grounded in the data. To this end, a variety of data, for example transcribed interview text and medical or other records, are analysed using grounded theory procedures such as constant comparison, theoretical sampling, coding, memo writing and sorting. Conceptual categories are constructed, the relationships between key ones specified, and theory developed in this process. The conditions under which these theoretical relationships are constructed, maintained and changed are delineated (Charmaz, 1990; Glaser & Strauss, 1967).

Strauss and Corbin's (1990, p.24) concise definition of grounded theory is often cited: "The grounded theory approach is a qualitative research method that uses a systematic set of procedures to develop an inductively derived grounded theory about the phenomenon." Glaser (1978, p.2) describes the method as follows: "Grounded theory is based on the systematic generating of theory from data, that itself is systematically obtained from social research. Thus the grounded theory method offers a rigorous, orderly guide to theory development..." He (2002, p.1), provides additional, and perhaps more operational, clarification regarding the theoretical aspects: "All that grounded theory is, is the generation of emerging conceptualizations into integrated patterns, which are denoted by categories and their properties. This is accomplished by the many rigorous steps of grounded theory woven together by the constant comparison process, which is designed to

generate concepts from all data. Most frequently, qualitative data incidents are used ".

After a relatively slow start, grounded theory has grown in popularity over the years and has made inroads into a myriad of disciplines, for example psychology, education, communication, health science and management. It has furthermore inspired the development of various computer software analysis programs (Cutcliffe, 2005; Strauss & Corbin, 1994). According to Ten Have (2004, p.136) grounded theory is "currently probably the most popular way of doing (and accounting for) qualitative analysis…" Cutcliffe (2005, p.421) states that grounded theory has actually developed into a "global phenomenon", whereas Fassinger (2005, p. 156) quotes Denzin (1997) in describing grounded theory as: "the most influential paradigm for qualitative research in the social sciences today".

Despite its popularity, grounded theory methods are, even after all these years, still generating considerable dialogue and controversy; in the words of Walker and Myrick (2006, p.547): "…it (grounded theory) is the most frequently discussed, debated, and disputed of the research methods". At the heart of the controversy lies a methodological disagreement, or split, between the two originators of the method that has resulted in public disagreement and eventually in each going his own separate way (Melia, 1996).

Glaser and Strauss came from two very different backgrounds. Strauss received his training at the University of Chicago, which had a long history and strong tradition in qualitative research. In fact, Kendall (1999) states that the Chicago School of sociology may be regarded as the place where symbolic interactionist theory was developed. Glaser, on the other hand, received his training at Columbia University, which had a quantitative research tradition (Glaser, 1992; Strauss & Corbin, 1990). According to Charmaz (2002), these dual origins have produced a mixture of advantages and disadvantages. She furthermore claims that Strauss's background gave grounded theory its open-ended emphasis on process, meaning, action and usefulness. Glaser's background that is more positivistic contributed

empiricism, rigorous codified methods and the specialised language of grounded theory.

The increased usage and popularity of grounded theory has however not resulted in focused refinement of the methodology's original canons and methods, but instead in the evolution of different variants, some of which deviate markedly from the original formulations. The method may in fact be seen as still maturing and branching (Annells, 1997a; Clarke, 2003; Dey, 1999). Many of the extant variants are often strongly disapproved of by Glaser (1992, 2002). This applies particularly to his colleague Strauss, who had in due course developed his own version of grounded theory that differed significantly from Glaser's, differences that continued and even intensified in Strauss's collaborative writings with Juliet Corbin (1990, 1998). Unfortunately, "what started as a most productive partnership between Glaser and Strauss ended in something akin to acrimonious divorce" (Dey, 2004, p. 80).

Although differences between the two versions will become apparent throughout the discussion that follows, the main disagreements will be specifically addressed later in this chapter, and will highlight the reasons why I chose to follow Glaser's (1965, 1978,1992, 1998) approach. Although there are minor differences, Glaser's approach is seen as being the closest to, and regarded by some as being, for all practical purposes the same as, the original method as delineated in 1967 in "*The discovery of grounded theory"* (Kendall, 1999; Rennie, 1998). Being the chosen version for the present research, Glaser's approach will naturally receive greater prominence in the discussions that follow.

Grounded theory paradigms

The *theoretical underpinning* of grounded theory is predominantly regarded as symbolic interactionism (Annells, 1997a; Corbin & Strauss, 1990; Coyne & Cowley, 2006; Cutcliffe, 2000; Fassinger, 2005; Goulding, 2005; Kendall, 1999; Kools et al., 1996). Symbolic interactionism focuses on the importance of shared meanings in human interactions, as well as the role of language and other symbols in these processes. The influential role of pragmatism in symbolic interactionism,

and consequently in grounded theory, has also been recognized (Charmaz, 2000; Clarke, 2003), including John Dewey's instrumentalism (Rennie, 1998).

Herbert Blumer (1969), one of the most influential symbolic interactionists, formulated three fundamental premises that this paradigm rests on:

1. Human beings act towards physical things and other beings in their environment based on the meanings that these things have for them.
2. The meanings arise from social interactions that people have with others. We communicate by means of language and other symbols; communication is the symbolic phenomenon.
3. The meanings are established and modified by means of interpretative processes.

Amongst others, these premises contradict traditional views regarding the origin of meaning, for example, that meaning is inherent to the objective makeup of the thing that has it, as well as that meaning is an expression of psychological elements (Blumer, 1969).

To summarise, through their interactions with one another, people are continuously creating shared meanings via the use of shared symbols such as words, gestures and dress. In the process, they construct their realities (Fassinger, 2005). Kendall (1999) and La Rossa (2005) accentuates the crucial role of language. La Rossa (2005) cautions that researchers who contemplate doing grounded theory based on theoretical perspectives other than symbolic interactionism, should do their utmost to ensure that language is central in that approach. The reason for this is that "words are the indicators upon which GTM-derived theories are formed" (p.838).

Despite the above delineation, a number of authors (see, for example Charmaz, 1990, 2000; Denzin & Lincoln, 1998; Guba & Lincoln, 1994; Ponterotto, 2005) maintain that the paradigmatic home of grounded theory is rather ambiguous and that it in fact is still being debated. In this regard, Charmaz (1990, p. 1164) writes: "Glaser and Strauss's earlier works have both phenomenological and positivistic emphases and therefore, sometimes may seem confusing and even inconsistent".

In her 2000 article, she identifies both positivistic and post-positivistic elements in the method. Ponterotto (2005, p.133) holds that the former (positivistic elements) refers to underpinnings of an objective nature, as well as coding and data analytic procedures that are explicit and rather reductionist. The latter (post-positivism) mainly refers to a verification orientation, which is characteristic of Strauss and Corbin's (1990) grounded theory version (Denzin & Lincoln, 1998; Ponterotto 2005).

Other paradigmatic locations pertaining to later versions of grounded theory include constructivism (Bryant, 2003; Charmaz, 2000), post-structuralism/critical (Fassinger, 2005) and "methodological hermeneutics" (Rennie, 1998, p.110). Fassinger (2005, p.157) has a rather useful, pragmatic perspective regarding the home of grounded theory, namely that it can be positioned anywhere on a continuum ranging from positivism to constructivism and even to post-structuralism (critical theory).

In this study, I take the classic view that is arguably encountered the most often in the literature, namely that grounded theory is underpinned by symbolic interactionism. My position is aptly summarised by La Rossa (2005, p.847) who states: "Based on symbolic interactionism and other language - orientated frameworks, GTM encourage the study of how `reality´ is socially constructed (e.g., how dying is socially constructed or how love is socially constructed)". In this research, I will therefore be studying how living with CMT (broadly speaking) is socially constructed.

However, as has been shown, the earlier versions of grounded theory (Glaser, 1965, 1978, 1992; Glaser & Strauss, 1967; Strauss & Corbin, 1990), which includes the version that will be followed in this study, contain elements of positivism and post positivism, particularly regarding coding procedures and data analysis techniques. The reasons for deciding upon Glaser's approach will be delineated later in this chapter. The fact that data analysis will contain post-positivistic elements is neither seen as negative nor positive, since I take the view put forward by Ambert et al. (1995), who are decisive that methods should be

selected on the basis of utility and appropriateness in the context of the research questions, "and not on the basis of a reviewer's epistemological preference" (p.888).

Aim

The broad aim of grounded theorists, at least according to Glaser's approach, is to identify and explain basic social and psychological processes (BSP's) (Cutcliffe, 2005; Glaser, 1978). Glaser (1978, 1992, 1998) states that there is an endeavour to understand the action of the participants in a particular substantive area; specifically what their main concerns or problems are as well as their continuous efforts to resolve them. Grounded theory research wants to discover the core variable that resolves the main concern or problem, a process that culminates in the formulation of a theory. In practice, qualitative data are mostly used, but quantitative data can also be accommodated (Charmaz, 2002; Glaser, 1978, 1999).

Essentially then, the aim of grounded theory is to inductively formulate a middle range theory about a particular substantive area (Glaser, 1992; Fassinger, 2005; La Rossa, 2005). Grounded theory can however also be utilised to formulate formal theory. Middle range theories are described by Glaser and Strauss (1967, p.33) as "…fall(ing) between the 'minor working hypotheses' of everyday life and the 'all inclusive grand theories'"(parenthesis added).

A distinction is also made between substantive and formal theories. Glaser (1978, p.144) summarises these as follows: "By substantive theory we mean theory developed for a substantive or empirical area of sociological inquiry - such as patient care, race relations, professional education, geriatric lifestyles, delinquency, or financial organizations. By formal theory we mean theory developed for a formal or conceptual area of sociological inquiry -- such as status passage, stigma, deviant behaviour, socialization, status congruency, authority and power, reward systems, organizations or organizational careers". La Rossa (2005) states that substantive theories are topic specific, whilst formal, or generic, theories, transcend issues.

These two levels of theory are both generated by means of comparative analysis. In the case of substantive theory, comparative analysis takes place between or among groups/units within the same substantive area. Formal theory requires comparative analysis of different kinds of substantive areas within that formal area. The difference between the two types of theory lies in their degree of conceptual abstraction (Dey, 1999; Glaser & Strauss, 1967).

The grounded theory itself, i.e. the research output, takes the form of a theoretical formulation or an integrated set of conceptual hypotheses about the particular substantive area. The hypotheses are in effect probability statements about the relationship between concepts, or categories (Glaser, 1998). Glaser (1992, 1998) regards these as the final product of grounded theory; further verification and testing are left to other, mostly quantitative researchers. As far as format goes, the theory may be presented in one of two ways: either as a set of codified propositions or as a running theoretical discussion or narrative using the identified categories and their properties. The discussion format is my preferred option because it gives a feeling that the theory is dynamic and always developing, according to Glaser and Strauss (1967).

Glaser (1978; 1992), building on criteria developed in the *Discovery of grounded theory* (1967), is decisive that a "good or proper" grounded theory must meet the criteria of fit, work, relevance and modifiability.

Fit refers to the requirement that formulated categories must fit the data from which they were developed in the data analysis process, and that they must adequately explain the data. If existing concepts are used, they must at all costs earn their way into the analysis and not be forced into it.

Work: For a grounded theory to work, it should adequately interpret and explain the behaviour in the substantive area (what is going on).

Relevance has to do with the requirement of the analytic formulations of grounded theory being able to explain actual problems and processes in the area of interest.

Modifiability refers to flexibility, that is to say, formulations and even theories can be modified as conditions change and/or new data is accessed.

The role of literature

Upon engaging with a new research project, it is best to start with as little preconceived ideas as possible (Dey, 1999; Glaser, 1998). The reason for this is that concepts and theory should emerge naturally from the data without colouration from prior influences. It should certainly never be forced into preconceived schemas and perspectives. Reading should be of a general nature only, for example to peruse different styles and the way in which others ordered their writings (Glaser, 1992). Glaser and Strauss (1967) actually advise researchers to ignore the literature and theory in the area under study until much later, after core categories have emerged. Ten Have (2004, p.139) appears to be rather sceptical of this: "Glaser and Strauss go as far as recommending theoretical ignorance as a strategy to foster open mindedness." Others, for example Charmaz (1990), are more flexible on this point, provided that prior knowledge is used as sensitising concepts only and never forced onto the data to make it fit. Naturally, at later stages when the study is framed within the literature, a thorough review of all relevant literature and extant theories should be undertaken.

Interrelatedness of the data collection and analysis processes

Unlike many other research methods, where all the data is collected before analysis commences, data analysis in grounded theory begins as soon as the first bit of data is collected. Thereafter, analysis and collection occur alternatively; each analysis directs the next interview or observation. The focus throughout is on questions and matters that enlighten the research questions, unless analysis at any stage indicates the irrelevancy of the latter (Corbin & Strauss, 1990).

Categories and their properties

Categories may be regarded as the cornerstones of the emerging theory. They are formed by grouping concepts that refer to the same phenomena together, and then assigning conceptual labels or names to the categories. Categories are at a higher level of abstraction than the concepts on which they are based and are also generated through the analytic process of constant comparison (Corbin & Strauss, 1990). The question whether categories may legitimately be regarded as variables,

is rather controversial. According to La Rossa (2005, p.843), many grounded theory interpreters, amongst them Glaser (1978; 1992), Charmaz (2000) and Creswell (1998) do indeed equate categories and variables.

Properties are conceptual aspects or elements of a category, more specifically the attributes or characteristics pertaining to a category, whereas *dimensions* encompass placing the properties somewhere along a continuum Categories may furthermore be broken down into sub-categories, which specify the category in terms of it's what, where, why, when and how qualities. Names for categories may be borrowed from the technical literature or existing theory, provided that the researcher ensures that they fit the data and are not forced on the data. In many instances, it is possible to name categories in vivo, that is to say, to use participants' own words. Generating ones own concepts does not carry the potential dangers of influencing the researcher with preconceived meanings and associations (Glaser & Strauss, 1967; Strauss & Corbin, 1990, 1998).

Theoretical sampling

Sampling in grounded theory is not aimed at representing a population, but instead is aimed at developing the emerging theory. As categories are developed and conceptualizations raised to higher levels of abstraction, gaps and needs become apparent, for example, certain categories may lack sufficient evidence. Another example is where the analyst is unsure if important information pertains only to particular cases. Researchers then purposely select new cases in order to obtain the required data and develop the categories. Theoretical sampling furthermore assists the researcher in defining a category's properties, the conditions that influence it and its relationship with other categories (Charmaz, 2002; Kendall, 1996).

Constant comparison

Comparisons are the main analytic tool in grounded theory. In the words of Dey (2004, p.88): "comparison is at the core of grounded theory, whether comparing bits of data to generate categories, or comparing categories in order to generate

connections between them. Comparison is the engine through which we can generate insights, by defining patterns of similarity or difference within the data".

Glaser (1978) distinguishes three types of comparisons in the context of theoretical sampling and theory generating. Firstly, incident is compared to incident to establish the underlying uniformity and its conditions. In this process, concepts are generated, as well as tentative hypotheses. Secondly, the generated concepts are compared to more incidents to elaborate and densify the theoretical properties of the concept and to generate more hypotheses. Thirdly, on an even higher conceptual level, concept is compared to concept. Boeije (2002) proposes a five step analytic approach to the procedure of constant comparison, of which comparisons within a single interview and comparisons between interviews are the most relevant to this study. In the former case, fragments of data are compared for similarities and differences in order to identify and develop categories, whilst in the latter case the focus is on patterns or combinations of categories.

Strauss & Corbin (1998) add a further type, which they call "theoretical comparisons" (p.94), to be employed when there is uncertainty how to code a particular incident or when an inability to think about incidents in different ways is experienced. Comparisons in this instance are with the literature and/or one's previous experience. As will be shown later, Glaser (1992) laments this facet of Strauss and Corbin's work. Although coding and constant comparison are intertwined processes in grounded theory, coding will now be discussed under a separate heading, as is often done in the literature.

Coding

Together with constant comparison, coding is regarded as a key analytic process in grounded theory. Either it encompasses a two or three-step process, depending on which version of grounded theory is used (Dey, 1999). Charmaz (2002, p.684) explains coding: "In essence, coding is a form of shorthand that distils events and meanings without losing their essential properties". Glaser (1992, p.38) defines coding as: "Conceptualizing data by constant comparison of incident with incident, and incident with concept to emerge more categories and their properties".

The first coding stage is called initial or **open coding**, which is defined by Strauss & Corbin (1990, p.61) as: "the process of breaking down, examining, comparing, conceptualizing and categorizing data". In open coding, a few fundamental questions are repeatedly asked of the data. Glaser's (1992) questions include: What category or property of a category does this incident indicate? What is the main problem faced by the people studied? What is the basic process that processes the main problem that makes life viable in the action scene? Questions by Strauss & Corbin (1990, p.77) include: Who, when, where, what, how, how much and why? Lastly, Charmaz's (2005, p.514) questions include: What is happening? What are the people doing? The unit of analysis may be a single line, a sentence, a paragraph or even and entire document (Strauss & Corbin, 1998), but the emphasis is on line by line analysis since it enhances detailed, close analysis of the data (Dey, 1999).

Data is scrutinised and events/actions/interactions compared with each other for similarities and differences. Conceptual labels are allocated to data fragments and gradually concepts that pertain to the same indicators are grouped together to form higher order concepts, namely categories. A category is described by Strauss and Corbin (1990, p.61) as the higher order grouping together of concepts that, in the process of comparison to each other, appear to pertain to the same/similar phenomenon. Furthermore, sub-categories may emerge, and both categories and subcategories are then developed in terms of their properties and dimensions. The process of open coding proceeds until theoretical saturation occurs, that is, a particular incident adds no new information and repeatedly indicates the same concept/category (Corbin & Strauss, 1990; Strauss & Corbin, 1990). Glaser (1978, p.56) regards open coding as the initial step of what he calls "substantive coding"; the second step is selective coding were the analyst engages in focused coding for an emerging core category.

A second intermediate phase in the coding process, called **axial coding**, was developed by Strauss and Corbin (1990) and is not found in Glaser's version of grounded theory. In fact, as will be shown in the next section, axial coding is controversial in the sense that Glaser totally rejects it having any role in grounded

theory. Axial coding basically entails the reassembling of data that was broken down and fractured during open coding. Here, categories are related to their sub-categories on the property and dimensional levels in order to render more complete and dense explanations about a phenomenon. This linking is done by means of a *coding paradigm* that denotes conditions, actions/interactions and consequences. Structure is linked to process and answers to the questions why, when, where, how come, by whom and how are sought (Strauss & Corbin, 1998). In coding, axial and open coding are not necessarily sequential steps and in practice overlap. Strauss and Corbin (1990, p. 99) feel so strong about their coding paradigm that they say: "unless your use this model, your grounded theory analysis will lack density and precision". Not everybody feels this way, though. Like Glaser, Charmaz (2002) also does not do axial coding, claiming that it adds complexity without improving the analysis.

According to Strauss and Corbin (1990), the final coding phase, namely **selective coding**, need only be attempted if theory building is the aim of the research. If the intended end result is conceptual description, the process may be stopped after axial coding. Glaser (1978; 1992), however, feels that conceptual description per se is never the aim of grounded theory. Selective coding is done by both Glaser and Strauss and Corbin. For the research findings to take the form of a theory, the major categories need to be integrated, related and refined. The focus in selective coding is on the identification and development of an emergent core category, which represents the central phenomenon and that integrates all the other major categories around it. Another aim is the filling in of poorly developed categories (Glaser, 1978, 1992; Corbin & Strauss, 1990).

Theoretical coding is another type of coding that Glaser uses, but not Strauss and Corbin. "Theoretical codes conceptualise how the substantive codes may relate to each other as hypotheses to be integrated into a theory" (Glaser, 1978, p.78). This is done by using one of Glaser's 18 coding families. Even though these are preconceived frameworks, Glaser maintains that the appropriate code family (the one that fits) should emerge from the research and not forced on the data. The

researcher is furthermore free to develop his or her own ones. For illustration purposes, I selected seven codes from Glaser (1978):

Coding family	*Examples*
Six Cs	Causes, contexts, contingencies, consequences, co- variances and conditions
Process	Stages, phases, progressions
Degree	Limit, range, intensity
Type	Type, form, kinds, styles, classes
Strategy	Strategies, tactics, mechanisms
Consensus	Clusters, agreements, contracts
Models	Linear, spatial

The first family, the six Cs, is seen as the "bread and butter" theoretical code, the first general code to keep in mind when coding data. One of Glaser's major criticisms against Strauss is that the latter focused on only one of the coding families, namely the six Cs, and in effect just called it the paradigm. Furthermore, he gave no recognition to Glaser's work on theoretical coding. All the other families, and Glaser even lists a few more in his 1998 publication, which allows for such a wide range of possibilities, are ignored by Strauss and Corbin (Glaser, 1978, 1992, 1998; Ten Have, 2004).

Memo writing

Memo writing is analogous to writing notes to oneself, a mechanism whereby the researcher engages in an extended ongoing dialogue with him or herself (Charmaz, 1990). Here, ideas and conceptual thoughts are written up, and codes are developed in terms of their properties and theoretical relationships between them. Glaser (1978) regards memo writing as crucial for generating grounded theory, an activity that cannot be skipped at all. Strauss and Corbin (1990, 1998) distinguish between different types of memos, namely code notes, theoretical notes, operational notes and various types of diagrams. On this matter, I follow Boychuck-Duchscher and Morgan (2004, p.610), who states: …"this reductionistic and fragmented approach to memo writing has the potential to overwhelm the researcher by its complexity and potential ambiguity." I found in practice that one

generic memo worked much better for me - I can concentrate on substance and need not worry about what type of memo it is that I am busy with.

Grounded theory aims to produce theories that are conceptually dense, that is, with many conceptual relationships. These relational statements are best presented by weaving them into a narrative, but a set of "if-then" propositions may also be used (Strauss & Corbin, 1994). For Glaser (1978), the sorting of memos into a theoretical framework according to analytic rules is a crucial step of integration and theory development. Decisions have to be made what must go where and relationships clarified. Conceptualisation thus takes place on a high level.

Divergences in grounded theory and the selection of a suitable version

Despite providing a large number of guidelines in their original book, Glaser and Strauss (1967, p.8) were not rigid and dogmatic regarding their procedures. Instead, they allowed for the possibility of others adapting and developing grounded theory. La Rossa (2005) states that this pluralistic design of grounded theory has indeed resulted in many adaptations and even drastic developments, of which the most profound (arguably) embraces the disagreements and eventual split between Glaser and Strauss themselves. He writes as follows in this regard: "Deciphering and evaluating the two versions-referred to as the Glaserian and Straussian schools by some… has turned into a cottage industry, with various individuals choosing sides or advancing their own similar-but-not-identical-to-GTM methodologies" (p.839).

Whereas this study concerns itself primarily with the differences and similarities between the two originator's grounded theory versions, it needs to be pointed out that the evolvement of grounded theory takes on considerable proportions, with many versions of the method seeing the light. These states of affairs lead to Dey (2004, p.80) remarking as follows: "There is no such thing as 'grounded theory' if we mean by that a single, unified methodology, tightly defined and clearly specified. Instead we have different interpretations of grounded theory…" Examples of grounded theory formats include dimensional analysis, developed by Leonard Schatzman (1991), in Kools, et al. (1996), constructivist grounded

theory, developed by Cathy Charmaz (1990; 2000) and situational mapping and analysis, developed by Clarke (2003).

With so many versions, Annells (1997b) aptly lists the following essentials, taken from the original formulations by Glaser and Strauss (1967), that should *always* be included for any variation to be regarded as a grounded theory method: (a) theoretical sampling, (b) constant comparative data analysis, (c) the need for theoretical sensitivity, (d) memo writing, (e) the identification of a core category and (f) the ideal of theoretical saturation.

For the purposes of this study, the accent is on comparing Glaser's (1965, 1978, 1992, 1998) and Strauss and Corbin's (1990, 1998) versions of grounded theory, with a view of selecting the most suitable one to implement. A discussion of these differences, and the delineation of the salient reasons for choosing the former, follows.

Since the publication of Glaser and Strauss's "*Discovery of Grounded Theory"* in 1967, more than a decade elapsed before another publication on grounded theory as a methodology saw the light. It was a solo effort by Glaser, namely "Theoretical Sensitivity" in 1978. From that point on, many publications on grounded theory by Glaser and Strauss saw the light, but never again as co-authors. In fact, over the years, grounded theory publications clearly reflected major methodological differences between the two, as well as between Glaser and other grounded theory researchers (Boychuck-Duchscher & Morgan, 2004).

Even though Strauss published his own version of grounded theory in 1987, it was his book "Basics of Qualitative Research", written in collaboration with Juliet Corbin in 1990, that really evoked controversy and generated debate that is still active today (Ten Have, 2004). Strauss and Corbin intended this book to help researchers, especially beginners, in the complex ways of doing grounded research. From the beginning, the book has been fiercely repudiated and opposed by Glaser, who felt that it was in many ways contradictory to the original assumptions and canons of grounded theory. In response, Glaser published his " Emergence vs.

Forcing - Basics of Grounded Theory" in 1992 in which he attempted to correct what he considered to be mistakes by Strauss and Corbin and "to set the average researcher back on the correct track to generating a grounded theory" (p.6). Glaser felt that Strauss and Corbin had deviated so far from grounded theory that they had in fact developed a new methodology in its own right, which he called " full conceptual description", which is definitely *not* grounded theory (Glaser, 1992, p.62).

Most, if not all, of the critical comments encountered in the literature about Strauss and Corbin's (1990, 1998) alterations to the original grounded theory method, embrace *the reasons why* I decided to follow Glaser's approach rather than Strauss and Corbin's, or any other altered, version. Hence, it is necessary that the most important criticisms be briefly discussed and linked to the present research.

Rennie (1998, p.102, 105-107) aptly summarises Strauss and Corbin's (1990) additions to grounded theory. He states: "Strauss and a colleague, Julia Corbin, have altered the original grounded theory procedures in several respects… their modifications to the basic method is fourfold: 1) the investigator's recalled experiences pertaining to the phenomenon under study are accepted as legitimate empirical data, 2) hypothesis testing is made integral to constant comparison, 3) consideration of the conditions influencing the phenomenon should not be limited to those indicated by the data themselves, and 4) the application of an axiomatic scheme that converts all social phenomena into a process is made mandatory" (numbering added). Each will be briefly commented on (in italics).

1. The original grounded theory holds that all relations conceptualised in the data by the analyst, should meticulously be supported by the presence of indicators, or signs in the data. Strauss and Corbin expand indicators to include the researcher's prior experiences that he or she introspectively accesses.

For me, who occupies the dual role of researcher and participant in this study, the dangers of bias is reduced by meticulously focusing on the data and using only conceptual indicators present in the data during analysis. Whereas, in the words

of Cutcliffe (2000, p.1478) "Few would dispute that qualitative methods invariably involve interaction between the researcher and the data", the risk of my subtly biasing the analysis and/or venturing into irrelevant relationships on the grounds of my prior experiences with CMT, (which may be idiosyncratic), is nevertheless considered an unnecessary one. This risk can be diminished and managed more effectively by using Glaser's approach, with his relentless insistence on presence of indicators in the data and emergence of concepts.

2. Strauss and Corbin forwardly hypothesize relations in the data (even though the data might not support relations at that stage of the analysis), and then return to the data in order to confirm or verify the hypothesis. They (1990, p.111) state: "… we deductively propose statements of relationship or suggest possible properties and their dimensions when working with data, then actually attempt to verify what we have deduced against data as we compare incident with incident. There is a constant interplay between proposing and checking ". Walker & Myrick (2006) draws our attention to the fact that Strauss and Corbin (1990) had markedly deviated from grounded theory's inductive focus by stating that their coding procedures involved both deductive and inductive elements.

Although a degree of deduction in grounded theory's analytic procedures is unavoidable (La Rossa, 2005), an inductive approach will largely be followed in this research, and the cycle of hypothesis formulation and subsequent returning to the data for verification, will be limited to an absolute minimum, if engaged at all. This is in accordance with Glaser (1978, 1992), who has always argued that the fundamental purpose of the grounded theory method is to develop theories that are induced from, and are accountable to, the data, and not to verify it.

Embedded in number 2 above is the matter of *verification* in grounded theory, a major point of dispute. Strauss and Corbin (1990; 1994) hold that a striving towards verification is essential. Steps to ensure verification are in fact built into their grounded theory and takes place throughout the entire research process. Glaser (1992) in turn feels that the aim of grounded theory is to generate

propositions and not to test them- testing and verification should be left to other researchers, as a separate study after formulation of the grounded theory.

This study deviates from Glaser's anti-verification stance, because a few verification measures in line with the "traditional canons of good science" will indeed be undertaken, albeit to a limited extent. There are three main reasons for this. Firstly, verification is considered a fruitful endeavour to counter the possibility of bias due to my being a member of the group studied. This aspect has been discussed at length under the heading "role of the researcher" earlier in this chapter. Secondly, by following Glaser's classical grounded theory approach, the possibility of forcing that may result from deducing something and then returning to the data to verify what I found, is reduced, if not entirely eliminated. Thirdly, since I am not an experienced user of grounded theory methodology, I feel that the few verification measures taken (please refer to the section on verification in this chapter) may add to the credibility of my findings.

3. Rennie (1998, p.106) states that Strauss and Corbin (1990) advocate taking into consideration *all* conditions thought to be applicable to a category, whether indicated in the data or not. In fact, the conditions are introduced on rational grounds and promote the use of Strauss and Corbin's conditional matrix. In addition, the analysing of macro societal levels, for example economic conditions, cultural values and so forth, deviates from classic symbolic interactionist underpinnings, which focuses more on micro societal issues (Annells, 1997a, Corbin & Strauss, 1990).

Since Glaser's approach does not include the conditional matrix, it will not be used in this research. In data analysis, only those conditions that are relevant to the conceptual categories (in other words, those that exert influence), are to be regarded as useable. Besides, this study focuses more on the experiential world of the individuals living with CMT and the structural conditions impinging upon them; the focus is not on automatically analysing macro societal influences per se. The latter, for example the myriad laws, policies, structures and so forth pertaining to the disabled, has been documented extensively (DPSA, 2001), and, as said, will

have to earn its inclusion into the analysis (must have a presence in the data) (Glaser, 1992; 1998).

4. This point by Rennie mainly refers to the use of the coding paradigm in axial coding. As the title of Glaser's (1992) book, "Emergence versus forcing", makes clear, the thrust of his main criticism is that, in Strauss's hands, the grounded theory approach has degenerated from a methodology to a set of methods with a focus on preconceived, forced conceptual description (Ten Have, 2004). In particular, this criticism regarding the forcing of data is directed at axial coding, which entails using a coding paradigm to relate categories to their *preconceived* sub-categories, namely conditions, action/interaction strategies and consequences (Dey, 1999; Glaser, 1992; Kendall, 1999). Glaser (1992) feels strongly that the paradigm boils down to nothing more than the blatant forcing of data and that it furthermore leads to elaborate description of concepts rather than the generation of theory. Rennie (1998) highlights an additional argument by pointing out that the coding paradigm amounts to forcing all social phenomena into being "processual", whereas the original method accentuated both structural and process concepts, depending on the data.

In view of the above, as well as reasons discussed earlier in this chapter under coding procedures, the coding paradigm is not considered an acceptable option in the present research. Instead, the emergence of concepts via traditional Glaserian (La Rossa, 2005) grounded theory procedures, including constant comparison, coding, memo writing, and sorting, will be focused on, and forcing of data in any way will be guarded against. Glaser's (1978) coding methods, with its focus on non-forcing, will in particular be adhered to. Even though Charmaz (2000) generally laments the positivistic elements in traditional grounded theory, her words regarding data analysis reflect my own view: "Glaser's comparative approach and emphasis on process provides excellent strategies for making data analysis efficient, productive, and exciting - without formulaic techniques. Every qualitative researcher should take heed of his warnings about forcing data into preconceived categories through the imposition of artificial questions" (p.514). To

re-iterate, the fundamental criticism of Strauss and Corbin forcing data by means of their preconceived coding paradigms and questions, does ***not*** *fit with the inductive, emergent nature of Glaser's grounded theory (Wilson & Hutchinson, 1996).*

My uneasiness about the above alterations by Strauss and Corbin to the grounded theory approach is one very important motivation for using Glaser's approach instead. Another entails Glaser's focus on basic social processes (BSP's). According to this approach, the discovery and subsequent formulation of basic social processes (in both substantive and formal areas of social life), is a fundamental aim of grounded theory (Charmaz, 2000; Clarke, 2003; Cutcliffe, 2005; Ekins, 1997; Glaser, 1978). Melia (1996) states that the absence of BSP's in Strauss and Corbin's (1990) book served as further evidence to Glaser that this duo had in fact developed a different method under the banner of grounded theory.

For Glaser, the generation of theory, via the concepts of a core category and its subcategories, which is mostly embedded in the process concept of BSP, is the fundamental aim of grounded theory. This does not entirely apply to Strauss and Corbin (1990), who state that the grounded theory process may be terminated before reaching the stage of theory generation, if the aim is conceptual description. Kendall (1999) is sceptical about Strauss and Corbin's (1990) methods' abilities in the context of theory generating and states: "I agree with Glaser (1992) that axial coding via the paradigm model is inconsistent with the work necessary to generate useful and dense theory (p.756)".

Another argument of Melia (1996, p.376) and Kools et al. (1996, p.315) that I identify with, is that Strauss and Corbin's method is permeated with a plethora of rules and cumbersome procedures that may actually hamper theoretical sensitivity. In this regard, the former writes as follows: "I always have a nagging doubt that the procedures are getting in the way; the technical tail is beginning to wag the theoretical dog" (p.376). The use of axial coding, particularly the coding paradigm, has the danger of focusing too much attention on "working the model", thus distracting from what the data is saying and concomitant theory construction

(Fassinger, 2005; Kendall, 1999). The latter (p. 751) recollects her experiences when she attempted to use the paradigm as follows: "I became so distracted by working the model to its natural conclusion that I stopped thinking about what the data were telling me in regard to the research question… working the paradigm model became an end in itself…" She continues in this vein and illustrates how the paradigm failed to lead to theory construction, which she overcame by using Glaser's approach. Stern (1994, in Charmaz 2002) asserts that Strauss and Corbin's additions to grounded theory, namely dimensionalising, verification, axial coding and the conditional matrix, actually erode grounded theory.

To summarise what emerges clearly in the above delineation, I have decided to essentially follow Glaser's classical grounded theory approach in this study. Outstanding positives for me in Glaser's approach include his idea of basic social processes, the primacy of generating theory, his focus on understanding the problem, or main concern, of the participants coupled to what they are doing to resolve it, his relentless insistence on non-forcing, the non-preoccupation with rules/prescriptions and his focus on conceptualisation.

Furthermore, his view of context is the usual, orthodox one and in line with the requirements of this study, whereas Strauss and Corbin (1990, p.101) add the rather idiosyncratic angle that context is "the specific set of properties that pertain to a phenomenon". According to Glaser (1992, p.65): "A context is a condition of overriding scope, under which a set of related categories and properties occur". By way of example, he explains that soldiers act in a context of authoritarianism. The fact that many regard Glaser's approach as the more difficult approach due to it being less formulaic and prescriptive in terms of operational steps (Kendall, 1999), is counterbalanced by the allowance for more creativity as well as the lack of distractions from one's data in the form of prescriptions. Lastly, Glaser's approach is generally seen as reflecting the original grounded theory approach (Rennie, 1998; Walker & Myrick, 2006).

Salient criticisms, issues and challenges

Many of the criticisms and related issues that qualitative research grapples with apply to a lesser or larger extent to grounded theory as well. In view of the tensions between the qualitative and quantitative traditions (discussed elsewhere), it is perhaps to be expected that much of this disapprobation stems from the positivist school of thought. Qualitative researchers have typically been referred to as soft scientists whose work is unscientific, merely exploratory and imbued with bias. Qualitative research has even been reproached for being an "attack on reason and truth!" (Denzin & Lincoln, 1998, p.7, exclamation mark my own).

Two of the most pervasive criticisms concern the issues of *rigour* in qualitative research, as well as limitations regarding the *generalisability* of qualitative findings, especially in the light of the small number of participants typically encountered in qualitative studies (small N) (Charmaz, 2000; Denzin & Lincoln, 1998). Because qualitative research mostly uses words as data and not numbers, is non-linear, and focuses on the subjective experiences of the parties, it is often accused of lacking rigour and verification measures. However, as was discussed at length elsewhere in this chapter, qualitative research can indeed be very rigorous and incorporate both in-process and ad-hoc strategies. In addition, Lincoln and Guba (1985) developed specific criteria to ensure quality in qualitative research.

The second area of concern resorts under (the alleged) lack of generalisability of qualitative findings, which includes Lincoln and Guba's (1985) similar (Kelle, 2006) concept of transferability. Criticism in this regard can hardly be more vivid than the original words of Lundberg (1929/1942), quoted by Kelle (2006, p. 303): "The scientific value of all these (qualitative studies) depends, of course, on the validity of the subjective interpretations of the authors as well as the extent to which the cases selected are typical. Neither the validity of the sample nor of the interpretations are objectively demonstrable on account of the informality of the method".

Early qualitative researchers responded by way of the concept of theoretical generalisation, claiming that the strength of theoretical reasoning in qualitative

research is superior to statistical inference. General social processes are at work in all cases, and are identifiable even in idiosyncratic cases. However, this approach rested on the assumption that social rules and history are stable and universal, and it was later replaced by micro sociological approaches such as symbolic interactionism. The latter claims the opposite, namely that social life is unpredictable and contingent. Lately, particularly post-structuralists and post-modernists hold the conviction that the context-boundness of *all* social phenomena effectively renders the idea of generalisation superfluous (Kelle, 2006).

Moving to grounded theory, what is the response to the generalisation issue? Glaser (1992) unambiguously states that the standard approach encountered in quantitative and verification studies, namely to generalise from a sample to a population, does not apply to grounded theory, or at least not to his version. This type of generalisation applies to unit analysis and not to the analyses of processes, which is the focus in grounded theory. Instead, what *does* apply to grounded theory, is the generalisability from a substantive theory, where the scope is rather limited, to "a process of larger scope with parsimony" (Glaser, 1992, p.117), based solely on the former's ability to fit, work and be relevant to the latter (more general) social processes. By way of example: how generalisable is *engaging with CMT*, the basic social process that emerged in this study, to the social processes in other inherited neurological conditions, and even in inherited diseases in general?

Regarding critical comments about grounded theory, Charmaz (1990, p.1164) posits that most criticisms of grounded theory stem from misunderstanding and/or misuse of the method. In addition, she aptly points out that many of these criticisms apply equally to both quantitative and qualitative research in general. Examples are a lack of thorough exploration that typically result in premature decisions and closure (such as what categories to use), the use of unnecessary jargon and not describing key terms adequately. Arguably, the proper way to deal with these issues, and that will be followed in this study, is to guard against it with a view of eliminating, or at least substantially reducing, it.

Fassinger (2005) points out that grounded theory has been criticised for its excessive fragmentation of data and for being "extremely time-consuming and expensive" (P.164). She (p.164) correctly summarises Woolley, Butler and Wampler's (2000, p.317) list of limitations as follows: "the methodology is labour-intensive, draws heavily on the conceptual skills of the researcher, requires explicit acknowledgement of researcher biases, is difficult to report succinctly because it relies on extensive examples from narratives and can be used only with small samples". Certain issues, such as labour intensiveness and costliness, either are unavoidable, or may be handled by way of optimal management.

Concerning researcher bias, I have already (throughout this chapter) elaborated clearly on my dual role as researcher/ participant, and have clearly stated the possible sources and management of bias. Drawing extensively from narratives and reporting on them elaborately (non-succinctly), is not in the present research seen as a limitation, but instead as an asset, especially in view of CMT being such an unknown disease (Crabtree, 2000), as well as the paucity of research on the psychosocial aspects of living with it.

Other potential problems with grounded theory referred to by Charmaz (1990, p.1164), include: (1) "glossing over" the epistemological assumptions of the method, including how it pertains to levels of explanation, (2) ambiguity regarding the use of researcher's prior theoretical perspectives, and (3) minimising grounded theory's relation with extant theory. An explanation as to how these concerns will be addressed in this study follows.

The epistemological aspects have already been clearly stated throughout this chapter. To reiterate, symbolic interactionism will underpin the study, with Glaser's (1965; 1978; 1992) grounded theory version, which contains post-positivist elements (Annells, 1997a&b), comprising the method that will be employed to analyse data and construct theory. Glaser (1998, p.8) states that "all is data". In a later document (Glaser, 2002b), he indicates that grounded analysts regard constructivist data, in other words data constructed intersubjectively between researcher and participant, as valid data. However, he is of the opinion

that constructivist data does not comprise a large proportion of grounded theory data, which is rather contradictory to this study. Interviews (data collection), will largely be unstructured and open ended, and constructivist principles will therefore be operative.

Data analysis in the present research will primarily be based on data that has been collected, in other words, interview transcripts and various documents. In line with Glaser's approach, the extant literature will be accessed later in the research process, and not at an early stage, as is recommended by Strauss and Corbin (1990). In rare cases where labels for concepts (and similar information) may indeed be taken from the extant literature, it will be from studies and literature that Glaser (1992, p.36) describe as "pure descriptions", which, according to him, may be accessed earlier in the analysis process. It is crucial that this information earn its way into the analysis via the process of constant comparison (Glaser, 1992).

Comparisons with extant theory will only be undertaken after a core category with its subcategories, has clearly emerged (Glaser, 1992). In this document, integration with extant theory will be delineated in the discussion chapter. It needs to be said though, that for the most part, these comparisons will have to take place with generic models about the adaptation to chronic diseases, of which many were developed via the hypothetico-deductive method. No existing grounded theory on living with CMT, or for that matter any other inherited neuromuscular condition, could be traced.

We will now proceed into the operational section of this chapter where the procedures followed, sampling, data collection and data analysis will be elucidated. The distinction between the praxis and theoretical sections is by no means absolute and indeed overlaps. Theoretical concepts may therefore surface again.

PART 2: THIS THESIS' GROUNDED THEORY PRAXIS

All research, including the present one, aims to answer the research question(s) of that particular study. These were formulated in Chapter 1, and were repeated verbatim in the beginning of this chapter. In this study, answering the research questions was done by way of a substantive grounded theory on how adults with CMT manage the manifestations, effects and challenges that stem from the disease (the second component of the research question) in order to optimise their adaptation to it and augment their well-being. The theory also embraces the first part of the research question, which amounts to how the disease subjectively affects them.

The version of grounded theory that informs the methods of this research, is the so-called "classical" grounded theory, formulated by Glaser (1965, 1978, 1992, 1998), which is closest (Rennie, 1998; Walker & Myrick, 2006) to Glaser and Strauss's (1967) original version. This method directed my processes of data collection, data analysis and theory construction. The formulation of an inductively derived theory is the only and fundamental aim in Glaser's grounded theory version. I will now explain how the grounded theory study was carried out. It starts by describing sampling procedures and characteristics, followed by data collection and, lastly, data analysis.

PARTICIPANTS AND SAMPLING

The Muscular Dystrophy Foundation of South Africa (MDF SA) supplied the names of all participants that participated in this research except one. The participant that was sourced differently had to produce written medical proof of his diagnosis. All relevant documentation provided by the participants, such as doctor's letters, as well as applicable employer letters pertaining to, for example, diminished work performance due to CMT, was included in the analysis. The type of CMT played no role. A requirement was a minimum age of 18 years, because this study focused on adults.

An earlier strategy, that is to say before the MDF route, to source participants via local neurologists and hospitals, was unsuccessful. Despite numerous calls and letters informing them about the project and asking for referrals, only one referral was received, which unfortunately never materialised.

The list of names provided by the MDF proved to be invaluable. It was however rather outdated because people had failed to inform the MDF about changes in domicile. According to the list, the largest concentration of people with CMT was in Gauteng province, with very few names scattered across the rest of the country. After telephonically updating the list, not too many names remained, which complicated theoretical sampling, mainly because of the limited number of people who were available for follow-up purposes. Sampling in the sense of selecting people strictly according to rather comprehensive predetermined criteria for inclusion in the study, was not an option.

Despite the difficulties, theoretical sampling was implemented as far as possible. For instance, based on telephonic information, a number of participants were included, and later interviewed on a second occasion, because they had for certain passed the mutant gene to their descendants, a theme that was necessary to follow up. Theoretical sampling was the main reason for conducting a second round of interviews in Gauteng. Purposive sampling was applied to a limited extent. Because this research focused on adults, people below the age of 18, an arbitrarily decided upon cut off point, were excluded and it was decided to concentrate on the Western Cape and Gauteng provinces. It was also attempted to include both sexes in equal proportion as far as possible, but as can be seen in Table 3.1, there were more females.

Eventually, 11 people with CMT participated in the research: eight from Gauteng Province and three, including myself, from the Western Cape. All the people who were invited to participate accepted the invitation. One CMT-affected person from Durban accepted, but not once returned any telephone calls despite promising to do so and was excluded due to lack of interest. The most important sample characteristics are depicted in **Table 3.2.**

Table 3.2: Sample characteristics

	N	%
Gender (N=11)		
Male	4	36,36
Female	7	63,64
Age group (N=11)		
30-40	2	18,18
41-50	3	27,27
51-60	3	27,27
61-70	3	27,27
Marital status (N=11)		
Married	10	90,91
Widowed	1	9,11
Employment (N=11)		
Retired	4	36,36
Homemaker	2	18,18
Working from home	2	18,18
Employed casually	1	9,11
Employed fulltime	2	18,18
Language (N=11)		
Afrikaans	6	54,55
English	5	45,45
Education (N=11)		
Pre-matric	2	18,18
Matric	4	36,36
Post-matric	5	45,45
Age when diagnosed with CMT (N=11)		
10-20	3	27,27
21-30	3	27,27
31-40	4	36,36
41-50	1	9,11

Nine participants were from the white and two from the Indian/Asian population groups. The average age of participants was 48, 6 (range 38-70), whereas the age at which they were diagnosed with CMT ranged from 16 to 47, with an average of 29, 36 years. A seven point Likert scale was administered, according to which the participants were asked to rate the severity of their CMT symptoms. A subjective experience of "most severe" was indicated by a score of 7, while a "least severe"

rating was indicated by a score of 1. Even though resorting more in the results chapter (Chapter 4), it may be apt to mention here that all candidates except one rated their condition as above average or average in severity.

Seven people used helping aids or similar (specialised) devices, for example walking sticks and orthopaedic apparatus, although not necessarily on a full time basis and/or of a "serious, or intrusive" nature (this does not refer to sturdy, lace-up shoes or boots, which more or less everybody wore). None of the participants was wheelchair bound, not even partially.

DATA COLLECTION

Three sources of data were utilised in the grounded theory analysis: (1) transcribed recordings of qualitative in-depth interviews, (2) biographical data questionnaires, and (3) medical and other relevant documents, for instance doctor's letters, where supplied. An example of a letter of diagnoses may be viewed in **Appendix D**. A fourth source of data, namely a battery of psychometric questionnaires, was only used for the purpose of triangulation, and will be discussed separately, in Part 3, of this chapter. For the sake of orderliness, the biographical questionnaire will also be discussed in Part 3, together with the other questionnaires.

With the exception of one person who chose to be interviewed at his place of employment, all participants were interviewed in their homes. Before commencement of any procedures, participants voluntarily signed a written consent form that addressed issues such as the aim of this study, anonymity and voluntary withdrawal at any stage (Warren, 2002). Refer to **Appendix B** for an example of the consent form. Six participants completed the biographical and other questionnaires directly after completing of the interview. The rest chose to complete them in their own time and return them by post. A pre-paid envelope was provided for this purpose. In the latter case, I explained in detail how to complete the questionnaires. I supplied my contact details, should any problems or questions arise, with a return call procedure that resulted in no cost for the participants. Initially, I thanked participants for their time by means of a shopping voucher, but after many objected to this, it was not maintained.

Interviews

The interviews lasted between 60 and 95 minutes and were audio taped with the participants' permission. In two instances, the spouse was present. All the interviews were conducted by myself. Interviews were transcribed verbatim. I guided the process, but an assistant did most of the typing because my finger coordination is rather impaired due to CMT. In addition, my speech recognition program could not be used with the Afrikaans interviews since it only recognises English.

The two participants from the Western Cape were interviewed first, followed by the nine from Gauteng. The latter was done in two trips five months apart. The reason for this was to finalise coding before proceeding with more data collection, mainly for theoretical sampling purposes.

The type of interview used was the qualitative in-depth interview (Johnson, 2002). Kvale (1996) uses the metaphor of being either a miner or a traveller to describe the interviewer's approach. As a miner, the interviewer mines for facts or nuggets of essential meaning, which resides in the subject's interior, waiting to be dug out. The knowledge nuggets remain constant and uncontaminated even after transcription to a written format. In the traveller metaphor, the interviewer embarks on a journey where he or she wanders with the people, conversing and asking questions so that they can tell their own stories of their lived world of experiences. The interviewer interprets the meanings in the stories and knowledge is constructed in the process. Since I am more comfortable with the traveller metaphor, I largely followed this approach.

Johnson (2002) states that one of the salient differences between qualitative in-depth interviews and other interviews amounts to greater involvement of the interviewer's self in the former. The interviewer's role includes the offering of some form of reciprocity in order to enhance mutual trust and aid self-disclosure. Complementary reciprocity applies when the interviewer is a non-member, while strict reciprocity is only possible if the interviewer is a current or former member of the group studied. Considering that this amounts to the interviewer giving

his/her opinion and sharing experiences on issues discussed, the advantages are clear to many.

Having CMT myself therefore enabled me to employ strict reciprocity in all the interviews. Membership, however, is a two-edged sword. Interviewers, who are members, should be aware of how membership knowledge might influence them and guard against pitfalls, such as preconceived ideas. Interviewers without membership knowledge, on the other hand, may have difficulty in recognising nuances and meanings.

The interviews were largely unstructured and open-ended. I, using techniques such as open-ended questions and prompts, ensured that the following broad themes were covered: (1) experiences regarding CMT in childhood, (2) diagnosis related experiences, (3) handling of stressors because of the disease, (4) present and future concerns, (5) the most salient contextual influences, and (6) support systems. Participants mostly addressed these themes spontaneously and very little directive behaviour on my part was necessary.

DATA ANALYSIS

Interview transcripts were analysed more or less concurrently with data collection as it proceeded in the three rounds as described, using grounded theory's constant comparative method (Glaser, 1965, 1978, 1992; Glaser & Strauss, 1967). After the first interview, the supervisor of this study and I had a working session during which we analysed, or coded, the particular transcript together. This was mainly done to ultimately enhance trustworthiness of the findings. In addition, a psychometrist presently involved in coding of qualitative data, checked the rest of the interview transcript's coding. The principle of completing the analysis of a script before proceeding to the next interview was adhered to as far as possible. Information from the background information forms, as well as medical and other relevant documents if supplied, was included in the analysis. This is in line with grounded theory procedures and in fact reflects Glaser's (1998, p. 142) notion that "all is data".

The analysis began with *open, or substantive, coding* of the interview transcripts. Data was studied line by line and broken down into units of meaning, or what Glaser (1998, p.140) calls "the incident". For me, regarding sentences or parts thereof as units of analysis worked better than the customary line-by-line coding. Each unit of analysis was confronted with the usual questions specified by Glaser (1978, p.57) such as: (1) what category or property of a category, does this incident indicate? (2) What is the basic social psychological problem(s) faced by participants? (3) What is the basic social psychological process that processes the problem? By using these procedures, concept indicators were identified, concepts emerged and the concepts were labelled. These codes took the form of gerunds in order to reflect movement, or process, in the data. By using action codes, the focus remained on what the people were *doing* and avoided the pitfall of merely listing topics (Charmaz, 2002). As far as possible, in-vivo codes were used, such as "finding a way", "never surrendering" and "investigating the basics".

By way of example of how open coding was done, an extract from an interview with a participant follows:

179. No, I don't see myself as disabled. Not at all. I may be slower or different,
180. but that's all. Like, you know, a lot of places we go to, they ask me to take off
181. my shoes. What they don't know is that for me it is quite a mission. Mostly I
182. wear these laceups. But nowadays I tell them that I'm slow; it takes me
183. longer to get things done. It is better that they know.

Coded as follows:
179-180: Resisting identity of being disabled.
180-181: Context.
181-182: Experiencing lack of understanding.
182-183: Informing others.

Coding was done directly on the transcripts, where a wide margin was left for this purpose. These codes, which for practical purposes may be regarded as "earlier type" conceptualisations of indicators in the data, were additionally, in due course,

transferred to separate pages (more or less a page per code). Here, as the incidents/ indicators under each code increased and it emerged more clearly in due course, a short description of the code, its properties and possible relationships with other codes, was recorded. The exact location of supportive evidence was also indicated, for instance P2: 346-351, meaning participant two, lines 346-351. By way of constant comparison, that is by comparing incident to incident, incident to concept and concept to concept, the conceptualisations were continuously re-worked, condensed (in some cases expanded) to eventually form rather loose conceptual categories. They were loose because they were ever evolving as coding proceeded. The relevant code-pages were sorted into the applicable categories, once again on a continuous basis.

At any time during the entire coding process, coding was interrupted to write memos. This was done whenever important ideas, insights or developments, in other words with important theoretical implications, occurred or were noticed in the data. Examples were the identification of possible relationships between concepts, when patterns/trends were noticed or when conditions such as contextual factors clearly exerted influence. A vast number of memos were produced, although, admittedly, many were no longer than half a page. Memo writing was found to be an invaluable tool in the analysis, especially in the integration phases.

The above procedures facilitated further development and refinement of the categories. By now, all categories had clearly been named, once again using in vivo terminology as far as possible. Sub categories of the categories had also been identified at this stage. Being at a higher conceptual level, the development of categories furthermore included the specification of the properties of each category. Constant comparison has been expanded to include the comparison of categories with sub-categories. The overriding aim was to identify and elucidate relationships between categories and between categories and their sub-categories. The way in which participants experienced diagnosis, for example, related to a myriad other processes, and did so in different ways.

Gradually, as coding and continuous re-working of categories continued, one broad, overriding process (category) that represented all the other categories emerged. The coding phase of *selective coding* had now been reached. This central phenomenon, the core category, or (as was the case in this study) the **core process**, integrated all the other processes, or categories, around it. Not only were the relevant categories of the core, now called sub-cores, related to the core, but also the core was able to explain how the sub-cores related to each other. From this point on, coding proceeded on a selective basis in the sense that it was now focused on developing the core process and its sub processes, or sub-cores. "Interacting with medical fraternity" is an example of a subcategory that was not developed further because it did not relate strong enough to the core. Admittedly, a property thereof, "gathering information from medical professionals" was used.

In line with the principles of theoretical sampling, later interviews with participants were focused on selective coding. The filling and developing of the core and its sub-cores thus continued. Gaps were filled and the relationships amongst and between the sub-cores and the core developed. This took place at a higher level of conceptualisation than was the case in open coding. Selective coding continued until no new concepts and properties emerged. Glaser (1978) refers to this point as theoretical saturation. In this research, the theoretical saturation point was reached after analysis of the tenth interview transcript.

When theoretical saturation was reached, selective coding evolved into the next and final coding phase, which is *theoretical coding*. In theoretical coding, the relationships between concepts are developed in terms of theoretical coding families, as expounded by Glaser (1978, pp.73-81 and 1998, pp.170-175). Glaser (1978, p.72) describes theoretical coding as follows: "… the theoretical codes conceptualise how the substantive codes may relate to each other as hypotheses to be integrated into a theory. …. they weave the fractured story back together again". The selection of a coding family amongst Glaser's (1978) 18 coding families clearly emerged as coding family number two, called "process" (p.74). This process also reflects a causal-consequence model.

With process being so salient in the data, how can it be structured and analysed, according to grounded theory? Glaser (1978, p. 74) provides the answer: "A process must have at least two stages. The analyst cannot talk of process and not have at least two stages". Therefore, to use Charmaz's (1995, p.661) wording: "I provide a stage analysis" of how people with CMT manage the symptoms and the effects of the disease in order to optimise their well-being.

The third part of analysing the data focuses on data gathering for triangulation and will be discussed next.

PART 3: TRIANGULATION

According to Neuman (2000, p.124), triangulation basically means that "it is better to look at something from several angles than to look at it in only one way". In order to examine the qualitative findings of this study from a different perspective, in this case the quantitative paradigm, a battery of self-report psychometric and other questionnaires was completed by all the participants. All questionnaires were scored by myself according to the prescribed scoring guidelines, and were verified by a registered psychometrist. In this section, the questionnaires will be introduced, whereas their results will be discussed in the next chapter.

Biographical data questionnaire

As the name clearly indicates, the aim of this questionnaire is to obtain biographical information from the participants. The questionnaire was compiled by myself and can be viewed in **Appendix A.**

Psychometric questionnaires

All participants completed a battery of self-report questionnaires considered applicable for triangulation purposes.

Carver Brief Cope

The Brief Cope (Carver, 1997) is a multi dimensional, self-report inventory that assesses people's coping strategies. The inventory consists of 28 items, to which participants respond on a 4- point Likert scale, with options ranging from one (I have not been doing this at all) to four (I have been doing this a lot). The scale was derived from the full length Cope (Carver et al., 1989) because that instrument with its 60 items was rather time-consuming to complete in certain contexts. There was also item redundancy (Carver, 1997).

The Brief Cope is considerably shorter than the original 60-item instrument. It consists of 14 scales of two items each, adding up to 28 items. From a practical viewpoint, the 14 scales may be regarded as "coping strategies". These are as follows: Active coping, Planning, Positive reframing, Acceptance, Humour, Religion, Utilising emotional support, Using instrumental support, Self-distraction, Denial, Venting, Substance use, Behavioural disengagement and Self-blame.

Like the Cope, The Brief Cope has two formats: dispositional and situational. Actually, the exact same items are used in both formats; it is only the wording that changes in order to alter the frame of reference. The dispositional items reflect a more stable and characteristic way of coping, for example "I criticise myself ". The retrospective situational format, adopted in this study, is more specific. The items are worded in terms of what the person did in a specific time period or situation, for example, "I have been criticising myself". The period decided upon was the three months prior to the study.

Internal consistency as measured by Crohnbach's alpha coefficient for all 14 scales, averaged across three administrations of the brief Cope in a particular sample, exceeded the minimum acceptable level of 0, 50.

WHOQOL – BREF

The full title of this questionnaire, rarely encountered in the literature, is the World Health Organisation Quality of Life Assessment - Abbreviated Version (or

Short Form). The scale is an abridged version of the hundred-item WHOQOL – 100, which was found to be too lengthy by some. The Bref, which measures quality of life, is a fairly new instrument that is used in various applications. An example is health services, especially health outcomes, where the effect of medical treatment on the quality of life of patients is assessed. There are 26 questions in total. Twenty-four are organised into four domains, or facets, of quality of life, namely physical health (PH), psychological (PS), social relations (SR) and environmental (EN). Each question is rated on a five-point Likert scale and refers to the two week period prior to completion of the test. The scores of each domain are transferred to a 100-point scale by means of the provided code. There is one question that measures overall, or a global, quality of life and another one that assesses global satisfaction with health (Taylor et al., 2004).

The instrument was chosen above the Medical Outcome Study Short Form -36 scale (SF-36), which is a very popular instrument for the assessment of health related quality of life, for a number of reasons:

1. Unlike the SF-36, and as far as can be established, the WHOQOL-BREF had never been used in research on CMT.
2. Both the full WHOQOL – 100 and the WHOQOL-BREF were developed in conjunction with more than 15 culturally diverse countries worldwide. South Africa is not on the list, but Zimbabwe is. The scales are therefore cross- cultural (Skevington, 1999).
3. Huang et al. (2006) found, in a large Taiwanese sample of 11,440 people, that the WHOQOL-BREF measures global quality of life (including health related quality of life), whilst the SF-36 measures health related quality of life.
4. The WHOQOL-BREF includes a domain that assesses the impact of environmental conditions, for example safety, financial means and access to health services, on quality of life. These factors were considered relevant to a developing country like South Africa.

The WHOQOL-BREF's psychometric properties had been established in rheumatoid arthritis by Taylor et al. (2004) and were found to be adequate. Test-

retest reliability was between 0, 71 and 0, 91. Chronbach's alpha coefficient was between 0, 64 and 0, 87, although the social relations domain did not meet the requirement. However, in a large scale Taiwanese study (N= 11,440) Huang et al. (2006) found that all the scales met the requirement. Regarding validity, "correlation with other measures of quality of life was supportive of concurrent validity" (Taylor et al., 2004, p.350).

BECK Depression Inventory (BDI)

This widely used questionnaire was developed in 1961 and has been revised and developed into different forms. The original format was used in this study. As the name indicates, the BDI measures subjective depression severity, or, more precisely, severity of symptoms and manifestations of depression. It is a self-report questionnaire, comprising 21 questions, each scored from zero to three. Internal consistency is high (Chronbach's alpha = 0, 86), with acceptable measures on various types of validity (Devilly, 2005).

The Perceived Stress Scale (PSS)

The PSS measures to what degree people appraise or perceive situations and general dynamics in their lives as stressful, specifically during the past month. Global stress, rather than specific stressors, and perceived stress rather than objective stress, is assessed. There are 14, 10 and 4 item versions, but the 10-item version was used because it has the best psychometric properties (Chronbach's alpha = 0.78) and is the one recommended by the developers. Each of the ten items is marked on a scale of 0 to 4 (Cohen, Kamarck & Mermelstein, 1983; Cohen & Williamson, 1988).

Rosenberg Self-Esteem scale (RSE)

Rosenberg, in 1965, described self-esteem essentially as being a favourable or unfavourable attitude towards the self. It refers to a person's sense of his or her value or worth and to what extent the person likes himself or herself. It is once again a short questionnaire, consisting of only 10 items. Six are positively and four negatively formulated regarding how the individual views him or herself.

Each question is rated on a four-point scale. (Martín-Albo, Núñez, Navarro & Grijalvo, 2007). In a study across 53 nations (N=16,998) Schmitt and Allik (2005) established Chronbach's alpha for the Rosenberg Self-Esteem scale as 0, 81.

The way in which the psychometric results relate to the qualitative grounded theory data, will be discussed later in the next chapter (results). As already pointed out, applicable data from the biographical questionnaires was included in the grounded theory and analysis.

CHAPTER 4

RESULTS

In this chapter, the results that emerged from this investigation will be presented. The prime focus of this study was to generate a grounded theory regarding the strategies that people with CMT employed to deal with the disease's manifestations with a view of optimal adaptation and well-being. This aspect will be the core of the chapter and constitutes Part one. For the purposes of triangulation, a battery of psychometric questionnaires was administered, and a few other verification techniques were implemented. These verification strategies will be presented in Part two.

PART ONE
THE GROUNDED THEORY

OVERVIEW

As was discussed in Chapter 3, the theoretical code that fits my study's data emerged as coding family number two, namely "process" (Glaser, 1978, p. 74) According to Glaser (1978) a process reflects movement; "something happening over time" (p.75) and must always (a requirement) consist of at least two stages or phases. A process may also embrace a "causal-consequence" model (p.75), which is applicable to this study. Therefore, the grounded theory model depicted in Figure 4.1, which is a summary of my results and that will be delineated in this chapter, may be described as a "cause - consequence - process" model.

In order to observe the emerging grounded theory of how people with CMT manage their condition and in so doing maximise their well-being, Glaser's grounded theory methodology guided me to ask the following two fundamental questions: 1) What is the main concern, or core problem, that people with CMT face, and 2) what are they doing to resolve it? The latter does not mean how they solve it according to some external, objective criterion of success; instead, it refers

to how they process the problem, what action they take, or both (Glaser, 1978; 1992).

The analysis of the data revealed a broad spectrum of problem related CMT experiences that culminated in the main concern or problem, which is *dealing with unpredictable disease manifestations.* This problem presented a challenge to CMT-affected people; their task was to constructively manage the effects of the disease, including the unpredictability component, with the aim of achieving an acceptable outcome. This may be enhanced well-being or some other outcome variable(s) as determined by the data.

The resolution side of the equation, namely the action processes that, put rather plaintively, encompass what the people were doing about the problems they face, constitute the focus of the grounded theory. In this study, the way in which participants resolved the main problem, that is to say the strategies they used to manage the rather unpredictable disease manifestations, emerged as the basic social and psychological process (BSPP) of ***engaging with CMT***. Engaging with CMT also manifested as the core category, or rather the core process, of the grounded theory. Engaging with CMT comprised three interdependent sub processes, which overlapped to some extent. These were *orientating, fighting back and optimising.* These three processes represented three mostly sequential stages that people with CMT pass through as they endeavour to, from their frame of reference, deal effectively with the symptoms, effects and consequences of the disease in order to maximise their well-being.

A diagrammatic representation of the grounded theory model that emerged from the data is depicted in **Figure 4 .1.** The format of this chapter, as well as the elucidations contained in it, will be based on this grounded theory model of *engaging with CMT.*

The delineation of the three sub processes, *orientating, fighting back and optimising*, will be the sum and substance of the chapter. This includes the distinctive features of each, the relationships between them and the structural

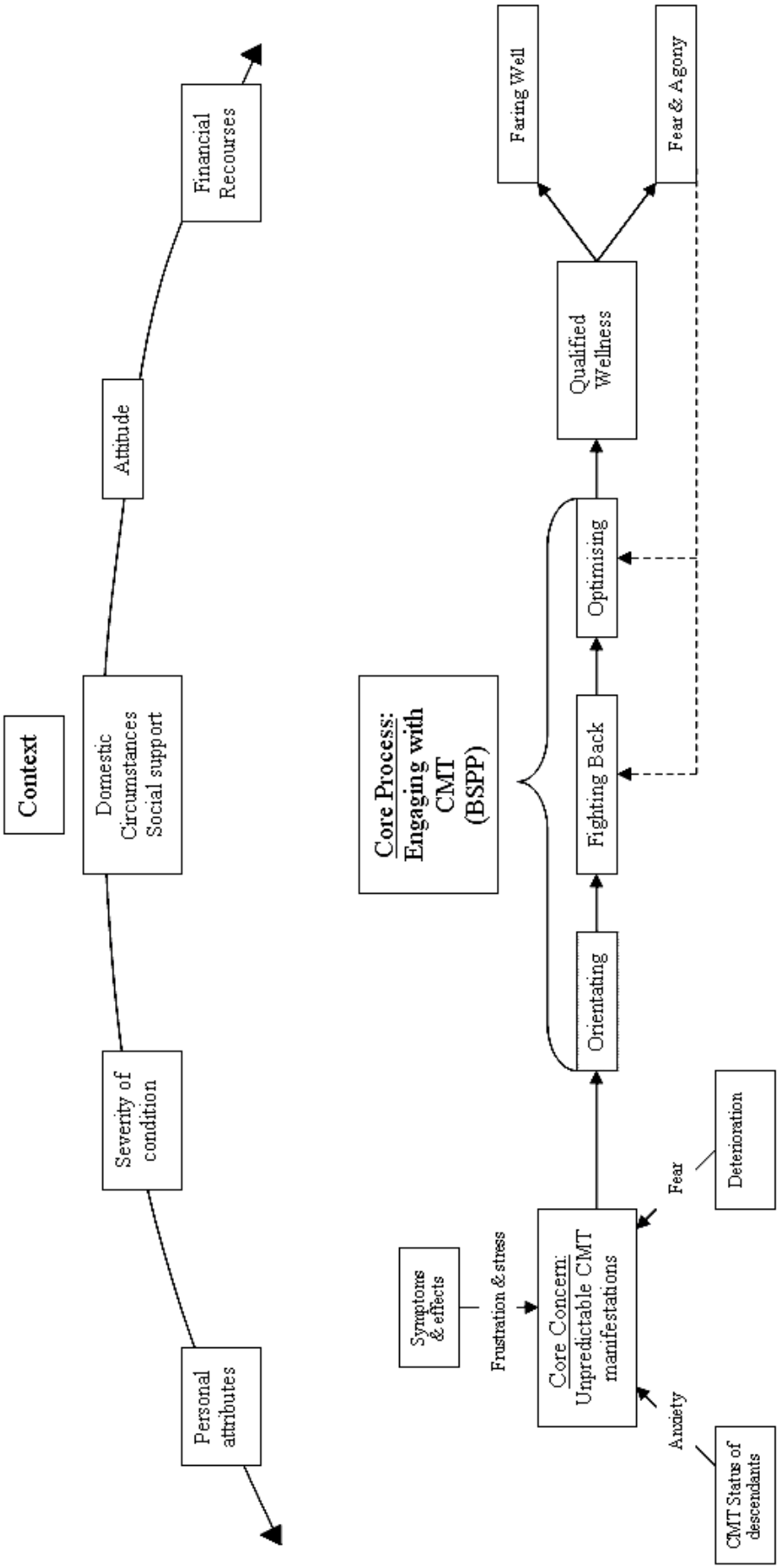

Figure 4.1: A grounded theory model of the process of engaging with CMT, depicting the resolution of the main concern, unpredictable disease manifestations.

conditions that affect them. The discussion will be substantiated and illustrated using ample quotations from participants. Whereas the aim was to use verbatim quotations, it was unavoidable to edit certain responses for the sake of clarity. This was restricted to an absolute minimum and great care was taken to ensure that the meaning of the quotations remained intact. In cases where the interviews were conducted in Afrikaans, quotations will be given in Afrikaans, followed by direct English translations. This is deemed necessary to ensure authenticity and credibility of the findings.

THE CORE CONCERN

As can be seen in Figure 4.1, the core concern, *how to deal with unpredictable CMT manifestations,* comprised three main sub codes. These were: (1) how to deal with the disease's symptoms and effects, (2) concerns about how far and how fast deterioration, or atrophying, will proceed and (3) concerns about the CMT status of the affected person's descendants. The main problems (1 to 3) constitute an integral part of the entire discussion because they influence so many processes. For the sake of clarity, they will nevertheless be briefly discussed individually as well.

Data analysis revealed a myriad of symptoms, effects, fears, threats and challenges that people with CMT had to contend with. Many affected people, for example, have balance problems and walking difficulties almost on a daily basis, whilst other symptoms, for instance dropping items when it is cold, manifested primarily during winter. Fears and anxieties tended to be more strongly related to long-term concerns, although certainly not exclusively so. The core concern of unpredictable CMT manifestations may be conceptualised as representing a challenge to participants. Analysis revealed at least two main challenges contained in the core concern and its three sub codes (symptoms and effects, deterioration and CMT status of descendants). These were: 1) how to deal with the symptoms and more practical disease manifestations on the one hand, and 2) how to cope with the unpredictable course of the disease and certain hereditary issues on the other (refer to Figure 4.1).

The more operationally inclined problems that the CMT-affected faced encompassed more or less the entire spectrum of CMT symptoms and effects that were delineated in Chapters 1 and 2, as well as one or two rare additions. A random list of these symptoms and effects, as reported by participants, is as follows:

- Finding suitable shoes
- Weakness
- Fatigue
- Poor running & performance in sport activities
- Getting weaker and weaker (atrophying)
- Walking difficulties
- Inheritance concerns (own children)
- Poor balance
- Self-esteem & identity issues
- Frustration
- Ignorance about CMT
- Lack of understanding (others)
- Falls to the ground
- Stigmatising & social effects
- Effects of cold weather
- Muscle cramps
- Walking barefoot
- Sprained ankles
- Lack of, or distorted, sensation
- Pain
- Inability to walk on heels
- Poor handwriting
- Work related problems
- Climbing stairs
- Having good days & bad days
- Driving a manual car
- Taking a shower
- Cooking & doing washing
- Low back pain
- High foot arches
- Hammertoes
- Foot surgery
- Chilblained hands
- Hearing difficulties
- Beginning to walk late as a child
- Vision Problems
- Consanguinity
- Asymmetry – right worse than left

Rare or unique effects included asymmetry (the right hand side of the body being more affected than the left), foot bones "collapsing" to the extent that severe bruising (haemorrhage) results, and blindness. Interesting effects included fear of taking a shower and taking a bath instead, and a marked increase in muscle cramping during the change of seasons. Although not an effect of CMT as such, consanguinity was nevertheless a unique problem, which had to be dealt with where applicable.

A major threat that people with CMT had to deal with was the uncertainty as to how long they will be able to maintain their current levels of physical functioning before deteriorating further. Equally so was their concern as to whether they had passed the CMT gene to their children. None of the participants in this study was at ease about these matters and fear and anxiety abounded.

To formulate a grounded theory concerns the formulation of a theory about the action or interactional strategies that people employ to resolve their main concern. The rest of this chapter will now focus on this facet.

THE CORE PROCESS: ENGAGING WITH CMT

Engaging with CMT emerged as the core category/process of the research. The following factors played a role in identifying the core category: (1) pervasiveness - it is a fundamental basic process that occurs, and accounts for change over time, irrespective of place, and (2) centrality - most other categories and their properties are related to it, and the core category also accounts for a substantial proportion of the variation in behaviour Glaser (1978). *Engaging with CMT* manifested as a complex, multi dimensional process comprising several interrelated sub-processes and strategies that ranged from reactive adaptation to the disease through countering its effects to the maximising of abilities and quality of life.

Engaging with CMT represented the way in which affected people coped with the manifestations of the disease, as well as how they managed the short and long-term implications of having it. To a lesser or larger extent, engaging the disease is encountered in all the stages of the core process. All participants, irrespective of

the influence of various structural conditions such as type and severity of CMT, age of diagnosis, sex, age, marital status, personal attributes and so forth, engaged the disease in one way or the other.

As the name partially implies, an element of combat and fighting one's way constitute an important component of the core process, although not necessarily the most important one in each of the stages. In the context of fighting, it involved a conscious decision by CMT-affected people to do everything in their power to keep standing and to fight the threats resulting from the disease for as long as humanly possible. This fighting spirit is aptly illustrated by a female participant in her early forties:

Ek dink my wil is net heeltemal te sterk om in te gee daaraan – ek het te veel go in my en ek is eerlik met jou, ek kan my nie in die toekoms sien agteroor sit met hierdie siekte nie.
I think that my will is much too strong to give in to it - I have too much go in me and I am honest, I cannot see myself in future sitting back with this disease.

Another older person verbalised it as follows:

Ek weier om op te gee tot ek nie meer kan nie. Die ou mense het gesê daar is nie 'n ding soos kannie. Nou, dit sal seker kom later (om afhanklik te raak van andere), maar ek vat dit dag vir dag.
I refuse to give in until I cannot go on any more. In the past, the old people used to say that there is no such thing as "cannot". It may perhaps come later (becoming dependent on others), but I take it one day at a time.

Engaging with CMT encompassed infinitely more than fighting and struggling. It comprised three rather distinct processes or categories, each with a number of sub strategies. The three emerged processes, namely *orientating, fighting back* and *optimising*, represented three sequential stages that people with CMT passed through as they manage their condition in order to optimally adapt and to obtain as good an outcome as possible (refer to Figure 4.1). In stage one, *orientating*, people affected by CMT acquainted themselves with the disease and its implications, both pre- and post-diagnostically. Stage two, *fighting back*, encompassed goal orientated efforts to counter and/or minimise the effects of the

disease, including dealing with future concerns. The name of stage three, *optimising* is almost self-explanatory, the essence being to optimise, or maximise well-being by way of various strategies.

The length of time that a person spent in a stage was variable and the transition point between the stages "blurry", to use Glaser's, (1978, p.100) terminology. Furthermore, those people who are affected may return to the earlier stages for any length of time and apply relevant strategies typical of that stage when important changes in their trajectory occur, as were typical in cases of profound deterioration. Both the speed of movement through the stages and the time spent in a stage were variable due to the impact of a myriad of structural conditions. Put differently, passage through the stages was enhanced or hampered, or at least mediated by, these intrapsychic and environmental (contextual) conditions.

The most important contextual variables that influenced the process of *engaging with CMT* are depicted in **Figure 4.1**. Personal attributes and attitude influenced the entire process of *engaging with CMT*, particularly by being background variables that played a role and mediated many action processes. The role of personal attributes is aptly illustrated in the following interview extract:

Ek dink ek is inherent in elk geval 'n fighter. Een ding wat ek geleer het toe ek vir daai organisasie vir gestremdes gewerk het; as iemand as volwassene gestrem word en hy was 'n go-getter, dan bly hy'n go-getter.
I think that I am inherently a fighter anyway. I learned one thing while I was working for that organisation for the disabled; when somebody becomes disabled as an adult and he was a go-getter beforehand, he remains a go-getter.

The severity of CMT also influenced the core process. The accents during the different stages of *engaging with CMT* were, for instance, different for seriously impaired individuals than for those with light symptoms. The availability and nature of social support was very important and helped or hindered people's passing through the stages. The role of financial resources varied between people. Issues such as being able to afford medical insurance, the nature of the benefits of

a medical aid and being able to make provision for the future, exerted influence on the core process and its sub-cores.

The four stages and the relationships between them, as well as the structural variables that affected them, will now be delineated. This detailed discussion will reflect the substantive grounded theory, summarised in Figure 4.1, as to how people with CMT manage the disease and its consequences in order to achieve a desired outcome. Copious use will be made of direct quotations, not only to provide the necessary evidence, but also to illustrate and clarify the constructs under discussion.

STAGE 1: ORIENTATING

Orientating usually commenced at any stage during childhood and continued for varying lengths of time through and beyond the crucial diagnostic phase, more or less until participants reached the stage where they had a good basic idea of what the disease encompassed and grasped the implications of "what they were in for". The process of orientating to CMT comprised three sub processes: dealing with pre-diagnostic manifestations, digesting the diagnosis and investigating the basics.

Dealing with pre-diagnostic manifestations and effects

The duration of the pre-diagnostic phase largely depended on the average age at which people were diagnosed with CMT. In this study, the average age was 29, with only three participants receiving a diagnosis before age 27 (at age 16, 18 and 26). It is therefore clear that affected people grappled for many years with the symptoms and effects of CMT without knowing what is wrong with them.

The effects of the disease, and most noticeably physical weakness in all its forms, usually manifested in various ways since very early childhood, for example sprained ankles, falls to the ground, and poor coordination. This resulted in considerable confusion and wrong attributions as to what was wrong. The action that parents and the medical fraternity took with the best intentions to help often

resulted in wrong, uncomfortable or even debilitating corrective measures being taken. One male recollected:

They operated on some bones, but they were barking up the wrong tree. They took a piece out of my heel to correct my foot that bent inwards, but they were wrong. Later, he continued: *I know one specific time my parents got shoes moulded to my feet, since it was difficult finding shoes. It was ridiculous. It looked like – the guys at school called them Noddy clogs. Then I got ripped off a lot and I went back to what I was wearing before.*

Generally, the physical dimension of the schools system presented almost insurmountable problems for the CMT-affected children. Teachers subtly discriminated against them by not entrusting certain tasks to them. Furthermore, the focus on sport and other physical activities were a nightmare for CMT-children. Without exception, these children were hopeless in sport and gymnastics. They were physically weak, had poor balance and could not run fast, yet more often than not were forced by teachers to participate in these activities. Although teachers might have been uninformed on CMT, this forcing of children into public activities where they experienced humiliation in front of peers and significant others nevertheless resulted in damage to their self-concept. The fact that the children did not know why they were so weak, caused them considerable stress and resulted in the processing of these dynamics with confusion and uncertainty. In many cases, this resulted in self-blame and aggravated the self-degradation. These dynamics were aptly illustrated by the following two individuals:

1. Subconsciously, your teachers don't pick you for certain things and don't ask you to do certain errands. You find that other children walk around with (inaudible) and you were not given that opportunity. It took me a long while to come out from all that. I had so many issues unaware of, that is perhaps why I did people skills. I could not do athletics. I hated it. This whole gymnastic thing, balance beams, you had to do it, and you were not given a choice. Also when growing up I was not given the opportunity to come out on my intelligence side. I was just an average pupil, not athletic at all. It had impacted greatly. Like I said, .I fell so often, there was not a week that I did not fall.

2. Dis hoekom hierdie goed vir my by die skole moet uitkom. Want jy is so afgekraak. Hulle het in daai dae nie geweet van ‘n selfbeeld nie, maar dis eintlik wat dit is. Hulle het jou selfbeeld totaal afgebreek. Hulle het altyd gesê jy is te sleg of jy wil nie saamwerk nie. Jy het so hard probeer en jy kan nie.

This is why these matters should get back to the schools. One was really diminished. In those days, they did not know about a self-image, but that is what it was. They totally broke down your self-image. They used to say that one is weak or did not want to cooperate. You tried so hard, but just could not manage.

In many cases, the unfairness, powerlessness and frustration that they experienced as children were remembered by CMT-affected adults with anger and bitterness, as was illustrated by the following two people:

1. ..... it was teachers who were relentless in their quest to force me into sport and physical activities, even though it must have been clear to them that I couldn't do it. Whether they were cruel or ignorant I still don't know.

2. Jy kan nie daai pen vashou nie. As jy ‘n tyd lank geskryf het, word die skrif al hoe leliker. Dan word daar vir jou gesê jy word al hoe luier. Jy het nie meer lus om te konsentreer nie, jy wil nie hierdie werk doen nie. Hulle het my oor die kneukels geslaan met ‘n liniaal. Dan kon jy dit nog minder doen. Veral in die winter.

You cannot grasp the pen properly. After a while, your handwriting deteriorates. Then they tell you that you are getting lazier. You do not want to concentrate any longer; you do not want to do the work. They hit me over the knuckles with a ruler- then you were even less able to do it. Especially in winter.

One way in which CMT-affected children coped with their weakness and other symptoms, was by compensating and by doing their utmost to comply with standards. They would typically train very hard to compensate or try to gain strength by exercising particular muscles, the former is illustrated by the following interview snippet:

I played rugby and I trained twice as hard as everyone else just to be good enough and strong enough and for my legs to be able to do it. I couldn’t run at all. I was totally useless.

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Compensating also took on other forms, for instance boys wearing long pants as far as possible because of feelings of inferiority about their thin legs. Affected children also learned to show a bold front to the world whilst hurting inside:

Dis waar ek die kuns ontwikkel het van buitekant smile jy, maar binnekant is jy fyn en flenters. Behalwe dat jy jouself bedrieg, doen jy jouself skade aan. Jy sit al meer van ‘n pose op.
That is where I developed the art of smiling on the outside but inside you are in pieces. Despite the fact that you are deceiving yourself, you are causing damage to yourself. You put up more and more of a pose.

The confusion and helplessness of the primary school years never totally receded, but as the children grew older, cognitive and other strategies of a more pro- active nature became more prevalent. This is illustrated by the following verbalisation, where indications of humour can also be detected:

But my main strategy was to compensate by studying and excelling academically. Not only did this work with teachers, but it even won me some popularity with the jocks of the school, in the sense that they were regularly begging me for my homework in order to copy it! (Laughs).

CMT impacts upon many individuals in a dichotomous way: one's feet and hands may be weak whilst the other body muscles remain strong. This phenomenon benefited some children who, for instance, did well in shot put but could not run fast. Others, although by far the minority, were fortunate to end up in a school where little pressure was put on them to compete in sport.

Social support from peers played an important role in the affected child’s well-being. In many cases, children with CMT were left alone by peers, that is to say, they were treated rather neutrally and were not particularly rejected or ill treated. In other instances, peers accommodated them and even helped them where they were weak. Where stigmatising and mocking by peers were present amongst participants, it occurred more in boys than in girls. The following man experienced both the positive and negative as a child:

Strangely enough, other children accepted me just fine and I don't recall them being nasty to me at primary school level. In high school, as a teenager, peers did mock me from time to time, especially my walking.

Many of these childhood and adolescent issues and strategies carried over into pre-diagnostic adult life, although greater cognitive maturity enabled better processing of the dilemmas faced. In terms of experiencing a reduced self as a consequence of CMT, or at least as a major threat to the self, the following quotation from a young married woman is applicable:

It (CMT) has caused a bit of bad feelings between my husband and me initially. Up until that point (before diagnosis), I thought it was something about me that was not right, but I did not know what was causing it. That had caused somewhat of a sour taste, I always felt that I was not good enough.

Naturally, the roles and responsibilities of adult life presented many new problems and challenges. Even a straightforward task like hanging washing presented problems to this woman:

In my 20 er jare het ek begin simpel goed agterkom – as ek bv. wasgoed ophang kan ek nie die pennetjie druk en hom indruk nie – ek het net nie die krag daarvoor gehad om dit albei te doen nie.
During my twenties, I began to notice simple things- when I hang the washing, I cannot press the peg to open it and press it home at the same time. I didn't have the strength to do both.

Working life in particular presented numerous problems, as is indicated by the following interview text:

I worked in a job that required meticulous and neat writing. At one stage, I experienced severe cramps in my hands. I had to do all my written work with a scriber and not by free hand.

As the years went by, more and more roles and responsibilities were disrupted by CMT, although the people themselves had not been diagnosed yet and were

therefore not able to put a label on what it was that affected them. Marriage, having children, household chores, promotion or increased responsibilities at work and more demanding social responsibilities were all impacted on negatively by the disease, although in varying degrees. Driving a car with manual transmission, for instance, became increasingly difficult. Gradually, the effects of the disease simply became too much for affected people to live with. Although they had previously consulted doctors with their symptoms, they now engaged the medical fraternity with the specific aim to find out what was wrong with them. The crucial step of diagnosis had begun.

Assimilating the diagnosis

The sub-process of assimilating the diagnosis entailed the ways in which CMT-affected people dealt with and digested the entire diagnostic process, the essence being how they reacted to the finding as such. Most participants (75%) who sought medical attention that eventually resulted in diagnosis, did so during their adult years. They took this step because their symptoms and conditions such as atrophying, as well as new manifestations caused profound disruption, discomfort and suffering. Two people were diagnosed in their high school years and in these instances, the process was initiated by parents.

Without exception, diagnosis was a long and uphill struggle, encompassing multiple referrals and in many cases wrong or irrelevant medical procedures. Ignorance about CMT led to affected people being sent from pillar to post, before ending up at the neurologist who made the diagnosis. More often than not, the first specialist that the general practitioner referred them to was an orthopaedic surgeon. A woman explains how a specialist was barking up the wrong tree due to ignorance about CMT:

Toe gaan ek na die ortopeet toe en hy sê hy dink dis carpal tunnel sindroom en hy gaan dit opereer. Toe op die operasietafel sê hy vir narkotiseur dis die eerste keer in sy lewe dat hy sien dat hier nie ‘n spier hier is nie. Toe sê hy gaan nou maar vir my die spier opbou en sien hoe dit gaan. Met ‘n opvolg besoek sê ek vir hom dis nog dieselfde – die goed val nog steeds uit my hande uit en ek kan dit nie optel nie – daar is geen verbetering nie. Toe stuur hy vir my na Dr ....by ..... hospitaal.

I went to an orthopaedic surgeon who thought it was carpal tunnel syndrome and decided to operate. Then, on the operating table, I heard him say to the anaesthetist that it is the first time in his life that he sees that there is no muscle there, and that he's going to build up the muscle and see how it goes. With a follow-up visit, I told him that there is no improvement, I am still dropping things and I cannot pick them up. He then referred me to dr..... at.....hospital.

Most people experienced the diagnostic process as unpleasant, uncomfortable and painful. The way in which diagnosis was handled, the perceived competence and sensitivity of the neurologist and the quality of CMT information supplied were very important conditions that either helped or hindered coping with the diagnosis. When handled insensitively, frustration and agony were the result, as is recollected by the following person:

My first test was a nightmare. They never prepared me. My feet were as cold as ice. Half past seven in the middle of winter. She told me afterwards they should have warmed my feet. I had no clue what was going on. I only heard the terrible scratching noise from the machines. In the meantime, this guy leaned forward over the bed and kept saying: " do it again, it is very interesting ". In the meantime, I am lying on the bed crying because it hurt so much. When it hurt really badly, I shouted at him.

The CMT-affected reacted to the diagnosis in divergent ways. There were those who experienced relief. For them, having lived with the symptoms for as long as they can remember but without really knowing what was going on, diagnosis brought dramatic clarification and in a way solved a life puzzle. One man described his experience in this regard as follows:

Receiving a diagnosis of CMT was not experienced by me as a traumatic, negative event. Up to that stage, I had experienced a lot of problems for being so weak and not knowing why. Now I knew that it was not my fault at all, that all the time I have suffered from a medical disease! It made a major difference to my self-respect and my way of thinking.

Being diagnosed dramatically influenced the lives of people with CMT in direct and indirect ways. Many adjusted their lifestyle and changed their outlook, as

well as the practical ways of doing things. By way of example, the experiences of a female in this regard:

After that (diagnoses) I knew what was troubling me and I changed my whole outlook, e.g. my walking was not very hasty, trying to do things at a much slower pace. Now I knew what was causing it, instead of rushing, I was more aware now that there was a step and that I must watch and look around.

Not everybody reacted positive, or even neutral, to the diagnosis. There were also those who experienced it as traumatic. Shock, despair and anger reflect typical debilitating ways in which these people processed the diagnosis. The following female was raised under the fallacy that women cannot acquire CMT because they are only carriers of the gene:

Ek is toe in 'n toestand - ek huil al die pad en wag vir die verslag. Gelukkig my verloofde – toe is ons nog nie getroud nie, ah... – ek huil al die pad en ek huil. Ek kom by die werk en ek huil, want ek sien myself ek gaan in 'n rolstoel sit..... Toe is ek kwaad vir my pa. Ek is verskriklik kwaad. Toe dog ek hoekom, hoekom?
I was in a state- I am crying all the way while I'm waiting for the report. Fortunately, my fiancé-we were not married yet, ah...- I cried and cried all the way. I arrived at work while still crying, because I see myself sitting in a wheelchair. Then I became angry with my father. Very angry. I thought why, why?

The age at which the person was diagnosed, as a structural condition, influenced the reaction to and assimilation of the diagnosis. Young people and children, for example, did not necessarily grasp the full implications, as can be seen in the following response (social support also buffered the effect of the message):

Honestly, I was still young at that time and didn't understand much of it (diagnosis). So I didn't worry much about it. My father went with me.

Irrespective of how the diagnosis was experienced, be it neutral, as relief or as devastation, it did not take long for the recently diagnosed CMT-affected to become alarmed and anxious as to what the future holds. They were uncertain of the implications of having this disease, which was exacerbated by the inadequate

information given to them at the time of diagnosis. This gave rise to the last sub-process of *orientating*, namely investigating the basics.

Investigating the basics

This sub-process amounted to the gathering of information as a way to gain some degree of control over future uncertainty. The hunt for information by the recently CMT- diagnosed were either fruitful, or frustrating, depending on factors such as how skilful they approached the matter, their perseverance and the importance they allocated to it. The following two quotations with opposing outcomes illustrate:

1. Ek het toe na die Mediese Biblioteek gegaan en toe 'n paar boeke uitgeneem en gelees wat sê hulle daaroor. Deesdae is daar op die Internet redelik inligting beskikbaar daaroor.
I went to the medical library, took out some books and read up on it (CMT). These days, a fair amount of information on it is available on the Internet.
2. *Even today I think there is not enough information about it. I tried looking for stuff, but I stopped now.*

People recently diagnosed with CMT also embarked upon a venture to unravel their inheritance in order to track down the origins of their disease. In many instances (most in fact), this proved to be a tall order. Typically, parents and grandparents either were totally unaware that they had CMT, or deliberately denied/concealed that anything was wrong with them. In many cases, they had passed away. These dynamics were aptly illustrated by the following verbalisations. In the second one, the woman blamed her mother for denying her the opportunity to take action and make provisions because of the disease:

1. Afterwards, I endeavoured to establish whom I inherited it from. Information was scarce but it appeared to be from my mother's family. To me it is clear that my late mother was indeed lightly affected, but she was never diagnosed. My grandmother was in a wheelchair, although I could never confirm that this was due to CMT. My uncle, who had passed away, was definitely affected, according to sources. My brother and sister did not inherit CMT – only me.

2. Kyk my ma het dit ook. Sy sê dit is nie so nie, maar dit kom van my ma se kant af; definitief. My oupa .... (onhoorbaar). Hy het vate en goed gemaak. Toe sê hy 'n vat het op sy voete geval, maar dit is nie so nie. Die ou mense was skaam vir so iets. Tot vandag toe stry my ma dis nie hulle familie nie, maar dit is so; dit kom van hulle familie af. Jy weet, as hulle miskien vir jou kon sê, dan kon jy voorsorg gemaak het.
My mother also has it. She denies it, but it definitely comes from her side. My grandfather.... (inaudible) he made barrels and things like that. He said a barrel fell on his feet, but it is not so. The old-timers were shy about these things. My mother denies that it is her family up to this day, but it is indeed so, it comes from her family. You know, if they perhaps could have told you, you could have made provision.

In certain cases the search lead to unusual findings that resulted in even more questions, for instance in the following case of consanguinity:

My husband and I are first cousins. Our mothers are sisters. Our grannies are sisters. His mother and father were cousins. My mother's parents were cousins. Maybe that's why the gene is so strong in my case. Strange, my husband is not affected at all. He is a strong, big guy.

Largely, the above quotations indicate that affected people's efforts to establish the genetic path and origins of the disease rarely resulted in clear-cut answers. However, it served a very meaningful purpose in the context of being recently diagnosed with CMT. People were trying to get some control over their situation, in which, at that early stage, confusion and uncertainty abounded. In fact, both the two actions of *investigating the basics*, namely *gathering information* and *tracing one's own inheritance,* arose out of the anxiety and uncertainty that followed the diagnosis. Being such early, almost "preliminary" strategies, they did not go far enough in answering the many anxiety-provoking questions about the nature and course of the disease, which confronted the recently diagnosed.

Realising the salient implications

Above two strategies nevertheless did provide enough answers to enable CMT-affected people to weigh their options and decide on the way forward. They knew enough to realise that the disease was not necessarily totally incapacitating, even

though the possibility existed that it may have resulted in serious impairment, even dependency, in the long run. The turning point, or critical junction, that signalled the end of the stage of *orientating,* was termed *realising the salient implications*. From these early strategies, people who have CMT were also in a position to reflect upon the general way forward, which, in this study, emerged as the decision to fight back. The decision to engage the disease was also done from a platform of tentative acceptance of their lot, in other words, that they had inherited this incurable disease. Verbalisations from two interviews illustrate this critical junction:

1. The person must first accept, this is my lot and this is how I am going to go forward...
2. My research on the disease, and especially the excellent information from the second neurologist, left me with many concerns and worries: the main one being what the future holds. I knew enough by now to realise that there was a realistic chance of me reaching old age without becoming full-time wheelchair-bound. If I looked after myself, I should be able to survive, although it may be a considerable struggle and uphill battle.

The lapse of time since diagnoses to the critical junction of *grasping the salient implications* varied greatly between individuals. The change was a very gradual one and the decision to fight back was hardly a once off event, but instead manifested over time. When it did, they proceeded to the next sub process of the core process *engaging with CMT,* namely *fighting back.*

STAGE 2: FIGHTING BACK

The Paperback Oxford English Dictionary (2002, p. 304) defines fighting as: "struggle to overcome or prevent". Whereas this description goes a long way in encapsulating the essence of this category, fighting back emerged as a broader concept, which also included dimensions where the focus was subtly different from the mere struggle to overcome or prevent. An example here is where, due to anticipated increased weakness, provision was made for eventually moving into an old age home or similar complex where care was available.

Fighting back was a key strategy that CMT-affected people used to cope with the disease, as well as to manage their living with the disease on a continuous basis. It should be stressed however, that it would be over simplistic and in fact incorrect to say that it is the most important strategy, since the three main processes or stages of *engaging with CMT* carried the same weight. For instance, if people did not successfully move through the *orientating* phase, they would not have fully realised, in other words with insight, what was in store for them in the future, hence they would not have been able to engage with the disease and make the commitment to fight back. For many, combatting the effects and threats of CMT, including holding on to "normality" for as long as possible, was the prime focus for a long period. It would indeed be fair to say that some affected people became rather stuck in this stage for a long time and only gradually and with a degree of reluctance moved on to the next stage.

Fighting back encompassed both practical, day-to-day actions as well as more long-term strategies. Typically, the former had to do with the activities of daily living, in other words, what affected people did in order to handle the day-to-day activities in which the disease restricted them. The latter had more of a future focus and entailed dealing with the envisaged long-term effects and consequences of CMT, for instance becoming weaker and weaker due to atrophying.

People with CMT, having by now *orientated* themselves to the disease, gradually came to realise that CMT does not necessarily result in total dependency on others over the long-term. Their own research revealed this, or neurologists informed them accordingly at the time of diagnosis. The dilemma they faced, however, was that one might become disabled, or dependent, in various degrees, for example in one's ability to walk, drive a car, write and so forth. Furthermore, people do indeed become wheelchair-bound due to Peroneal Muscular Atrophy or CMT, albeit not regularly. Where will the disease take them? More specifically, how far, if at all, will their muscles atrophy? Can they influence the process? These were burning issues that they grappled with throughout the entire process of *engaging with CMT*.

The gradual decision to fight back evolved because these people realised intuitively that they had a fighting chance. Unlike certain progressive diseases where the outcome is frequently of a more severe nature, for example certain types of muscular dystrophy, the CMT-affected saw that the scenario was not necessarily as grim for them. If they persevered and worked hard to limit the negative implications as far as possible, if they took care of themselves, the possibility existed that they may survive without sacrificing too much subjective quality of life. There were four strategies in the stage of *fighting back,* namely *wrestling with worst-case scenarios, persevering, empowering themselves and accepting reality.*

Wrestling with worst- case scenarios

Worst case scenarios here refer to the worst possible outcomes of the two pervasive conditions that were introduced in the beginning of this chapter; those that had to do with the unpredictability component of the core concern. They were: (1) extreme concern by the CMT-affected that they may deteriorate to the point of becoming dependent on others, and (2) inheritance concerns, including anxiety whether their descendants have inherited CMT, extreme concern about their well-being if they did, and how to handle the possibility of future children. The focus here is how the CMT-affected dealt with these worst-case scenarios.

Except for the uncertainty component and the fact that both the above engendered considerable fear and anxiety, the two conditions were rather distinct. Deterioration is variable, that is to say, a person's condition may deteriorate in various degrees, from little to much. Furthermore, degrees of impairment and even the extent of dependency on others have a subjective component in addition to objective criteria. For instance, some people may consider themselves dependent if they use a wheelchair occasionally on shopping trips, whereas others feel this way once they cannot take care of their personal grooming any longer. The subjective understanding of certain terms such as "dependent" in this case, makes it much more difficult to communicate effectively with others.

Theoretically, worst-case scenario in the case of the CMT status of one's descendants is much more dichotomous - the children inherited CMT or they did not, although they could be more or less seriously affected than the parent who gave it to them. It could however take years for the symptoms to manifest, or to do so noticeably. Once the child is born, the focus of the parent shifts to managing his or her own thoughts, feelings and anxieties about the offspring's possible CMT status, as well as optimising his or her care for the child. The issue whether to have any more children emerged as a delicate matter.

Being a rather isolated, but nevertheless influential condition, inheritance concerns and the way in which they were handled, will be discussed first. This will be followed by the ways in which deterioration was dealt with. The latter connect with and flow more naturally into the rest of the chapter.

Dealing with CMT status of descendants

In general, a major anxiety provoking concern for affected parents who had children of their own, was the chronic uncertainty if they had passed the CMT gene on to the children or not. Anxiety was aggravated by the many uncertainties regarding inheritance in general. Most parents knew enough about CMT to realise that the onset of the different types of the disease vary. An ostensibly unaffected toddler, for instance, does not exclude the onset of the disease at a later stage. Most also knew that their offspring may be affected less (or more) severely than they themselves. The child therefore could be very mildly affected, with the possibility of parents not noticing or subconsciously minimising minor symptoms. Parents reported that their anxiety was of a chronic nature. It always loomed in the background, but, as will be shown, did not necessarily result in fruitful action to address it.

Parents on the whole were aware of, but did not possess adequate knowledge about, the existence of genetic testing in order to establish the presence of certain CMT types. Very few had investigated the matter properly and no children had actually been subjected to these tests up to the date of the interview. Rather

unexpectedly, one parent who intended to have it done, encountered ethical problems when he wanted to have his child genetically tested:

I wanted my kid to have the DNA test as well. There was a whole ethics problem (regarding the issue of subjecting a minor child to genetic testing). The professors were very pleased that I wasn't that type (type 1A, which can be readily detected with genetic testing) because they knew there was a fight coming up (to have the child tested even though he was a minor).

A more puzzling matter than the dynamics regarding the lesser known genetic testing was that very few parents had taken their children for the conventional neurological examinations and diagnosis. Less than half of the participants had actually taken this action, of which two took the children for diagnosis in late adolescence or early adulthood. Parents grappled with the trade off between embarking on the route to more certainty and the gloomy finality of a positive diagnosis, with all the implications it entailed. This included the elimination of hope that the child was unaffected, in other words, that the worst-case scenario would become a reality. This is not to say that parents with CMT had no insight into the advantages and disadvantages of diagnosis, as is reflected in the following mother's comments:

Voorverlede jaar wou ek al gegaan het, want ek wou vir xxxx laat toets het, dis nogal vir my, uhm.., soos ek sê ek gee nie om as hy dit het nie, maar ek wil voorbereid wees, wil weet wat vir my wag. Dan weet ek hoe om dit te hanteer. Die toetse is baie seer en ek wil hom nie onnodig daaraan blootstel nie. (Meeste deelnemers het die elektriese geleidingstoetse as pynlik ervaar. Dit behels die toedien van skokke, geleidelik toenemend in sterkte, aan ledemate, of aan die senuwees binne die ledemate, om presies te wees).

Two years ago, I intended to go because I wanted to have xxxx tested. To me it is rather, uhm..., as I said, it does not matter if he has it, but I want to be prepared, I want to know what awaits me. Then I know how to handle it. The tests hurt a lot and I do not want to expose him to it unnecessarily. (Most participants experienced the electric nerve conduction tests as rather painful. It involved administering electric shocks, which increased gradually in strength, to limbs, or to the nerves inside the limbs, to be precise).

At the time of the interview, the above-mentioned mother had still not taken the child for diagnosis.

The data therefore indicate that parents did not readily embark upon the route of diagnostic testing; instead, they chose to monitor their children themselves in the hope that they will not detect the presence of the disease. This strategy of *monitoring*, which generally stretched over many, many years, typically resulted in a never-ending cycle of detecting a possible CMT symptom, with accompanying panic and elevated anxiety, followed by relief when the symptom subjectively "proved" to be benign.

Keeping a watchful eye on children for indications of CMT were not necessarily a full- time, conscious focus, but instead more of a general orientation. In many cases however, the monitoring was meticulous. The uncertainty and monitoring even extended to the grandchildren. The concerns of a middle-aged mother/grandmother with CMT in this regard can be seen in the following interview text:

Want my dogter se jongste enetjie lyk baie goed; dink nie dat sy dit het nie. Maar die oudste enetjie wat nou standerd xxx toe gaan, het dit. My een seun, hy het dit nie. Glad nie. Maar nou ja, hy gaan nou trou en ek weet nie of dit gaan uitkom in sy kinders nie. Dis die ding.

My daughter's youngest one looks very good; I do not think she has it. The eldest one who goes to standard xxxx now, has it. My one son does not have it at all. However, he is now going to marry and I do not know if it will come out in his children. That's the thing.

When children of CMT affected parents reach adulthood and move into roles such as marriage and having children of their own, concern about their own CMT status typically escalated and became burning issues. It was very rare to find grown-up children being totally ignorant about their parent's CMT status and therefore the possibility that they might have inherited the gene, even though they might have been only lightly affected. Those who already had children of their own, grappled with decisions to have more. Under these circumstances, they typically put pressure on the parent, as was illustrated in the following instance:

The children are also rather honest in their analysis of the situation, which put further pressure on me. For example, my daughter talked to me about the probability of their offspring inheriting CMT because her husband is a very sport-orientated type of person who will be "broken" by having an affected child. What do you say?

Did parents know that they had CMT when they had their children? In most cases, the answer is yes, as reflected in the interview transcripts and background information forms. It was rare to find that affected parents were totally unaware of the possibility that they could be passing on the gene. Typically, they were unaware with the first children, but knew that they had CMT with later children.

The decision to have children in the context of CMT was, with very rare exception, discussed by the two partners and the decision to proceed was a joint one. In some instances, because they were aware of the possible inheritance implications, parents tried to ascertain the risks involved. Genetic counselling was an avenue they explored, although not always with satisfactory consequences. Others based their decision to have a child on knowledge that they acquired through reading or from what the neurologist told them. Some people may regard the following advice by a neurologist as controversial, whilst others may approve:

My Pa het daai dag toe ons daaroor praat vir my gesê ek moet nooit kinders hê nie – toe sê daai dokter vir my Pa as hy weet hoeveel oorerflike siektes daar is wat kinders kan kry, is ons s'n niks nie. Ek mag maar kinders hê. Hy sê toe vir my Pa hierdie siekte maak jou nie dood nie. Dit was sy woorde – dit gaan my nie doodmaak nie – die kanse is 50% dat my kind dit sal hê.
My father told me never to have any children. The doctor then told my dad if he just knew how many inherited diseases there are that children can get, ours is nothing. I may have children. He then told my dad that this disease will not kill you - these were his words - it will not kill me - the chances were 50% that my child would get it.

There were also parents who followed the strategy of urging their children not to have children of their own:

When I found out about it (that he has CMT), I talked to the children and advised them not to have any children, but they would not listen. My youngest daughter's child has it, the worst of all.

The matter of how parents felt in retrospect, that is to say, how they, having lived with CMT for such a long time, presently feel about having children, will be dealt with in the third stage of *engaging with CMT*, namely *optimising*. In *fighting back*, it was more about grappling with the risks involved in having CMT-affected children and what to do about their fear if children had inherited the disease. Although the aim was to get peace of mind by knowing, thereby reducing anxiety and possible guilt feelings, in many instances the overriding motive was to enhance the well-being of the children. Parents could allegedly take better care of the child by knowing his or her CMT status. The fifth quotation, counting back from this point, illustrates this. On the other hand, by not knowing, the child could be raised without the danger of stigmatisation.

What happened in instances where there was clarity about the positive CMT status of children, in other words where parents *knew for certain* that they had passed the gene on to the child? In these cases, intrapsychic strategies were the dominant action that parents employed. There is no known cure for CMT and direct action with a view of altering the problem situation was therefore not an option. What remained was cognitive and emotional processing of the situation, as well as activities to optimise their care for the child in all its facets.

One way in which optimising of childcare manifested, was very high levels of parental involvement in the lives of affected children. The involvement took on many forms, but the emotional aspect was at the heart of it, reflecting a level of empathy that in a way was only attainable in these unique conditions, where the "responsible" person was the parent. In addition to empathy, parents were actually hurting for their affected children; living with them on a daily basis caused considerable pain and guilt feelings. Just listen to the pain and agony of these two CMT-affected mothers:

1. Its hard to see your child struggling. I see people laugh at him, the way he walks, the big one. When you see that as a parent, it hurts a lot. At a shopping centre the other day youngsters were sniggering, because of the way he walks. That wants to make me cry.

How is he dealing with it? Sometimes I am just scared to ask him how he is dealing with it. He has never come to me. He does get tired. We can't go places where there is a lot of walking, because inevitably he gets tired. I cope much better than he does. He still comes to me in the morning to close up the top button of his shirt. He sukkels with it. (He struggles with it).

2. Jy weet, baie oggende dan kom ek by sy kamer in, dan sit hy op daai bed. Dan kan jy sien hy kan nie op nie, al wil hy, dan val hy terug. Dan sê hy los net so bietjie. Hy is ontsettend iesegrimmig, jy kan sien. Hy kruip toilet toe, maar sal nie vir hulp vra nie.

You know, many mornings when I enter his room he sits on that bed. Then you can see, he cannot get up, even if he wants to, then he falls back. Then he says just leave me for a while. He is very grumpy, you can see it. He crawls to the toilet, but will not ask for help.

By way of example, the first mother responded by being extra affectionate towards the CMT-affected child:

I feel most for the one who has it, because he can't do certain things. When I ask him to fetch a glass of water, he drops it all over the place.

Both the responses of agony and affection, including taking special care of the child, extended into adulthood, despite the CMT-affected child in some instances being married and having children of his or her own. In fact, the concern then extended to the grandchildren, in other words, anxiety whether they inherited the disease or not. This mother was still agonising about her married daughter with CMT, who already had children of her own:

You know, the bitterest part about CMT is that you want to do everything, but you cannot. Like my married daughter, she is affected so severely that she has a hunchback. She really struggles.

This particular mother also has a son who inherited CMT from her. Despite the fact that he is self-employed and independent, his mother is very protective of him and supports him emotionally. She has actually been so thorough that she has provided for alternative people to take care of him if she is no longer capable of doing it.

As an integral part of their torment, affected parents experienced considerable guilt feelings. Implicitly or explicitly, they felt responsible for having given their children this incurable, progressive neurological condition. Guilt feelings were present irrespective of whether parents were aware of their own CMT status at the time when the child was conceived or not. In other instances, parents blamed themselves for "mistakes" they were convinced they had made in the child's upbringing. They were aware of the disease's physiological basis and that it could only be inherited, but nevertheless felt that, somehow, their mistake altered or influenced the course of the disease. A mother's confusion and self-blame is illustrated by the following:

Sometimes I blame myself. He wasn't walking until he was two and half. When people asked and said he is almost three. I should have let him go through the crawling stage until three if he had to and walk at four if he had to. I still think if I didn't do that....I beat myself up about that sometimes.

Loving and caring for the affected child were not the only strategies: parents also tried to add value by trying to counter the effects of the disease in the child, thereby entering the process of *engaging with CMT* in the child's life, specifically the sub-strategies of *fighting back* and *optimising*! As the following comment illustrates, what exactly to do was not always clear:

Sometimes, I send my son for bio kinetics, because he is unstable, he can't stand. I hope it helps him in the long run. Can you tell me, how far should I push my son to exercise?

As can be seen from above exposition, most parents were still experiencing anxiety about the possibility of their children having inherited the disease at present, in other words at the time of the interview. This finding will be taken up at the end of the chapter where outcomes are discussed. Similarly, the strategies of intense involvement and care, despite reflecting CMT parent's best intentions, did not succeed in adequately reducing the pain, agony and stress of these parents. Perhaps nothing else could have been done. It is however not the task of the

grounded theorist to evaluate the effectiveness of people's resolutions, but instead to only focus on how they process and resolve their main concern (Glaser, 1998).

Managing Deterioration

Because deterioration in CMT tends to be a slow and gradual process over many years, affected people were not always focussing on it. Some remained on a plateau for years. Higher levels of awareness existed when the more profound (and regular) losses occurred, as well as in instances where individuals monitored their bodies more meticulously. In most cases however, as the years went by and affected people lived with the disease, the signs of deterioration became masked by the struggles of daily living, resulting in reduced awareness. Eventually however, a point was reached where either the atrophying body or the concomitant diminished physical abilities forced themselves into awareness, triggering alarm and anxiety. In other instances, it was a gradual process. These experiences are reflected in the following interview text:

When the kids came along, I did this, that and the other. I didn't deteriorate. Now that I become older, I can see that I am starting to waste away more. When I look at my hands I can see now that now that I am getting older, it's starting to waste away more. My calves are still the same. There is still nothing there.

The threat of atrophying so extensively that one becomes wheelchair bound or dependent on others was experienced as very, very anxiety provoking by all participants. Their responses were peppered with indications of this fear, of which the following is a typical response:

My future concerns?.....to lose any of the abilities that I have, and not to loose any (in-audible), for example personal grooming. I would not want any one else to take care of it. You can always ask others to buy food, or somebody can send you some, but your own grooming....what you do for yourself. I don't want to become dependent. General things, your bath, things you take for granted.

The profound disruption in their lives brought about by deterioration is dramatically illustrated by the following male:

Ek kom dit agter aan goed wat jy kon doen en nie meer kan doen nie. Met klein spierbewegingtjies sukkel ek al hoe meer mee. Ek het nou as kind leer klavier speel, maar ek kan dit nou nie meer doen nie. Jy kan nie jou vingers sit waar jy hulle wil sit nie, so kitaar speel is nie eers ‘n opsie op hierdie stadium nie. Maar ek loop deesdae al hoe swaarder. Ek het dit (agteruitgang) ook agtergekom met die muurbalspelery waar ek nie meer die rakket ordentlik kan vashou nie, ook met die skrywery waarmee dit al hoe moeiliker gegaan het en die voet wat vashaak omdat hy hang – sulke goed wat vir my sê hier is agteruitgang.

I notice it in things that I could do but cannot do any longer. I struggle more and more with small muscle movements. I learned to play the piano as a child, but I cannot do it any more. You really cannot put your fingers where you want to and playing the guitar is not an option at all at this stage. Walking is becoming increasingly difficult. I also noticed it in squash where I can no longer hold the racquet properly, also my writing, that is becoming increasingly difficult and my foot that is caught because it hangs - things like this indicate to me that there is deterioration.

In addition to the uncertainty regarding the extent of deterioration, the inconsistent rate and speed of the proceeds were equally problematic and disruptive, as, for instance, stated by the following person:

And after school, when I was diagnosed, it wasn’t that serious. I was still walking around normally and everything. And probably the last four or five years it kind of got a lot worse. In a fairly short period of time and then it flattened out again.

CMT- affected individuals mourned their losses, that is to say, losses in abilities or bodily functions due to atrophying. Irrespective of where they were in the downward deterioration cycle, or even if they were temporarily on a plateau, mourning took place each time a significant loss occurred. The following quotation aptly explains the mourning phenomenon:

Elke verlies wat jy ervaar is soos ‘n sterfgeval. Daar is elke keer ‘n routydperk. Jy steek dit vreeslik weg, maar die oomblik as ek saans gaan lê, huil ek my kussing nat oor daai verlies wat jy nou weer moet verwerk. Jy kom op ‘n plato; dan is jy weer reg en kan jy weer cope. En dan op ‘n kol kom jy agter ek kon nog laas dit gedoen het, maar kan nou nie meer nie. Jy moet in die hoekie gaan sit as jy daar beland, anders onderdruk jy dit. As daardie emosies dan uitbars, het jy probleme.

Every loss that you experience is similar to the situation when someone dies. With each loss, there is a time of mourning. You try to hide it, but the moment I lie down at night, I cry so much about that loss that I must now deal with again, that my pillow gets wet. You get on to a plateau, then you are okay and can cope again. Then at some stage, you realise that you cannot do things that you could do. ..... you have to go and sit in a corner if it happens, to work through it: otherwise, you suppress it. You have problems if those emotions erupt.

The CMT- affected responded to the deterioration threat with a myriad of short, medium and long-term strategies, ranging from purely cognitive processing to very concrete actions. In a broader context, it may in fact be reasoned that most of the strategies employed by affected people to manage their CMT, in one way or the other addressed looming deterioration. Many strategies were nevertheless more specifically directed at handling the disease's practical effects and consequences, for instance finding alternative ways to do tasks, rather than the causal condition of deterioration per se.

One important aim of deterioration-related strategies was to make provision against future atrophying. Making financial provision was considered important. Taking out insurance policies, for instant endowment policies, was the obvious route for some. Disability insurance was not always accessible; a CMT-affected person stated rather despondently: "*Financially I cannot get any disability insurance"*. For working individuals who belonged to a pension fund, it was crucial to hold on to their jobs until normal retirement because of superior pension benefits.

Providing for one's future safety and security when they were physically weakened was very important to participants. This was strongly related to the place of residence. The phenomenon can hardly be better explained than the following participant did:

I bought myself a townhouse in a security complex where I want to retire. You feel much safer there. I know I am going to be vulnerable- your entire body is advertising "soft target". I will have to think about not carrying large sums of money on me; will have to conduct business in a different way.

Also concerning the place of residence were plans and strategies to arrange or alter it with a view on practical convenience when one became weaker. It included actions such as installing rails next to steps, levelling of the walking surface and so forth. Some affected people also contemplated acquiring a property without steps. Others provided for the possibility of an old age home if the worst-case scenario became a reality.

Most CMT- affected people pondered on the matter of future social support when deterioration is taking its toll. Actions they took included communicating to one's spouse and other support sources about the matter and making appropriate arrangements. By way of example, a CMT- affected woman who acted rather pro-actively:

Hy (eggenoot) maak nie kos nie. Hy kan wel tee maak en koffie maak. Ek het nou vir hom gesê as hy af gaan, sal hy net moet kyk wat ek doen. Ek sal hom leer. Hy moet beginne kos maak.
He (husband) does not cook. He can make tea and coffee, though. I have told him now, once he retires, he will have to watch what I am doing. I will teach him. He will have to start preparing food.

A practical step that affected people took in order to get some control over the uncertainty was simply to go and see a neurologist in order to establish how far they have deteriorated. At best, they could get an indication of the rate at which they are going backwards and then base future projections on this. Some individuals based their future projections on comparisons with how rapidly other CMT-affected family members had deteriorated. Others meticulously monitored their bodies for signs of degeneration. They were furthermore very sensitive to pick up comments from significant others about their physical functioning, for instance that they were lately walking funnier than before. Typically, when deterioration was detected, anxiety would escalate. There were also people who were more pro- active in their provision for future decline:

.....jy kan beplan wat is jou modus operandi vorentoe. Een ding wat jy kan doen is om te lees. Wat ek gedoen het en steeds doen, is ek gaan op die internet en lees, soos nuusbriewe. As ek iets

sien waarmee iemand sukkel, dan bêre ek dit. So bou ek vir my 'n databasis op, sodat as jy op 'n punt kom waar daardie persoon is, is dit nie vir jou onbekend nie en leef jy makliker daarmee saam.

..... you can plan your modus operandi ahead. One thing that you can do is read. What I do is go on the Internet and read, for example, newsletters. When I see something that somebody struggles with, I file it. In this way, I compile my own database. When I get to the stage where that person is, it is not foreign to me and I can live with it easier.

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With rare exception, people also put their faith in the hands of the Lord and held firmly onto it, as is reflected in the following quotation:

I think generally I would like to just pray that it won't come to that point (dependency) *and I will hold on to that prayer. I sincerely believe the Almighty never gives us a burden we can't handle and He will not give us something to do that you're not comfortable with. If it is, it will be a big challenge and I am not able to face that challenge. I'll rather pray that I am never in that position and if it ever comes to it that I must go down that road, the Lord must just take me with my faith intact.*

This section attempted to isolate and illuminate a number of specific problems and strategies regarding the deterioration phenomenon. As already mentioned elsewhere, fear of deteriorating to the point of dependency was a very pervasive condition. A number, but not all, of the strategies of *engaging with CMT*, and in particular *fighting back*, that will be discussed in the rest of this chapter and elsewhere, had at the heart of it explicit or implicit attempts to alter, halt or slow down the process of deterioration over time, and/or at least to limit the consequences of it.

Persevering

As a strategy, persevering with goal orientated efforts in order to survive the short and long term consequences of CMT proved to be equally pervasive as the structural condition of possible rampant deterioration, discussed above. *Persevering*, although most prominent and noticeable in the stage of *fighting back,* was encountered throughout the entire process of *engaging with CMT*. The sub-process of *persevering* entailed: (1) a firm commitment to keep standing and to

cope for as long as possible, (2) practical, reactive strategies in response to the symptoms of the disease, (3) making provision for future developments in the disease's trajectory and (4) a more intrapsychic process involving identity issues.

Never surrendering

Never surrendering, which resembles a fighting spirit, encompassed making and living a firm, lifelong commitment never to surrender to the disease's manifestations and not to lapse into self-pity, no matter what. The interview responses of individuals with CMT were peppered with examples of persisting and refusing to give up. Two people's verbalisations were selected amongst many:

1. Ek dink my wil is net heeltemal te sterk om in te gee daaraan – ek het te veel go in my en ek is eerlik met jou, ek kan my nie in die toekoms sien agteroor sit met hierdie siekte nie.
I think that my will is by far too strong to give in to it - I have too much go in me and to be honest, I cannot see myself in future sitting back with this illness.
2. ... daai wilskrag, solank ons kan moet ons. Ek weier om op te gee tot ek nie meer kan nie
..... will power, as long as we can we should. I refuse to give up until I cannot cope any longer.

Non-surrendering also entailed a commitment not to feel sorry for oneself, that is to say, not to live in sackcloth and ashes because of CMT. This amounted to a mental attitude to be strong and did not imply that CMT affected people doggedly rebelled against the inevitable or that they persevered with futile efforts in the face of profound deterioration. Available resources and the circumstances of each person also influenced the extent of their living the non surrender-commitment.

Finding alternative ways

This strategy amounted to finding alternative ways and means to do tasks and fulfill roles that became problematic due to the effects of CMT. These actions were either born out of necessity, that is to say, stemmed from practical needs, or were employed to maintain autonomy and independence. Contextual conditions in the form of problem-solving and creative abilities determined or at least

mediated people's success with their efforts to generate alternative strategies. A female's comment almost reflected this in vivo:

Dan moet ek 'n ander manier kry om dieselfde ding nog steeds te doen. Mens raak dus vreeslik innoverend en kreatief, want jy moet gedurig aanpas. Almal is nie in staat om dit in dieselfde mate reg te kry nie. Party is meer kreatief en kry dit reg, terwyl andere net sê hulle kan dit nie meer doen nie.
Then I have to find a different way to do the same thing. You therefore become really innovative and creative, because you have to adapt continuously. Not everybody is capable of succeeding in this to the same extent. Some are more creative and manage to get it right, whilst others just say they cannot do it any more.

In general, however, people with CMT proved to be capable of much creativity in their quest to find alternative solutions to problems. The following remarks were made by a participant who has a type of CMT that, according to the person, resulted in serious visual impairment:

Ek kon op 'n stadium nie eers tandepasta op my tandeborsel kry nie. Tot ek agtergekom het, druk die tandepaste in jou mond uit en sit die tandeborsel in jou mond, dan werk dit uit.
At one stage, I could not even manage to put toothpaste on my toothbrush. Until I found out that if you squeeze the toothpaste into your mouth first and subsequently use the toothbrush, it works.

Examples of how affected people solved problems related to activities of daily living abounded. A few commonsense examples were: sitting down instead of standing up while doing household chores, typing instead of writing, heating the study to warm hands and aid writing, utilising the computer and wearing boots to counter ankle sprains. Where available, individuals also used specialised equipment as far as possible, for example bottle openers and special pliers to thread cotton through a needle. A male who regarded himself as being seriously affected by CMT, verbalised his efforts to maintain control over everyday life as follows:

You learn different things. When I am in the factory, I always have a cordless screwdriver handy, because I can't work the screw manually. And a pair of pliers, so that if I drop

small screws, I can pick them up. I think through time, you realize it is a problem. Like I can't button up my shirt – so I make sure that when my shirt is ironed, it is buttoned up already so that I can just pull it over my head. See, you always work out ways around it.

The changes forced by the progressive deteriorating nature of the disease were in some cases far-reaching, as is reflected in the following response:

Mens leer om aan te pas. Daar is bv. baie goed wat my linkerhand by my regterhand oorgeneem het. Ek is regshandig, maar doen veel meer met my linkerhand, soveel so dat ek partykeer met my linkerhand skryf - nie so netjies nie- omdat die regterhand nie meer wil werk nie. So mens moet aanpas – as 'n ding nie op een manier wil werk nie, probeer jy iets anders.
You learn to adapt. There are for example many things that my left hand took over from my right hand. I am right handed, but I do much more with my left hand - to the extent that I sometimes write with my left hand, not so neat, because my right hand does not want to work any more. So one has to adapt, if something does not work in one way, you try something else.

Getting control over one's is life in the context of finding alternative ways and means were even more attainable where contextual conditions such as ample financial means existed:

I bought this system from the United States for my pool. It is a big pump; it sucks water from the bottom and pushes it out so you have to swim against it. It has a speed control and a mirror at the bottom, so your position yourself and swim against the current. Unfortunately, it cost an absolute fortune.

Commendable in conditions where alternatives were limited due to physical limitations, CMT- people still attempted to obtain results:

I tried to improve each "grey" area that they pointed out, but with negligible success. I worked very, very hard and long hours to compensate, trying my best to add value for my employer.

Earlier in this chapter, getting suitable shoes was listed as a major problem that confronted the CMT-affected and that they had to solve. The most common solution was to relinquish the futile quest for suitable fashionable shoes and to go

for practical footwear that supported the feet and ankles. These were mostly boots or lace up shoes. A woman explained:

Vroeër het ek meer as 140 paar skoene gehad, alles ontslae van geraak, kan glad nie ‘n hak dra nie. Ek dra al hierdie goed wat my hele voet vashou.
In the past, I had more than 140 pairs of shoes. I had to get the rid of them all; I cannot wear a shoe with a heel at all. I wear only the things that support my whole foot.

Resisting identity of being disabled

To what extent did people who have CMT regard themselves as disabled? This proved to be a sensitive and difficult issue with which all participants grappled. One possible, and perhaps apparent, line of reasoning could be that this would largely depend on the seriousness of their condition, in other words, the degree of impairment present. However, data analysis revealed the true complexity of the matter. Despite the fact that all participants bar one rated the severity of their CMT as above average or average on their background forms, only two, of which one had another serious disability forcing major dependency, regarded themselves as belonging to the disabled community. The vast majority were decisive in their view that they were not disabled and firmly resisted taking on this identity. They in fact felt they were not abnormal in any way.

Three people's statements are indicative of this identity-resistance. The first is a male in his 30s who visibly was seriously affected in terms of symptoms and who rated the seriousness of his CMT as above average, the second and third are two women who rated their condition as average and above average respectively:

1. I do not consider myself as disabled. It is more in your head.
2. I don’t see myself as disabled. I do not park in a disabled bay. Before I used to look for the closest parking, but I’ve decided to change my attitude. Now it’s a chance to walk.
3. I have worked through many things, a lot of personal demons and situational demons, so I would say I am very grateful. Society puts a lot of things on us and expects you to do a lot of things. If you don’t do it that way, then you are not normal. And I tell a lot of people it is not a matter of being normal or abnormal; it is about doing it differently. You do it one way and this person does it the other way. They’re not abnormal. I think I am

very strong and passionate about that. I don't like people to say "normal kids do this or normal people do that". There is nothing abnormal about us (people with CMT). We just do it differently. The English language has so many adjectives you can use. Don't use abnormal.

The third response in particular reflects this woman's struggle with her identity in the context of CMT over a long period, as well as how she cognitively worked it out, eventually got clarity in her own mind and integrated her condition into her self-concept. I should add that this woman functioned under conditions where she was blamed and stigmatised by her spouse's family for "bringing this inherited disease into the family".

Another male who had broken records in his sport activity, aptly illustrated how CMT threatened his public image and therefore negatively affected his social identity:

The thing is, on the front of the newspaper I've got to say: "I've got CMT". The guys here at work – what will they say? And my kid's friends. They will ask who this cripple is. What are they going to think?

In the case of women with CMT, the matter of public appearance was particularly noticeable because they felt that their femininity was threatened:

Vir my was dit baie swaar. Selfs jou vroulikheid het 'n probleem daarmee. Ek kon nooit mooi skoene dra nie. Met 'n hakkie of so nie. Toe die punt-skoene in die mode gekom het, het ek dit ook probeer, maar dit het nie gewerk nie.

It was difficult for me. Even your femininity has a problem with it. I could never wear pretty shoes with a heel or so. When the pointed shoes came into fashion, I tried it, but it did not work.

Resisting, or grappling with, an identity of being disabled often spilled over into various strategies that aimed implicitly or explicitly to hold on to "normality", autonomy and independence. This was taken to the extreme in the following case:

If I am taking steps to make things easier, I feel that I am losing (because he wants to do it the "normal" way). The people at work know not to take things away from me. When I

am trying to put a nut on a bolt, they won't take it away from me, because then I fight. They know I will keep struggling until I manage.

In some instances concealing CMT from others served various practical purposes, for instance countering possible stigmatisation and discrimination in a competitive environment like sport or pending promotion at work. Concealing their CMT from others was also a strategy that some used to protect their identity and not to move into the role of a disabled person. In other words, it had to do with resisting the identity of a disabled person. This proved a difficult task and the futility of these efforts was dramatically illustrated in the following case:

One day I was with a friend and I was wearing these (ankle-foot orthosis). His daughter said "what's under your socks?" I told her it is braces and she asked what it's for. I told her that I have a problem with my feet, I can't walk properly, so these help me to walk. She said: "Why do you wear socks over it" and I said I was hiding it, because I am embarrassed. And she said "that's really stupid" and walks off. And I think this is a seven year old and I thought, maybe she is telling me something.

Empowering

The strategy of empowering entailed efforts to equip and empower the individual so that he or she was enabled to effectively deal with CMT and its effects. It encompassed a wide spectrum of actions ranging from those aimed at self-development, through practical strategies aimed at, for example, strengthening the body, to spiritual empowerment.

Similar to so many other strategies, a large component of empowering may be conceptualised as goal orientated efforts by the CMT affected to get more control. This entailed not only more control over the uncertain future, as was introduced in the section on deterioration, but also control over the environment, which was becoming increasingly difficult for people with CMT to master due to their weakening bodies. By way of equipping the self, including the physical body, with skills, competencies and abilities, better survival in their environments would (hopefully) be ensured. A practical example of this is where a woman who never

deemed it necessary to obtain a driver's licence because her husband had one, now goes for lessons in order to get her own. This would enable better coping in her environment.

The strategy of *empowering* comprised three sub-strategies, namely empowering the self, empowering the physique and empowering the spiritual aspect.

Empowering self

Whereas many strategies encountered in *engaging with CMT* may be conceptualised as aiming directly or indirectly to empower the self, it emerged here as a rather distinct strategy aimed more at mental, rather than physical, enablement. Strengthening the self in this context amounted to both cognitive work as well as work on the self-concept.

The quest for control over the disease by the CMT-affected could hardly be more apparent than with the *broadening of their knowledge base* about the disease, including how it affected them, as it was encountered here. Not only did this strategy empower the affected individuals with insight that enabled better coping, but was also invaluable in helping them to unravel many uncertainties about their own particular symptoms. Perhaps to be expected with such a rare disease such as CMT where information is not readily accessible, the quest for information and efforts to unravel the workings of the disease continued from where it left off in *orientating*. There, however, the aim was to get a basic idea what it was all about, whereas here it entailed more focused efforts to get to the bottom of their CMT.

The gathering of information about CMT did not fundamentally differ from what participants did in *orientating,* but by now was much more intense because of the ever-increasing number of questions and concerns that needed clarification. Concerns such as inheritance and deterioration remained burning issues. Examples of additional matters that were being researched, included: (1) whether CMT affected people should follow a special diet and, if so, what it would be comprised of, (2) how much if any and what types of exercise should be engaged

in, (3) what genetic engineering entailed, (4) the wisdom of surgery as a means to rectify defects, and (4) dietary supplements.

The sources of information also remained relatively the same as in *orientating*, but here, once again, were much more intense, making thorough use of all the relevant resources. Internet websites with good, reliable information on CMT were now better known, as were publications on CMT, for instance brochures by the Muscular Dystrophy Foundation of SA and other institutions. In addition to utilising neurologists and other medical professionals, more use was made of applicable medical support services, for example the branches of physiotherapy, biokinetics, and orthotics, to obtain information on neuromuscular diseases. In some instances, individuals joined international CMT organisations or the local equivalent, namely the Muscular Dystrophy Foundation of SA, or at least regularly received their publications. One person said in this regard:

Is there anything I want to ask you? I don't think so. I have done so much research into CMT, there's not much. I belong to CMT America and I get all their magazines.

In certain instances, the research done by participants empowered them with the latest information and even cutting edge theories about the disease, as in the first quotation below. In other instances, the information was more on a "raise the eyebrows, interesting to know-basis" as in the second quotation:

1) Omdat dit glo ook ‘n outo imuun siekte is, wonder ek – ek het byvoorbeeld al in my skouers ook hierdie vretende pyn ervaar.
Because it is apparently also an autoimmune disease, I have wondered - I have for example also experienced this gnawing pain in my shoulder.
2) As jy en ek ‘n kilometer geloop het, het ons eintlik 4 kilometer geloop – hulle het dit so uitgewerk; ek het iewers daardie statistiek. Jy moet als met 4 maal. Dis hoekom ons so moeg word.
If you and I have walked for 1 kilometre, we have actually walked four kilometres. They have calculated it, I have the statistics somewhere. You have to multiply all our efforts by four. That is why we get so tired.

The strategy of broadening of knowledge about CMT empowered affected individuals with external information that contributed to their effective management of the disease. A concurrent process took place on a more personal level and concerned the unravelling of their own CMT. The aim with unravelling CMT was empowerment of the self with insight and understanding about how the disease affects their own bodies, thereby enabling better coping with and management of these effects. In many instances, this strategy encompassed embarking on the exploration of their bodies, focusing on where and how it was impaired by the disease. Despite confusion and even more questions being generated by this process, affected people somehow managed to unravel many puzzling aspects about what is going on in their bodies. The following woman was trying to clarify for herself why she was falling so often:

Then also I notice when I am agitated or very tired, I tend to fall over my own feet. You are not concentrating, the brain does not get the message quick enough, it has a lot to do with the nerves. The brain does not get the message to the leg and that's why you fall. I notice that it is because I have too many things on my mind.

Another person unravels why she experiences a distorted sensation:
"Something that happens to me is that it feels on my hand or wrist as if I've got a jersey on, a hot spot, and then there is nothing. It is a rotten message coming from your brain. The sensation is wrong."

An interesting phenomenon that puzzled many CMT-affected people, and one which nobody successfully unravelled, concerned a contradiction. It entailed the anomaly that, although their feet have diminished sensation and in fact are typically described as being "dead", or "I can't feel my feet", most people reported that, when barefoot, even minor items or protrusions (one participant said even rice grains) on a surface cause pain sensations. Not that many CMT people often walked barefoot. The lack of support made them feel insecure and they avoided walking barefoot as far as possible.

The strategy of *self-development* entailed the broadening of skills and capabilities by way of attending relevant training programmes and activities.

Some participants embarked upon further studies, be it long term formal programs such as university degrees and higher scholastic qualifications, or shorter informal courses and seminars. The nature of the studies was diverse. It ranged from matric to post graduate University degrees, as well as various non-graduate communication and counselling courses. The following person had come a long way since she left school before finishing matric:

I am working towards a degree now. I didn't get matric and tried many other avenues so that I don't have to go through matric. That was a big issue for me. Then I did lay courses with different organisations and got certificates and all that. Now I am doing it through the University of .... .

Studying, as it emerged here, furthermore had a fighting dimension to it, which is to say in addition to self-development, as is illustrated by the following CMT associate when he said:

I knew that I would have to do much, much more than healthy people to make progress in my job. I did many in-house and external courses on various topics. Most were management courses. I also embarked upon postgraduate studies. I could not rest on my laurels.

An important process that people with CMT underwent was *reorganising of their self-concept.* Although the CMT-affected fiercely resisted taking on the identity of a disabled person, as was discussed elsewhere, they were nevertheless confronted with the reality that they did indeed have CMT. Furthermore, as they over time lived with the disease, they were forced to make adjustments to the way they do things and to their lives in general, in order to accommodate the effects of the disease. All this resulted in CMT-affected people experiencing more and more pressure towards better integration of the disease with their self-concepts. The following response serves as an illustration of how body image was negatively impacted upon by CMT:

Ja, in die volwasse jare tel dit (die fisiese) nie so baie nie, alhoewel ek dink tog mense takseer jou maar so bietjie op jou voorkoms. Hulle sien jy makeer iets en dis tog op ‘n manier asof hulle nie soveel vertroue in jou vestig nie – soos bv. in 'n ou met ‘n mooi sterk fisiese voorkoms nie. Dit het maar op die mens se siel ‘n effek.

Yes, in your adult years it (the physique) does not matter so much, although I think that people still judge you to some extent on your appearance. They see that something is wrong with you and it is as if they do not put so much trust in you as in a guy with a nice, strong physical appearance. It does indeed have an effect on ones soul.

Although a diagnosis places a label on their condition, self concept-work in the context of dealing with weakness and other symptoms has actually been going on since childhood and even early childhood. In those years, some children were fortunate to survive without severe negative implications for the self-concept. Many however, grew up in less favourable school and other environmental contexts where they did indeed suffer damage to their self-concepts. An example of this is the sport culture in schools that put enormous pressure on children who were physically weak and the stigmatisation of the weak and marginalised by so many elements in society.

In both above instances, wrong or irrelevant attributions for the observed weakness in children by significant others affected the former’s self-concept. Many attributions were benign and featured rather neutral explanations, for instance "childhood clumsiness" or "a careless child". Embedded in this was the implication that the child will outgrow it in adolescence or early adulthood. Other attributions were more negative, for example "useless child" or "weakling". Pre-diagnostic adults also faced challenges in the context of self-concept but were better able to deal with it due to more mature cognitive abilities. It nevertheless remained difficult for them to account for their poor physical performance. Many activities such as dancing, social sport and even walking long distances were not for them and passed them by. The majority simply accepted that, for some inexplicable reason, they could not manage these activities, or they avoided them. Many attributed the problems to "just not being a physically strong type of person". They suspected that something was wrong with them but did not know what.

The dramatic event of diagnosis put an end to all wrong and irrelevant attributions. After an initial period of orientating to the diagnosis, self-concept work became more prominent. Patterns in the data indicate that those with many early negative experiences tended to view diagnoses as a relief, whereas the more neutral and positive cases tended to see it as the opposite. In the former case the relief of knowing what was wrong resulted in increased self-confidence and even enhanced self-esteem, although the relief itself was short lived because stress about the uncertain future soon surfaced. The following response is indicative of this:

Receiving a diagnosis of CMT was not experienced by me as a catastrophe. Up to that stage, I had experienced a lot of unhappiness and really stressed so much for being so weak and not knowing why. Now I knew that it was not my fault at all, that all the time I have suffered from some weird disease! It made a major difference to my self-esteem and my way of thinking..... But although I knew now, I was afraid of what awaited me in the years to come.

In the second group, characterised by less debilitating childhood self-experiences and for whom diagnosis was more negative, diminished self worth was evident in some cases. Neutral attributions concerning weaknesses and other symptoms were now superseded by the reality of being labelled as a person with a progressive, incurable disease, with all the concomitant stigmatisation and negative implications.

Irrespective of the effect that diagnosis had on them, all CMT-affected people were confronted with the task of better integrating the disease into their self-concept. In many instances, this task reflected elements of fighting back, as was indicated in the following response. Notice how diagnosis dramatically resulted in enhanced self worth:

It was only after I was diagnosed, however impromptu it was, that I realised this is my life. God created me just as he created you and I am not going to make excuses for something I had no control over. Look, if I cook something and I burn something, it is my

own negligence. So I am not going to make excuses for something I had no control over. This is me, this is who I am and as the English say: accept me once and all and that's the attitude I took, and since then I found that my relationships are better; people around me are better, because I did not realise that I have this confidence. Now I knew: this is it, this is me – take it or leave it. I found that it made a major, major difference.

The more assertive manifestations of the self sometimes surfaced as nonchalance, as the following verbalisation illustrates:

Jy weet, ek het agtergekom dat as jy met iemand gesels (in die konteks van CMT) en die ou lyk asof hy wil begryp, dan sal ek aan hom verduidelik, maar as hy vir my 'n koue skouer gee, dan sê ek vir hom, man – dis jou verlies, As jy nie ingelig wil word nie, is ek nie bereid om met jou te sukkel nie.
You know, I have noticed that if you are conversing with someone (in the CMT context) and the person appears to be willing to comprehend, I'll explain to him, but if he gives me a cold shoulder, I'll tell him man this is your loss. If you do not want to be informed, then I will not even bother.

Work aimed at accommodating the disease into their self-concepts by CMT-affected people appeared to be an ongoing, cyclic process linked to the uncertain course of the disease. When they were on a plateau or when environmental conditions were favourable, adequate functioning was possible and a more positive self-concept more often than not manifested. When atrophying surfaced again, or when debilitating effects of the disease took its toll, for instance a painful fall to the ground, public humiliation and so forth, the harmony of self was threatened once more. Acknowledging that they were vulnerable and deteriorating despite their best efforts aided self-integration.

It should be stressed that integrating CMT into the self-concept did not mean accepting the identity of being disabled. As explained in *perservering,* regarding them as disabled was fiercely resisted by the majority of CMT-affected people. Having this incurable neurological condition was not regarded by them as a qualification for the category of disabled people.

Empowering the body

The strategy of empowering the body was a crucial disease management activity that people who have CMT engaged in. It encompassed strategies and actions that aimed to equip the body with capabilities, skills, reserves and strengths, as well as to rectify as many bodily defects caused by CMT as possible. The aim of this was twofold, namely: (1) to strengthen the body in order to prepare and provide for future deterioration, in other words, to build up reserve capacity as "insurance" against it, and (2) to correct and/or manage physical defects and manifestations to enable better mastery of the environment. Put tersely, by empowering the physique, better functioning, as well as longer and better endurance was envisaged. This process also included actions that did not directly impact upon the physical, but instead had an indirect effect. An example of this is the individual avoiding conflict because it sapped energy and left the body weakened.

EXERCISING, DIET AND WEIGHT CONTROL

This equipping strategy was directed at keeping the body and especially muscles supple, lean, strong and fit in order to function optimally. To this may be added endurance, with the implicit long-term intention to have longer staying power against atrophying. The following quotation is representative of many:

.....it (CMT) affects me badly. All I can do from my side is to remain as fit as possible and hope it (becoming dependent) does not happen.

Exercising included activities such as walking and swimming. Interestingly, the latter was the only type of exercise that everybody agreed benefited them, although cramping was a problem in the case of very cold water. Some exercised in order to improve certain disease related problems, for instance pain alleviation:

Ek het knaend lae rugpyn. Baie van ons ouens kla daaroor. Dis deel van die siekte – al wat jy moet doen is om lekker strekoefeninge te doen.
I have chronic low back pain. Many of us complain about this - it is part and parcel of the disease. All one needs to do, is stretch- exercises.

Whereas the intentions of the CMT-affected for exercising were clear, various conditions hampered the process. Considerable confusion existed as to whether exercising was at all a good thing, how exactly to go about the exercising endeavour, what forms were best, and, very important, where ones limit lay, in other words, how far one could push oneself. Many people embarked upon exercising programmes only to be disillusioned, discovering that their weakened muscles cannot respond adequately. The following interview text by a participant is indicative of this disillusionment:

I cannot lift my foot up when I do weights. I've tried exercising it, but no power. Nothing. It's crazy, I believe if there is a muscle there you should be able to grow it, but it just gets so weak so quickly.

Unfortunately for some, money was wasted on exercise programs that added no value, as the following person found out:

Ek was by die gym aangesluit, maar ek het my geld gemors. Ek kan nie op die roeimasjien nie, want my voet gly uit; ek kan nie op die fiets nie, want my voet gly uit; as ek die gewigte aftrek, is dit oneweredig – die enigste ding wat ek gevoel het daar vir my gewerk het, was treadmill, maar ek moet vashou, anders val ek.
I joined the gym, but wasted my money. I cannot go on the rowing machine or a bicycle, because my foot slips out, if I pull the weights down, they are unequal. The only thing that worked for me was the treadmill, but I had to hold on, otherwise I fall.

Gymnasium exercises were more fruitful when done under the supervision of a personal trainer. A woman, who started at the gymnasium after years of inactivity, did just that and stated:

I started gymming recently, about a month ago. I take it easy, but I am not on my own. I got myself a personal trainer to help me and watch over me. My husband suggested that I should get a personal trainer. I told him about my condition; he is aware. When he pushes me, I say I am getting tired now; you shouldn't push me.

Unfortunately, because CMT-affected people were uncertain as to how strenuous their exercises should be and furthermore struggled to get information about this,

overworking of their weakened muscles and joints occurred. According to the following person, the overworking caused permanent damage:

'n Kollega van my het my gevra om saam met haar te oefen. Sy is 'n maratonatleed. Ons het drie keer per week, een uur op 'n slag, die trappe van die Uniegeboue uitgehardloop, op en af. Na 'n paar weke het my kniëe begin oppak. Hulle is nou oor die muur. As ek lang afstande loop, dan pyn hulle.
A colleague of mine, who is a marathon runner, invited me to train with her. We ran up and down the stairs of the Union buildings for about one hour at a time, three times a week. After a few weeks, my knees began to pack up. They are now permanently damaged. When I walk long distances, they ache.

The CMT-affected did not follow any special *diet* as such but most focused mainly on fresh, healthy foods in general, of which fresh fruit, vegetables and lean meat were the most obvious examples. Everybody was conscious of overweight being particularly bad for people with neuromuscular disease because of the increased burden on muscles and joints. Being aware however did not necessarily mean compliance and a few people admitted to carrying unnecessary weight. Overall, diet and weight control strategies were actively pursued by the vast majority of people and were commonly encountered in the data, of which the following quote is representative:

I do not eat fattening stuff, because I do not want to carry all that extra weight. When my pants are tight, it's time to cut down. No special diet.

Multivitamins, or specific vitamins especially B, C and E, as well as anti-oxidants, were nutritional supplements that people with CMT took, but more as a general precaution and not against any symptom or effect of the disease as such. One participant maintained that creatine worked for him. Considerable confusion was detected in the data about what worked and/or was safe to take.

In terms of habits such as smoking of cigarettes and drinking of alcohol, CMT-affected people followed a conservative approach with moderation being the

norm. Only one participant smoked, and this person was not a heavy smoker and intended to give up. If they used alcohol at all, it was done in moderation.

GETTING HELPING AIDS

This strategy was aimed at rectifying bodily shortcomings that resulted from CMT. The desired outcome amounted to enhanced quality of life because of better mastery of the environment. Not everybody used helping aids and even if CMT-affected people had acquired them, they did not necessarily use them full-time. Furthermore, the type of helping aids employed ranged from "drastic" measures aimed at stabilising joints to a mere walking stick to aid balance.

Supportive footwear, especially sturdy lace-up shoes and boots, was the commonest and arguably the most affordable helping aid that just about everybody with CMT benefited from. It has been said, perhaps on a lighter note, that being smart in the context of productivity, means functioning according to the 20/80 principle. By this is meant that one should ideally exert 20% effort in order to obtain an 80% gain or benefit, and not the other way around. If ever this was true, it is indeed so in the case of supportive footwear for people with CMT.

Without supporting the feet and ankles, these people had very little confidence in all contexts involving ambulation. A legion of sprained ankles, painful falls to the ground and toe injuries left them with considerable apprehensive anxiety about threats from the surface they were moving on. It included uneven surfaces, irregularities, protrusions, kerbstones, small pebbles and so forth. Add to this the diminished sensation in the feet, poor balance and drop-foot syndrome, and it becomes clear why supportive footwear was so beneficial. Even simple, common footwear like sturdy slippers made a big difference, as the following text illustrates:

Vir die winter het ek vir my ‘n stewel pantoffel gekoop wat my voete ondersteun en warm hou. Toe trek ek nou die dag ‘n paar sandale aan en ek was verbaas om agter te kom hoe selfs daardie pantoffels my voete ondersteun.

For winter, I bought myself a pair of firm, boot type slippers that support my feet and keep them warm. The other day I put on a pair of sandals and I was surprised to find out how much even those slippers support my feet.

Where prescribed, or deemed necessary and consequently acquired on own initiative by the individual, ankle-foot orthoses (an image may be viewed in **Appendix C)** was another helping aid that made a profound difference, that is to say, if correctly designed and applied. The following two people who both have had orthoses for some time illustrate this:

1. Dit is nou my plastiek voete. Weet jy, met hierdie goed loop jy so maklik, jy sweef oor die aarde! Ek het gesukkel voordat ek dit gehad het. Jy loop nou soveel makliker.
These are now my plastic feet. You know, with these things you walk so easily, you float over the earth. Before them, I struggled, but now walking is so much easier.
2. Yes, it is brilliant (Ankle-foot orthosis). I can hardly walk without these things.

Usefulness, although very important, was not the only property of importance. Another influential property of this sub-process was acceptability, which was determined by a few factors. First, there was the impact of using helping aids on the self-concept and identity. This aspect had been dealt with elsewhere, but will be briefly recapped here. Despite the fact that people with CMT to various degrees succeeded in integrating the disease into their self-concepts, they resisted the identity of being disabled. Often, helping aids, especially visible ones such as braces, ankle-foot orthoses and even walking sticks, were interpreted either as threatening independence or as being outright symbols of disability. They were therefore resisted. The following interview passage, in which a CMT-affected parent willingly accepted advice from her teenage child, is indicative of this:

I was very tempted. My son actually said I should not get one (a walking stick), because he said I am only ----- years old and I will lean on that more and forget about working on my own balance. He says what's going to happen is you get lazy. Rather use walls or shopping trolleys when you can, but do not go this route. I was tempted and even looked at some when we were in Durban, those that the spies have. But he said no.

Appearance also played a role in determining to what extent people identified with the aid. Could they picture themselves wearing it? Will it threaten their public image? Lastly, comfort and ease of use were equally important. The older types of orthoses were frequently manufactured out of metal, which was heavy and tended to be uncomfortable. The later, much lighter versions were a major improvement. The following interview extract illustrates this:

Die eerste keer toe ek by 'n ou kom wat vir my sê ek sal iets moet kry om my voete voor op te lig, het hy vir my van hierdie ystergoed gemaak. Ek was kinderlik ongelukkig en het my voete permanent weggesteek. Dit was lelik. Ek het langbroek gedra en daardie ding het sulke hake gehad en my klere stukkend gemaak. Ek was baie ongelukkig. Toe kom ek op 'n dag by 'n ander ou uit wat hierdie tipe goed maak en toe het hy vir my hierdies van plastiek.
The first time that I consulted someone who told me that I should get something to lift my feet upwards at the front, made me those metal things. I was very unhappy and permanently hid my feet. It was ugly. I wore long trousers and those things had hooks, which tore my clothes. Then one day another guy who makes these things made these for me from plastic.

To conclude, in their fray against CMT, the majority realised the potential benefits of helping aids and expressed willingness to consider them should there be a real need in future. Many were not yet ready for them, though.

UNDERGOING SURGERY

Largely, surgery that participants underwent as an attempt to correct deviances caused by CMT, were done during childhood or at some other stage before diagnosis. Since a diagnosis of CMT had not been made, wrong surgical procedures that added very little, if any, value were a common occurrence. Perhaps to be expected with such an unknown disorder such as CMT, this phenomenon was also to a lesser extent encountered post diagnostically. Be that as it may, CMT-affected people had mixed experiences with surgery. The following two quotations from different people indicate negative and positive outcomes respectively:

1. My toes began to curl up. They operated on both feet, one after the other. I was in hospital for about one month. When I eventually got home and tried on some shoes, I

noticed that the second toe of my right foot stood up and it was impossible to get my foot into the shoe. Back to the doctor who had to operate again because he had messed up.
2. Soon after I left school, I went for an operation to straighten my little toes. None of my shoes was suitable any more and the pain was just too much. The operation was a big success and solved the problem 100%. It is the only operation I ever had related to this condition.

For some people with CMT surgery was the only option since no other strategy or treatment was considered a viable alternative by medical professionals. One middle-aged woman has had 12 operations on her feet since the age of 16. The muscles in her feet were so weak because of atrophying that they could not hold the bones of the feet in position. The result was that the bones protruded through the flesh, causing bleeding and chronic infection, thereby forcing surgery. She is under no illusion about the unfortunate outcome for her:

Ek gaan die gebruik van my voete verloor. Ek het geen gevoel in my voete nie. Alle spiere is aangetas.
I am going to lose the use of my feet. I have no feeling in my feet. All the muscles are affected.

In sum, people with CMT were inclined to carefully weigh the recommendations of the medical fraternity and their prescriptions regarding surgery against criteria of necessity and perceived degree of added value to their physical functioning. It was a case of embarking upon the route of surgery as a last resort.

UTILISING MEDICATION AND ALTERNATIVE TREATMENTS

The employment of treatments and services rendered by medical professionals and related support services with a view to empower the body, thereby enabling better coping with CMT, were the gist of this action-strategy. It included both professional and self-initiated strategies and actions. Almost entirely reactive, that means in response to the disease's manifestations, employing these resources were not necessarily successful, for instance the use of certain alternative treatments, but at least it empowered CMT-affected people with some degree of control.

The first strategy to clearly emerge was the use of various types of **medication**. By far the most popular was the use of magnesium and calcium against muscle cramps. These minerals were found by many to be beneficial, as can be seen in the following quotation:

Nee ek slaap in die nag, maar my bene begin bietjie ruk. Ek het nou vir my 'n kalsiumaanvulling gekry met magnesium en dit help defnitief, die magnesium help. Ek het nie meer daardie erge rukpyne nie.
I do sleep at night, but my legs begin to jerk. I got myself a calcium supplement with magnesium and it helps, definitely the magnesium helps. I do not experience those acute spasmodic pains any longer.

It should be added that many dealt with cramps in the more conventional way of forcing the cramp out by standing on the foot. Related to cramps and in many cases the result of it, the symptom of pain confronted CMT-affected people and generated various self-management strategies. Both cramp-pain and diverse muscle/joint pain were much worse in conditions of cold, which for all practical purposes meant during winter. Using heaters and warm clothing, particularly warm socks and gloves, were the first line of defence. Many people resorted to more drastic measures, for instance prescription medicine, with mixed results. An affected person's strategy, representative of similar strategies by other, did just this and in this particular instance, it paid off:

Against pain, I use one of the older forms of anti-depressants. Not because of depression, but because it raises the pain threshold. It helps for the gnawing pain in my legs at night.

Others were not so fortunate. Nothing they did appeared to be successful in reducing the pain to acceptable levels. One can detect the desperation and despondency in the following response by a participant:

If I've been very busy, I just can't sleep that night. No amount of medication will take it (the pain) away so I don't even bother to take any. I'll have a very restless night because of my feet and I just wait. I do not know whether to stuff it in hot water or massage it, I do

not know what to do. I just wait. You're too exhausted, you can't really rest. You put cushions everywhere.

Another resource utilised by people with CMT was **alternative medicine/treatment.** The types of remedies and treatments were diverse and included rubbing their hands with aloe gel to aid sensation, using of a wedge-shaped cushion, a vibrating foot spa and submerging the feet in very hot water followed by cold water. In terms of alternative treatments administered by trained professionals, a few participants had tried acupuncture and any of the others, for instance reflexology, hydrotherapy and so forth.

Alternative medications and techniques were controversial. Some affected people never even considered using them, whereas a slight majority had at least tried some remedy at some stage. The basic nature and attitude of the person, being contextual variables here, influenced this process in the sense of very low risk takers not experimenting with these unknown remedies. The approach of the more explorative was typically to experiment with any reasonable option that might work, an approach demonstrated by the following individual:

Nee, ek het hulle almal getoets – ek wil weet of 'n ding werk.
I have tested them (alternative treatments) all. I want to know if something will work.

In a way, it is understandable that CMT-affected people experimented with the alternative approach. After all, there are only a very limited number of conventional medications available for symptomatic treatment of CMT, with no cure for the disease available yet. The CMT-affected however discovered that these remedies and treatments were no panacea, rendering erratic outcomes in terms of success, if at all. Fortunately, most maintained a good sense of humour:

Ek wonder partykeer of al hierdie goed wat ek probeer het my nie teruggesit het nie. Dan hoor jy van ko-ensiem Q en dan probeer jy dit vir 'n ruk en dan dink ek dit help nie, ek mors net my geld. Lag saam... Ja, jy weet jy mors jou geld, maar jy probeer dit nog steeds. Lag.
At times, I wonder if not all these things that I have tried have put me back. You hear about Co-enzyme Q and then you try it for a while and then you think no this doesn't help,

I'm wasting my money. (Laughs). You know you are wasting your money, but you still try it. (Laughs).

Some may regard the following verbalisation by a CMT-individual as being a rather balanced approach:

I do take alternative medicine from time to time but I have found that very few remedies work for me. I have never consulted a trained professional in this regard. Perhaps I should have. There is one homeopathic ointment that I find occasionally helps for aching muscles and a tissue salt called Kali Phos that tends to counter exhaustion, but nothing to speak about, really. These things are worth a try, if you can afford it. They are not cheap any more.

The utilising of **physiotherapists**, a strategy that perhaps resorts more under conventional treatment approaches, did not emerge prominently in the findings. Very few people reported having benefited from it (in fact, only one was decisive that he had benefitted), whilst the majority either had never considered using physiotherapy, or had only benefited from it to a limited extent. By way of illustration, a participant with a positive outcome:

Then I went to a physio and said I was struggling to walk. I go to her all the time, and that's when she – those braces and splints, you know these things. (Shows it to me). That's when she recommended these.

Empowering the spirit

Spiritual empowerment encompassed getting one's relationship with the Almighty on a sound basis if deficient, as well as relying on God in the management of all aspects of living with CMT. This sub process typically came into prominence in the later stages of *fighting back.* If individuals had a close relationship with God before being diagnosed with CMT, their spiritual life subsequently deepened and broadened. In addition to the extent of religiosity, another property of this subcategory that emerged was degree of involvement in church and religious activities. The marked increase in these activities was so dramatic that it was not an uncommon occurrence for individuals with CMT to be elected to positions in

the church structure. The following lady, diagnosed with CMT, described her moving closer to God as follows:

Jy weet ek het nader gekom uhm, ek gaan nie sê ek is 'n groot Christen of so nie, maar ek sal sê ek vra die Here altyd om vir my by te staan en ek wil nader aan Hom gaan, jy weet, want dis al man wat jou kan help. Wat vir jou bystaan.....
You know, I have moved closer, uhm, I won't say that I am a big Christian or so, but I'll say that I'll always ask the Lord to support me and I want to get closer to him, you know, he's the only man who can help you. Who will support you....

In conditions where individuals felt uncomfortable about moving on unfamiliar surfaces and/or an unequal terrain, church attendance was adversely affected. Then again, most social activities that involved moving over unknown surfaces were avoided. Naturally, this did not necessarily mean diminished religiosity. The following response is indicative of these dynamics:

Nee, ons gaan nie kerk toe nie. Dan het ek nog 'n probleem om daar rond te loop. Luister elke Sondag op die radio. Ek glo in God en alles, maar ek gaan nie kerk toe nie, want dis nog 'n plek waar ek gaan sukkel.
No, we do not go to church. Then I have an additional problem there to walk about. I listen to the service on the radio every Sunday. I do believe in God and everything, but I do not go to church, because it is just another place where I am going to struggle.

Even though the data indicated the seeing of a higher, God-given purpose with their CMT by participants, this aspect emerged much stronger in the last stage of *optimising*, and will therefore be discussed in that stage. Here, in *empowering the spiritual aspect,* the focus was more on utilising God and religion as a **resource** in the management of the disease. In doing so, the attitude of people with CMT was not at all manipulative, but instead a genuine, deep trusting in and relying on God, as well as using His strength and wisdom. This is aptly illustrated by a 40-year-old individual with CMT who stated:

You just trust in the Lord. Every morning you get up and ask God to give you the strength to carry on. We pray five times a day. If God is on your side, whom do you need? Only God can help you. You just need to raise your hand.

CMT-believers believed that living according to God's will permeated all facets of life and that His divine guidance should be sought in even the most of operational problems that emanate from the disease. Nowhere however did putting trust in the Lord emerge more prominently than in the case of the more serious effects of CMT. An example of this was the threat of rampant deterioration, and the following interview extract dramatically illustrated it:

I work on the premise, if you make provision, you indirectly ask for it (becoming wheelchair bound).You're trying to say, without the will of God this is what you're going to do. I would rather let the Almighty take care of it, because He never gives you a burden you cannot handle. I am not going to ask for that, because that is not what I want. You need to believe, your faith is very important. Religious wise you should not have a plan B. The Almighty provides plan A, B and C. We just follow Him.

Relinquishing full control and transferring it, or part of it, to God, aided acceptance of a fundamental reality that had gradually evolved and that now increasingly confronted people with CMT: the fact that the course of the disease was less susceptible to their best efforts to alter it than originally thought. The latter is characteristic of the final sub-core of *fighting back*, called "*accepting reality*". All people with CMT were confronted with this reality, not only the religious ones.

Accepting reality

Accepting reality may be regarded as a transition point between the stages of *fighting back* and *optimising,* the last stage of *engaging with CMT*. Similar to the ending of the former stage, the transition was in fact so gradual and tentative that it was more of a process in itself than a static "point" or brief occurrence.

Having fought CMT and all its manifestations for so long, that is to say, ever since diagnosis and even prior to that, CMT-affected people gradually, over many years, came to recognise that, despite their best efforts, they were still getting weaker and weaker. Their bodies were atrophying relentlessly, although in the majority of cases very slowly and erratically. So slowly in fact that many did not detect deterioration until it became very prominent, for instance inability to execute certain manual tasks any longer, or after tripping and falling due to more profound drop-foot.

In view of the fact that CMT-affected people have lived within their atrophying bodies for so long, awareness of deterioration was always present, but not necessarily to the same extent as in this much later stage of *engaging with CMT*, where the focus was more on accepting reality. Instead, once again, it amounted to a matter of degree and prominence. Engaging with the disease commenced way back when people were *orientating* to it, and the subsequent *fighting back* was an activity that was so comprehensive that it often masked the effects of deterioration. There were also many short-term successes that aided this lower awareness of deteriorating, for instance the learning of new competencies, the finding of alternative ways to do tasks and empowering of the body. Gradually, however, the reality of atrophying could no longer be avoided, denied or countered and affected people were forced to face up to it. In this sense, it was more a case of recognising, or of facing, reality, before it could be accepted. An individual with CMT, who regularly employed the strategy of concealing the disease to others, explained his process of facing reality:

Over the last two years, I have become more comfortable with it (CMT). This is what I have and I cannot hide it any more. It has become just so obvious. You might as well let people know. At least they don't feel uncomfortable.

Another CMT-individual, a real die-hard who visually displayed very serious symptoms, began to concede and reluctantly recognise the inevitability of the deterioration process. Her fighting spirit and resistance to a wheelchair may indicate that she was still in the earlier stages of the process of *acceptance of*

reality, but, on the other hand, no participant at any stage of *engaging with CMT* was comfortable about the possibility of sitting in a wheelchair:

Deelnemer: "Ek sal nie gaan sit nie. Gister het hy (eggenoot) vir my gesê ons moet vir my ‘n rolstoel kry. Toe sê ek vir hom, wat, vir die hospitaal?" Eggenoot: "Daar gaan ‘n dag kom dat sy sal moet sit in ‘n rolstoel". Deelnemer: "Ek weet, ek weet. Nicol (ek, die onderhoudsvoerder), jy weet dit ook. Dit gaan met jou ook gebeur, jy sal ook in ‘n rolstoel moet sit".
Participant*: "I refuse to sit down. Yesterday he (spouse) told me that we should get a wheelchair. So I told him why, for the hospital?"* Spouse: *"The day will come when you will have to sit in a wheelchair".* Participant: *"I know, I know. Nicol (me, the interviewer), you know it as well. It is going to happen to you too, you will also have to go and sit in a wheelchair".*

In contrast to the rather reluctant acceptance of reality by the above individual, others more readily conceded, as is illustrated by the following excerpt from an interview with a rather young person:

…. when I read the stuff I get from the USA, I’ve got it pretty bad. When I take my age to other people --- (mentions own age) yrs old, they are having slight problems. I have serious problems already. That is a bit of a worry. Realistically, in 10 years, at some stage, I could be in a wheelchair. At what age, I don’t know.

The insight and acceptance characteristic of the process of accepting reality, were strikingly illustrated by the following individual who described the dynamics when he met another person who also had CMT:

So he told me he must just exercise more, he is sick of people feeling sorry for him. So I told him look here is my number, give me a shout if you ever want to talk. He was obviously where I was previously: there is nothing wrong with you, you just need to exercise more you'll be fine you don't need to worry about nonsense like this. So I thought just wait till he gets older then you'll start too.

For a very small minority of people, facing reality meant accepting the identity of a disabled person. This was contradictory to the majority, as already delineated

elsewhere. The following response illustrates accepting the identity of being disabled:

Ek het die laaste twee tot drie jare begin dink aan myself as 'n gestremde – as jy die belastingvorm invul vra hulle ook vir jou of jy gestrem is – die laaste twee jaar het ek ingevul dat ek myself as gestrem beskou. Vantevore het ek dit nie regtig so beleef nie, maar nou begin dit vir my so te voel.
For the past few years, I began to think of myself as disabled. When you complete your tax form they I ask if you are a disabled. For the last couple of years I have answered that I do regard myself as disabled. In the past, I never really experienced it as such, but now it begins to feel this way.

How much, if at all, did the fighting spirit of people with CMT recede when they were accepting reality? Data analysis indicated that it was not so much a matter of degree but instead the nature of fighting that was changing. Whereas formerly the accent was on countering the effects of CMT, in other words resisting being overcome by it, the focus now shifted more to the maximising of abilities, skills, competencies and quality of life in general. This amounted to optimising everything that they had left in their bodies and lives in general. Included in this was guarding over the body and caring for self, which means nurturing it, in order to preserve the strengths that they had left. This is reflected in the following response by an individual with CMT:

Ek besef ek kan nie onrealistiese eise aan myself stel nie – ek moet funksioneer binne die perke wat my liggaam vir my oplê. Ek probeer hom versorg, want hy moet vir my nog 'n ruk hou. Jy wil nie skade aanrig nie.
I realise that I cannot make unrealistic demands on myself. I have to function within the limits laid down by my body. I try to care for it (his body) because it still has to last for some time. One does not want to cause damage.

Accepting the reality that strategies such as opposing and resisting were not successful in halting the weakening process, most definitely did not mean that affected people surrendered to the disease. The commitment to never surrender was still as valid as ever, but the battle now shifted to a different frontier. Having come to terms with reality, people affected by CMT experienced more inner peace, yet were still motivated to carry on combatting the disease. This amounted

to persevering with the strategies of *engaging worst-case scenarios, never surrendering and always empowering*, but these processes now receded into the background. Efforts were now directed at the new focus that gradually emerged, namely making the most of their situation in order to optimise their living with CMT. The last stage of *engaging with CMT*, called *optimising*, had now been reached.

STAGE 3: OPTIMISING

Introduction

Living with CMT in many ways amounted to a continuous process of optimising. No matter what the threat, demand, environmental condition or context, people with CMT endeavoured to optimise their well-being and in many instances accomplished remarkable results. However, the exertion in terms of optimising up to this point, the energy and drive for it, was directed more at dealing with and overcoming the obstacles and problems emanating from the disease and not primarily at optimising per se. The focus was more on survival and mastery. Affected individuals typically were committed to fight the disease and its manifestations for as long as possible. This combatting inclination commenced tentatively in the stage of *orientating* and got momentum during the stage of *fighting back*.

Enhanced optimising of their quality of life as it emerged in this final stage of engaging with CMT reflected a much broader and deeper perspective on living with the disease than before. This mature outlook was indicative of the CMT-affected having being depurated by the struggle over many years, resulting in their acquiring a high degree of inner peace, linked to a more balanced view. Included in this was enhanced insight where both positives and negatives concerning the situation were considered, whereas before the focus was more on dealing with the negative manifestations. Furthermore, there was a drastic reduction in anger and bitterness, or at least these negative emotions were dealt with much better. A

typical response indicative of greater inner peace and positive focus was the following:

…aan die begin was ek baie opstandig – deesdae het ek wat dit aan betref baie meer vrede in my gemoed. Ek skop nie meer soveel teen die prikkels soos vantevore nie – probeer nou maar die beste van ‘n slegte saak maak en probeer my kragte so aanwend dat dit ‘n verskil kan maak.
… in the beginning, I was very insurgent- these days I have more peace of mind regarding it (CMT). I am not kicking against the pricks so much as before- I am now trying to make the best of a bad situation and to use my strengths in such a way that it will make a difference.

Optimising well-being for most CMT-affected people went beyond seeing meaning in their lot. It included focusing on activities that added value to their lives, as well as looking after themselves. A man, for example, who was confronted with the choice of going on early retirement or staying on in his job, saw it as follows:

Eventually I took the option of early retirement due to medical reasons. I must say that my employer offered me the alternative of a "lighter" position, but, after analysing the situation, I concluded that it wasn't much lighter and that the risk was too great. It was time to look after myself.

Informing others, in particular loved ones, about how the disease was progressing also now became a higher priority. Concerns about the future, including future anxiety, remained high; after all, it would be naive to expect future-anxiety to disappear altogether under conditions of CMT. However, individuals were able to deal with it better. One CMT woman described how she dealt with future anxiety by changing her outlook:

I don't want to become dependent. Every day is a blessing for me. I live one day at a time. Some days are so bad I can’t even comb my hair. But then there are things that you do that you take for granted, general things like taking a bath. I am very grateful for all of that.

Notwithstanding the ever presence of future anxiety, the enhanced inner peace and mature insight characteristic of *optimising* contributed to participants experiencing stress and anxiety (in general) as more controllable, at least to a greater extent than before. Although not exclusively so, these states also created favourable conditions for humour to surface. Examples of humour during the interviews include a participant demonstrating to me in a very funny way how she walked with high heeled shoes when she was "forced" to wear them, and another how he lost his balance in a camping shower, resulting in an embarrasing, but funny, situation. Some even described our way of walking (and similar manifestations such as running) humorously, as the following extract from an interview illustrates:

Man, daai dag loop ek weer met my voete so hoog in die lug, dit lyk asof ek sente optel en… (onhoorbaar weens gelag); My man wil hom ook doodlag.
Man, on that day I was walking as usual with my feet very high, it appeared as if I am picking up coins and…(inaudible due to laughter). My husband also wanted to kill himself with laughter.

In this stage, CMT-affected people still persevered with their quest for autonomy and, corresponding with all prior stages, the vast majority still resisted being regarded as disabled. It should be reiterated that they were still actively struggling against the manifestations of CMT. The fighting spirit in terms of refusing to be overcome by the disease, never receded throughout all the stages and even beyond. Optimising expanded the engagement by including the "making the most of"-component. This amounted to channelling energy towards maximising as many phenomena related to their CMT as possible in order to enhance adaptation and well-being. The "struggling against" and fighting elements, as well as the "overcoming" focus, moved into the background.

It should be pointed out that the terms well-being, quality of life and wellness are used interchangeably and not according to their true theoretical definitions where differences between the three may be encountered.

The stage of optimising comprised two sub processes of equal importance, namely optimising interests and broadening perspective.

Optimising interests

Optimising interests had two subdivisions or strategies, namely optimising one's own interests and optimising the interests and welfare of others.

The process of optimising one's own interests encompassed a wide variety of actions ranging from taking care of one to focusing on actions of personal significance. An individual with CMT aptly condensed the essence of this process by way of the following almost matter of course utterance:

He (a person with CMT) should take care of himself to have a good life.

The terminology "acting in own interest" has embedded in it the implication of prioritising, or re-prioritising, so that high priority, if not the highest priority, was given to actions that were performed to enhance self-interest. Perhaps the most apparent example of this was where people with CMT simply refused to do tasks or carry out responsibilities if they felt that it was too demanding or if they could not cope with it. Two women with CMT stated:

1. Cocktail parties are an absolute nightmare. Nowadays I refuse to attend them. There is no seating. You try to balance the glass, your snack, yourself, and your handbag. You can't do it, you actually cannot do it.

2. Luister na jou liggaam. As jy voel jy kan nie meer nie, moet jy ophou. As jou liggaam vir jou sê jy kan nie hierdie spoed volhou nie, dan doen jy dit nie.

Listen to your body. If you feel that you cannot manage, you should stop. If your body tells you that you cannot maintain this speed, then you do not do it.

Putting the self first, in other words acting in one's own interest, in many instances had a decisive element to it. CMT-people had over the years learned to stand up for themselves in a variety of situations. Now, this behaviour culminated in assertiveness and decisiveness in situations where the affected person felt fatigued or vulnerable. This was done to preserve the self and the body. These

dynamics are reflected in the following two quotations, taken from different interviews with CMT-affected individuals:

1. I had such high expectations, growing up with parents who expected you to do so much, that I actually feel embarrassed to say I need to sleep today. I would not be caught dead sleeping in the afternoon if my mother was around. Now I tell her I need to go home, I need to rest.
2. Previously, I pushed myself if I had to do something. If I knew you would come at half past nine, I would be ready and waiting by nine. It's not like that any more. If I am late, then I am late. Finish, because I have good days and bad days.

Re- prioritising also encompassed a cognitive process. Over time, individuals gradually came to realise that they had their priorities wrong by, for example, spending too much time at work or by exerting too much energy in an activity. This was illustrated in the following quotation:

I probably work 12 hours a day, six days a week. I work hellava hard. I feel this is time I should be with my wife and my kids, doing things while I can still move about. Maybe in 10 years or 20 years time I retire in a wheelchair. That is pointless. I am wasting a lot of time at work. That is worrying me and I am trying to get more people in so that I don't have to work for so long.

Focusing on *activities of personal significance* ranged from the more individually orientated interests, for instance certain hobbies, to contributing in both local and broader communities. People with CMT involved themselves in these activities and interests because it enriched their lives and countered typical disease related stressors, for instance symptom discomfort, negative thoughts and even self-pity. Where people were retired, the leisure activities frequently involved the rest of the family and in particular the spouse. A retired individual summarised focussing on activities of personal significance as follows:

Over the years, I've come to appreciate that it (the course of CMT) is out of my control and that the best thing I can do is to get on with my life and make the best of it. Focus on the things that mean a lot to me: my family, hobbies, music, nature, to mention a few. My

wife and I have tackled many new and exciting ventures- overseas travel, camping and so forth.

Hobbies included a wide range of activities from crossword puzzles to bonsai cultivation. Then there were also leisure activities that involved more physical effort, as well as capital expenses for those who could afford it. A working CMT-affected male explained:

I think I miss walking in the mountain most. I countered that by buying a 4 X 4 off-road vehicle, so that I can still go into the mountain. I am very fortunate that I have a good job and everything.

For the majority of the CMT-affected, optimising of welfare went far beyond expansion of own interests. Analysis revealed a significant (not the statistical variant!) external focus reflecting involvement on a much broader frontier, including the promotion of the welfare of others. CMT-affected individuals, especially in their later years, experienced participating in various community activities as very fulfilling and rewarding, especially to add value to a particular course. In some instances, this necessitated self-development in order to successfully occupy leadership roles in organisations and institutions. The type of work proved to be challenging and rewarding, as the following individual discovered:

I train basic counselling at an organisation empowering women. I am voluntary counsellor for them once a week and then I go to them when they call me. I do ----- (name of religion) counselling, since I studied ----- (name of religion) family law. I find it very challenging to work with people from other racial groups, to learn about their culture.

Getting involved in church and/or related religious activities were by far the most popular type of community involvement encountered in the data. This should be read in conjunction with *empowering the spiritual aspect*, as discussed in this stage of *fighting back*. In that sub process, the aim was more on utilising religion as a resource whereas here, in optimising, the focus has shifted to self-fulfilment and contributing. Furthermore, not everybody who underwent spiritual

empowerment in *fighting back* necessarily got involved in church matters to the same extent.

Getting involved in spiritual activities in a sense was a natural step for those who put their trust in the Almighty. Not only did it enable richer spiritual growth and empowerment, but furthermore offered the opportunity of contributing to something meaningful. In addition to spiritual enrichment, the meeting of new friends, enjoying a sense of belonging and networking, was amongst the advantages that CMT believers experienced. The next interview excerpt illustrated the social support component:

At the church-it was not long before they chose me as a Bloc leader - I met an incredible amount of people these past three years. I have the most wonderful friends. When I for example take a bottle down to open it, they just automatically take it from me and do it for me.

An interesting phenomenon to emerge clearly from the data was *making a contribution*; an endeavour that aimed ultimately at promoting the welfare of the CMT community, as well as other disadvantaged people. This process entailed a few variations but all had in common the adding of value by means of informing others about CMT and/or by sharing of own experiences. In so doing, it was hoped that hardship and suffering could be prevented or at least alleviated, especially for children and disadvantaged people affected by the disease. The following lady, whose school years were traumatic because of CMT, was passionate about the fact that school authorities be informed about the disease because harm to CMT- affected children and others with impairments could be avoided:

Ja, niemand weet nie. Niemand dink regtig daaraan dat daar ‘n rede moet wees daarvoor nie (val en lompheid weens CMT). Niemand gaan tog net gewoonweg loop en val sonder ‘n rede nie. In die tussentyd is mens so afgekraak. Dis hoekom hierdie goed vir my by die skole moet uitkom.
Nobody knew, they never actually thought about the reason for it (clumsiness and falling due to CMT). No person will just go ahead and fall to the ground without a reason. And

then what they did was just to degrade you and break you down. This is why it is important to me that these things get back to the schools.

To illustrate making a contribution in order to benefit those who may not be aware that they have CMT, as well as the marginalised, the following apt interview passage was selected:

Yes, I think I am almost morally obliged to make this thing a little better known. That is why I am thinking this completely disabled -- (name of sport) thing. I am not that keen, but if it is a way for me to get on the front page of a newspaper by breaking a record and they say I have CMT – these are the symptoms. You know, how many people are sitting in underprivileged areas that have it, they do not know, and a set of braces could mean they could walk again. I have the opportunity to get newspaper space and if I can use that to tell people this is what CMT is, this is what your feet look like, and this is what it does to you. Tell all the people in – (name of city) who has it and do not know and then they will realise this is what I have. I really want to do it.

It was rare indeed to encounter a participant that was not interested in participating in a CMT support group. Geographical distribution was, and still is, a major obstacle to a face-to-face support group, but individuals nevertheless expressed great interest in such a group or some variant of it. This matter will be taken up again in the next chapter. The main perceived advantages of such a group were meeting other people with CMT and the sharing of experiences.

A facet of optimising the interests and welfare of others, and one with which many CMT-individuals grappled, was the matter of informing others about CMT; the main issue being how much and what type of information to supply to stakeholders, including loved ones. These matters also received attention at earlier stages of *engaging with CMT*, but in ways applicable to those contexts. During *orientating*, at the time of diagnosis, informing significant others did take place, but the full implications of having the disease were not always realised at that stage. During *fighting back,* the focus was elsewhere and informing stakeholders was more on a "need-to- know" basis. Here, in *optimising*, informing others was more of a goal directed, considered effort based on the aim to foster the

well-being of all stakeholders touched by the disease, such as the employer and the family.

When employed full time, informing the employer about their positive CMT status, as well as how much of the concomitant threats and implications to reveal, proved to be a rather complicated matter. Although this has been the situation all along, in other words, in the two former stages as well, affected people who were in this last stage of *optimising* felt more compelled to optimise the interest of all parties. The danger of stigmatisation when the employer knew about the disease, with possible negative effects on one's career, always loomed in the background. On the other hand, revealing CMT to the employer could result in more suitable placement, optimising the interests of both parties. Data analysis revealed no clear pattern in this regard, mainly because of different environmental and other conditions operating in each case. Examples of these were the age of the affected person, years of service, the nature of the occupation, the employer's human resource policies, degree of seriousness of the CMT and the dispositions of the individuals involved.

What did emerge more clearly was that it became more or less impossible to avoid the issue of informing the employer under conditions of more profound symptoms. This typically occurred during the later stages of CMT when the effects of atrophying became more noticeable, resulting in diminished work performance and in, some cases, other staff gossiping about what was wrong with the particular person. When people were gossiping, pressure to reveal CMT increased. A professional person employed full-time handled the informing-matter as follows:

Ek het daai tyd toe ek agtergekom het daarvan (CMT) vir my kollega daaravan gesê en mense het daarvan uitgevind en toe was daar mettertyd baie gissings ens. Toe het ek in die werkgewer se nuusblaadjie so 'n uiteensetting gegee van wat die situasie behels. As jy wil aanhou werk hierso, dan moet die mense darem weet. Ek voel die ---(naam van beroep) gaan nie verloor omdat ek die siekte het nie, ek gaan nog steeds my werk doen.

At the time, when I found out about it (CMT) I told a colleague, and so the people came to know about it. In due course, it led to a lot of speculating and I subsequently wrote an

exposition about my situation in our in-house newsletter. If you want to carry on working here, at least the people should know. I feel that my disease would not result in my employer suffering any losses. I will still be able to do my work.

Another felt that it was in everybody's interest to inform his boss about his CMT at the time of his promotion to a very (for him) demanding position. It took place at a very late stage of the disease's course when atrophying was very disruptive. He described it as follows:

My last position was just too demanding and to make matters worse my boss never impressed me as a person with excessive empathy. Fortunately, I did inform him about my disease when he appointed me in my position. It was important to me that they should know.

For many CMT-affected people, it never came to disclosing their disease to the employer because they did not stay long enough, recognising out of their own initiative that they have to change to a more suitable job, retiring and, in the case of females, getting married and/or having children and so forth.

Another troublesome issue for people with CMT was what, if at all, to tell their children and then at what age to do so. All participants' spouses in this study were fully aware of the CMT situation and this matter therefore involved them too. Informing children was not such an important issue at younger ages, in other words the early primary school years. As children grew older, they usually became more aware of the fact that something was different about the affected parent's physique or physical functioning. If the children did not spontaneously inquire about this, the situation could have prompted the parent to disclose CMT. The following excerpt illustrates these dynamics:

My kid – yes, he knows. He does not know the full extent of how genetically it all works. When we play cricket, I have to sit down after a while – I can't go any more. And then he says "fine, we'll rest for ten minutes". So I've told him- he knows.

Parents who considered it to be to the advantage of the child to know, were inclined to supply information of a more general nature, as can be seen in the above response. It should be added that, if the child had clearly inherited CMT, information of a more comprehensive nature probably would have been supplied. This, however, was inferred and did not emerge visibly in the data. Other parents had more paternalistic views and/or values about informing children on sensitive or controversial issues. Typically, these CMT-parents considered it not to be in the interest of the child to know about the disease, or to know as little as possible. They were concerned about stigmatisation of the child and all the negative consequences that went with it. The following quotation illustrates:

Our family did not go too deeply into it, because society puts a lot of emphasis on it (having impairment). You make it difficult for your child to excel, because you have these expectations of ....(inaudible response). The less they know the better. You just say that he does things his own way.

Broadening one's perspective

The process of broadening one's perspective was influenced by a myriad of variables and conditions. Two that stood out and that were closely intertwined in the data, were the nature and degree of insight related to the broader CMT-phenomenon on the one hand, and the individual's approach to life, in particular life with CMT, on the other hand. In general, having mature, balanced insight usually, but not always, resulted in a more or less corresponding approach to life. An example of a discrepancy was where CMT-affected individuals experienced undue bitterness and resentment about their CMT lot, yet their approach to the matter of having children with positive CMT status reflected surprising maturity and lack of anger.

By way of broadening one's perspective, the CMT-affected individuals were able to enhance their wellness by way of at least two mechanisms. First, they countered tunnel vision and possible cognitive restriction that so easily arose out of negative emotions such as hopelessness, anger, resentment and so forth. This rather pro-active strategy not only opened their minds for new and more

possibilities, leading to better coping, but also led to a more balanced view and approach to life in general. For instance, many people who have CMT now also saw the positives in their situation, if present. The second mechanism entailed reduced levels of stress, the result of enhanced inner peace that accompanied the broadened perspective. Both mechanisms led to a myriad of outcomes, for example less bitterness and a deepening in appreciation for loved ones. Having walked the road and having accepted the reality about their CMT situation, there was a deep gratitude amongst participants for the stress and hardship that support systems had been subjected to because of CMT. A renewed realisation of their importance, past, present and future, also surfaced.

CMT-affected people were, especially in the later stages of their living with the disease, actively working at broadening their insight and approach to life. These efforts ranged from being grateful, that is to say counting their blessings, to the discipline of refraining from worrying excessively about the future. Furthermore, viewpoints regarding various issues that were formed over many years were now expanded or altered in order to optimise quality of life with CMT.

On a practical level, many affected people concluded, and were in fact living in line with this approach, that excessive concern and stress about the unpredictable course of the disease were largely counterproductive. Most felt that they had done all they could, in other words they had tried their best to fight the disease and to provide for the uncertain future, and were now just getting on with their lives. This went beyond accepting reality, as described elsewhere. It should be stressed that surrendering to CMT was still not an option, instead it amounted to making the most of an uncontrollable situation and living as full a life as was possible under these conditions. For many, it also meant focusing on the here and now. The following two interview excerpts illustrate these shifts in focus:

1. *Nowadays I don't look too much into the future, I don't let that get me down. I live for today, with my team and myself.*

2. .....ek vat dit dag vir dag. Ek gaan net aan. Solank ek die lewe kan geniet en dit vir my voel die omstandighede laat dit toe, sal ek dit doen. Mens moet seker maar kyk na vorentoe en wat gaan

gebeur en so, maar ek voel so, ek gaan net aan. Dit is altyd in my agterkop ek gaan dit nie lank kan doen nie. Ja, solank ek dinge kan doen, doen ek dit.

..... I take it one day at the time. I just carry on. As long as I can enjoy life and conditions allow it, I shall. One probably should look ahead at what is going to happen and so on, but this is the way I feel, I just carry on. It is always at the back of my mind that I won't be able to do it (physically demanding tasks) for much longer. As long as I can do things, I do it.

Seeing the positives in their situation of having CMT were encountered often in the data, in many instances rather unexpectedly in the context of the particular case. It was not a case of the rather far-fetched scenario of CMT disease having any inherent positives, but rather a matter of counting blessings in the circumstances. Furthermore, it was intertwined with being grateful for abilities that remained and for the status quo not being worse than it was. An example typical of the latter is the following response by an individual who was seriously affected by CMT:

Would I be angry with God? No, how can I be. I've got a great wife, a great kid, a great job and I am fortunate enough to live in a great place. I could be living a couple of kilometers up the road in a shack. Then my limbs might work properly, but I would have to live like that! I can't be angry about anything.

There were a surprising number of rather specific indirect outcomes of CMT that affected people experienced as more favourable. Moving closer to God had already been dealt with elsewhere. Two further outcomes were selected to serve as illustrations of this phenomenon:

1. I have good health; this is no sickness where you are afflicted in your mind. I don't have that and I am grateful for that.

2. Weet jy, ek en my Pa het nooit goed klaargekom nie – maar vandat ek jare gelede uitgevind het dat ek CMT het, was ek en my pa so close – dit is ook wat my positief daarna laat kyk.

You know, my dad and I never got along well, but we have been very close since I found out that I have CMT. It is another reason why I view it as positive.

By far the most frequently encountered "positive" consequence of having CMT was an enhanced empathy for disabled people and for people in need. In many instances, this represented either a shift in values or the cultivation of new ones, for instance commiseration for the marginalised and having more empathy in general. This process furthermore influenced the outlook on life that individuals with CMT had. The following interview text strikingly illustrates these dynamics:

The other day in the Spur, a woman sat behind us with a baby in one of those rockers, covered in a blanket. My husband wanted to look at the baby and I joined him. When the blanket was lifted, we saw a child lying there with only half of both his arms present. There were tiny fingers here and there on the arms. Whereas a normal person would have recoiled back, I asked the mother how old the child was. It turned out that he had some disease. The mother asked me if I wanted to hold the child, and I said yes. I could touch the child and there was no resistance or creepiness. You do see life through different eyes. You are not so prejudiced towards other people. If somebody tells me that they struggle with their feet, I have empathy. Your outlook on life does indeed change.

Certain individuals were adamant that CMT made them stronger people; in the sense that it had unlocked hidden facets of their personalities or that they were toughened by all the hardships. Furthermore, they had to acquire special coping skills to deal with it all. Others approached the matter from the opposite side, in other words, what they would have become had it not been for CMT. Three people's responses were selected as examples of these approaches:

1. It (CMT) has made me a stronger person. Since I've realised that I have it, something inside of me has come out that I did not realise I had. (She was referring to inner strength and a fighting spirit)

2. Maybe if I had physical ability, I might have done something else, not good, I might not have been the person I am right now.

3. Dalk as ek niks makeer het nie, was ek dalk 'n groter bliksem, mens weet nooit.

Perhaps, if there had been nothing wrong with me, I would have been a greater bastard, you never know.

Then again, there were those who felt that CMT brought them nothing positive, for instance, the following short, decisive statement by an affected woman:

Daar is nie ‘n spesiale voordeel daarin om CMT te hê nie.
There is no special advantage in having CMT.

Reflecting back upon one's life with CMT, an activity very characteristic of this last stage of *engaging with CMT* went beyond mere reactive thoughts about the past and embodied qualities of a mature and balanced outlook on various related issues. This included reflecting upon difficult issues such as the wisdom of having children in view of the genetic risks, how to respond to those who had stigmatised or hurt one in the past, and ones basic philosophy on gradually becoming weaker and weaker. Being grateful for their achievements and blessings in the context of CMT were also distinctive here. As already discussed, a lighter side, that is to say a sense of humour, also featured more and more.

Even at this advanced stage of living with the disease, reflecting about the wisdom of people with a CMT diagnosis having children remained as sensitive and difficult a matter as ever. Even after all these years, CMT-affected parents still found it difficult to separate their opinion on having children in general from their opinion on having had their own particular children. It was nevertheless to a lesser extent here. Most individuals, having travelled the road up to this stage, were now able to reflect upon this matter with more insight and wisdom than before, although some confusion was still encountered.

Various conditions influenced the degree of insight. First, reflecting back meant that parents drew upon their own experience and knowledge. Second, being the last stage of *engaging with CMT*, most by now had lived with their children for some time. This resulted in parents having a clearer subjective opinion about their children and grandchildren’s CMT status, although many were still anxious in this context. When descendants were CMT positive, parents were much less in favour of CMT affected people having children. This in turn, was influenced by how severely the descendants were affected. Third, background or contextual variables such as moral societal norms about the blessing of having children, abortion, disability, the role of parents, as well as the physical means to support them,

influenced insight and understanding. The following quotation illustrates a well considered (not in right or wrong terms) viewpoint:

I know someone who was tormented because his child inherited muscular dystrophy from him. Therefore, I asked him one day what would have happened if the child were born with a weak heart or with just one kidney. Would it have made a difference? In other words, what guarantee do you have that your child will not inherit something deviant? Why then discriminate against us?

The standpoint of people with CMT on the matter of having children of their own, as it emerged in *optimising* was threefold: (1) unconditional approval, (2) conditional approval and (3) disapproval. Interview extracts to illustrate each are as follows:

1. *Yes, I still would have had children, even if I knew I had CMT.*

2. Dis ‘n bitter moeilike kwessie. Ek sou steeds kinders gehad het, al het ek geweet. Ek sou die risiko gevat het, maar ek sou net een of so gehad het.
It is a very difficult matter. I would still have had children even if I knew. (that she had CMT). I would have taken the risk, but I would have stopped after one or so.

3. Nee, ek sou beslis nie kinders gehad het as ek geweet het nie. Want dan het hulle onnodig met hierdie probleem gesukkel. Sou liewer kinders aangeneem het. Veral hierdie kleinkind van ons. Nee, mens kan dit nie aan iemand doen nie. Maar as die kind daar is, moet jy die beste van ‘n slegte saak maak.
No, I would definitely not have had children if I knew (that he had CMT). Because then they would have suffered with this problem unnecessarily. I would have adopted children instead. Especially our grandchild. No, you cannot do this to another person. However, once the child is there, you have to make the best of the situation.

Parents who utilised genetic counselling when they had children, advised their children to do the same when they themselves wanted to start a family. Lastly, not everyone had succeeded in clarifying the matter of having children. Just listen to the confusion and indecisiveness in the following response:

At that stage, when we made the decision not to have another child, it was kind of a reactionary decision. You have just heard that you have the disease and we decided to stick with one child. It's difficult, one would probably think different if you don't have any

children. When we had --- (name of child), we were young and had many plans- perhaps we would have decided to have the child regardless. However, considering what I know and feel today, I could have decided that the risk might have been too great. But then again you do not know if you would have experienced feelings of emptiness that could have led to you deciding to have a child.

At this advanced stage of *engaging with CMT,* reflecting back furthermore encompassed meditating upon, and searching for meaning and a higher purpose in the context of having CMT. To illustrate this, the following excerpts from two interviews were selected:

1. Jy voel daar moet iets heilsaams hieruit voorspruit, wat 'n ander ou nie kan kry tensy hy hierdie paadjie (CMT) geloop het nie.

You feel that something wholesome must result from this (having CMT), that another person cannot acquire unless he had walked this road.

2. *Perhaps I got it (CMT) because the Lord wanted to send me on a certain path. My path went in a very different direction this past three years- everything has changed. I was always: just leave me alone, now it is: I have learned so many lessons...*

Most people, upon reflecting back on their lives with CMT, did so with gratitude and appreciation. Individuals were grateful for many things, for instance all their achievements despite being affected by CMT, and for numerous things not having turned out worse than they did. In this context, an individual who was placed on early retirement due to CMT, commented:

According to the neurologist's report at the time of diagnosis I am 50% disabled, and I would much rather focus on the 50% that I have left! There are many ways to kill a cat- it is still possible to live a good life. I myself achieved so much that I am thankful for. For example, the last position that I occupied was in management! I certainly had a rewarding life and still have. I am not bitter or angry with God or anybody for having CMT. In fact, I praise His name for my life, which has been, and still is, very fulfilling indeed.

Having a positive attitude (and a fighting spirit!) could hardly be more strikingly illustrated than the following woman, who was already quoted elsewhere, but in a different context. She said:

Jy weet, die ortopeet het nou met die laaste operasie (3 dae gelede) vir my gesê dat ek gaan my voete verloor. Hy kan vir my niks meer doen nie. Hy het gesê operasies sal nie meer help nie. Maar jy weet wat, ek lewe nog!! Wel, ek hoop nou maar hulle kry dit reg om voetoorplantings te doen. Ek verstaan dit nie, hulle kan harte oorplant en alles, hoekom nie voete nie? Ek sal die voete kies van die wat so gereeld toi-toi ('n soort georganiseerde openbare protes). Dan moet jy sien wat doen ek alles!

You know, the orthopaedic surgeon told me with my last operation (three days ago) that I am going to lose my feet. There is nothing he can do for me. He said that operations would not help any more. However, you know what, I am still alive! Well, now I hope that they manage to do feet transplants. I don't understand it. They can transplant hearts and everything, why not feet? I would choose feet from people who regularly toi-toi (a kind of organised protest in public places). Then you must see me go!

CMT-affected people were also grateful towards their support systems, especially family members. The support systems included friends, extended family and community organisations, for instance the church. The main support system was the family; there was a deep appreciation for the spouse and other family members for the many sacrifices and loyal support over so many years, recognising that it could not have been easy for them. This is demonstrated by a CMT-affected woman whose husband is approaching retirement:

As jy bystand het soos ek, kan jy voorsiening maak vir die jare vorentoe. Ek besef hy raak partykeer iesegrimmig, hy is ook moeg. Is nie meer vandag se kind nie. As hy nou eers by die huis is (na aftrede), kan mens mos nou lekker leef. Jy weet, hy gee vir my baie raad en dan probeer ek dit.

If you have the support that I have, you can make provision for the years ahead. I realise that he (spouse) gets grumpy from time to time, he is also tired. He is not today's child any more. Once he is at home full time (retired) we shall be able to live really well. You know, he gives me so much advice, and I try it all.

In the following instance, the main support person was a child; the affected mother's appreciation is strikingly reflected:

I lean on my middle son a lot. Always ask him to help me. Sometimes I just call him to have him there. He is really my pillar. He does such a lot for me. Sometimes I feel that I put too much pressure on him. The one he marries must remember he is half mine!

Another person had a deep consideration for those around her:

Daar is iets wat vir my baie belangrik is. Jy het die mense om jou nodig, maar jy mag nooit hulle lewens versuur omdat jy ‘n probleem het nie. Jy kan wél vir hulle sê, probeer om hierdie vloer so skoon as moontlik te hou, want dit maak my voete seer. Maar jy moenie gedurig "nag" en kerm nie. Dan moet jy maar net aanpas, anders skep jy vir hulle ook ‘n probleem.

There is something that is very important to me. You need the people around you but you should never be the bane of their lives because you have a problem. You may ask them to try to keep this floor as clean as possible because it hurts my feet. But you should not nag and whine all the time. Then you must simply adapt, otherwise you create a problem for them too.

In the process of optimising their well-being, most CMT-affected people had, over a very long period, developed several personal philosophies or viewpoints about their fate in inheriting the disease, as well as what it meant to be living with it. These philosophies simplified living with the myriad of CMT engendered stressors, for instance symptom manifestation, threats and various ambiguities. They also played a role in countering debilitating emotions like self-pity and resentment. A few of these viewpoints had already been discussed, amongst them were taking it one day at the time, making the most of the abilities that were left and putting all one’s trust in the Lord to provide.

In addition to the philosophies already discussed, a few apt viewpoints, philosophies and reflections were selected for clarification purposes and to serve as conclusion of the elucidation of *engaging with CMT,* which was the core strategy that affected people employed to manage their living with this disease:

1. Ek het besluit dit gaan my nie onderkry nie. Ek dink omdat ‘n ou ‘n ander uitkyk het op die lewe as jy hierdie siekte het – dinge wat vir andere nie belangrik is nie – stupid goed wat ‘n ou mee sukkel, dan besef ‘n ou wat belangrik is in die lewe. Ek het ‘n lewensfilosofie dat elke mens keuses het as iets met jou gebeur. Gaan jy die lewe aanpak en sorg dat dit jou nie onderkry nie? Jy

het een van twee keuse, wat sal dit jou baat om jouself jammer te kry? Ek glo verskriklik sterk in die keuses wat ‘n ou in die lewe maak.

I decided that it (CMT) is not going to get the better of me. I think it is because you have a different outlook when you have the disease- things that are not important to others- stupid things that you struggle with- then you realise what is important in life. My philosophy of life is that each person has choices when something happens to you. Are you going to tackle life and see to it that it does not get the better of you? You have one of two choices, how will it benefit you to feel sorry for yourself? I strongly believe in the choices that one makes in life.

2. *Life is too short to fall into self-pity. Life has a way of throwing you a curved ball and you must be ready to catch it. You cannot brood about things. Don’t let it depress you, get you down.*

OUTCOMES

Actions taken by participants to resolve or manage their main concern, or basic social problem, have outcomes or consequences. Glaser (1992, p. 6) defines consequences concisely: "They are simply an outcome from whatever happened before." Consequences in grounded theory tend to be of a temporal nature; a consequence of certain actions, for example, may become the cause of the next action. Strauss and Corbin (1990) point out that even failure to take action has consequences. An illness that becomes worse due to the failure to follow a regimen, is an example of this.

As was elucidated in the beginning of this chapter, the main concern or problem that people with CMT faced amounted to *how to deal with unpredictable CMT manifestations.* Their main task was to manage the symptoms, effects and consequences of the disease in such a way that an acceptable outcome be attained. It subsequently emerged that this task was accomplished by way of the core strategy, or basic social psychological process (BSPP) of *engaging with CMT*. The latter comprised three sub processes: *orientating, fighting back and optimising*, and these represented three largely sequential stages.

The fact that, in grounded theory, consequences of actions/strategies frequently become causal conditions of the following actions/strategies was also observable in the findings of this study. Even though the end of each sub core was described as transition points or critical junctions, they were simultaneously consequences as well. The three processes comprising *orientating,* namely dealing with pre-diagnostic manifestations, digesting the diagnosis and investigating the basics, resulted in the consequence of *realising the salient implications*. Grasping implications of the diagnosis then gave rise to the next action strategy in the process, namely fighting the disease. The same dynamics are applicable to the stage of *fighting back*. Having fought CMT extensively over time resulted in the consequence of *accepting reality*, which in turn led to the last action strategy, *optimising*.

Getting back to the overall outcomes of *engaging with CMT*, it was stated in the beginning of the chapter that an acceptable outcome amounted to enhanced well-being or some other outcome variable as determined by the data. However, the word "enhanced" proved to be rather problematic. It was for instance very difficult for participants to comment about enhanced and improved well-being because their condition deteriorated over the years. In other words, there were problems with baseline comparison. Furthermore, the rate and extent of deterioration varied over time within and between individuals, necessitating continuous readjustment. Most participants, however, were able to subjectively say if they felt they were faring well under the conditions or not. Put differently, they were more comfortable to express their feelings in these terms. Faring well, or just *wellness*, is therefore considered a more suitable outcome variable.

To the majority of participants, the gist of *engaging with CMT* amounted to the element of struggling and fighting against the disease's manifestations until the bitter end. Participant's interview responses were for example permeated with statements that they "refuse to sit down or to lie down", that they will tenaciously persevere until the bitter end and that they "refuse to give up". Linked to this fighting spirit was the fact that all participants except one fiercely resisted being regarded as disabled. By this is not implied that coping with CMT had been easy.

Actually, the contrary is true. The first quotation on the next page is an indication of just how tough it was for some.

However, the fighting spirit, the mental energy and drive to persevere and survive stood out and were reflected more or less in all the sub strategies of *engaging with CMT*. These may be summarised by the concept *tenacious holding on,* which was intertwined with the CMT-affected person's perception of whether they were faring well or not. Tenacious hanging on simply meant not succumbing to the disease by giving up, feeling sorry for oneself and diminishing the quest for living well or faring well. It referred to a mental attitude, the spirit to continuously engage with the disease and to hold on to "normal" ways for as long as possible. Most importantly, not to lapse into debilitating depression or hopelessness.

This extensive component of *engaging with CMT* referred more, but not entirely, to the first leg of the core problem, the part that had to do with the handling of the disease's symptoms and effects that are more tangible. It encompassed dealing with the myriad practical day-to-day problems and challenges experienced as a result of living with CMT. For many, this embodied the gist of living with CMT, reflecting continuous action right at the coalface. This is the upper portion of the triangular sub- problems depicted in Figure 4.1. None of the participants really failed in this area, although the degree of perceived success differed between people. When the majority talked about them faring well and having an acceptable quality of life, they were inclined to refer to this component. The disease's practical manifestations impacted greatly on daily living and quality of life; managing to cope on this frontier for most meant that they were faring well.

Two extremes are presented to serve as illustrations of outcomes. The first came alarmingly close to being a negative outcome. The individual is presently managing fairly well. It is also an indication of how serious the disease may affect people emotionally. The person claimed to have had depression superimposed on CMT, but it was impossible for me to separate the two. The second is an individual with very prominent symptoms, such as profound drop

foot and high stepping gait, who, having engaged with CMT over a long period, has almost transcended the disease and now does not see it as such a major threat:

1. Ek het ook al partykeer baie naby aan selfmoord gekom – ek het eendag met die haelgeweer hier onder my ken gesit hier buitekant – op die ou end het my moed my begewe – as ek nou terugkyk is ek dankbaar – ek sou my vrou en kind 'n onguns gedoen het en die werk van die Here baie skade aangedoen het.
At times, I also came very close to committing suicide. One day I sat outside with the shotgun pointing to my chin. In the end, my courage failed me. When I look back, I am grateful. I would have done my wife and child a disservice and would have caused a lot of damage to the work of the Lord.

2. *My advice to somebody with CMT is to never have a biopsy. He can live a good life, CMT only has a high nuisance value, but it is not like having Duchenne (a type of muscular dystrophy) where you have a very short life span and are suffocated. It's just the distal muscles that are affected. I think he should be fine; he can have children, but must be aware that they can inherit it. If they do, there are far worse things to inherit than CMT. One can live a normal life with CMT. Just a bit slower. But it is definitely a nuisance to have.*

Two other principal outcomes referred to the unpredictable component of the core problem, those that had to do with how participants managed uncertainty on the two frontiers of atrophying and having children. They refer to the bottom two problems of the triangular problem depiction in Figure 4 .1, and amounted to the following:

1) To what extent did *engaging with CMT* succeed in reducing stress and anxiety that were due to atrophying? Put differently, did these people succeed in coming to terms with the threat of deterioration?

2) To what extent did *engaging with CMT* alleviate distress on the two issues of inheritance? These were the uncertainty if one had passed the CMT gene to one's descendants and, if indeed so, to what extent have they (the parents) made peace with it?

In the sub strategy of managing worst-case scenarios, it was stated that most strategies of *engaging with CMT* in one way or the other addressed the pervasive

structural condition of deterioration. Strategies and actions such as wrestling with worst case scenarios, persevering, empowering, exercising, getting helping aids and broadening perspectives, to mention a few, all aimed directly or indirectly to influence the course of deterioration or to limit the damage caused by it. The latter included coming to terms with it and reducing undue anxiety. Did *engaging with CMT* accomplish this? The answer is no, not fully, for the majority of participants. Despite the fact that CMT-affected people did not always focus, or think about, deterioration in the context of their busy lives, the possibility of becoming dependent due to atrophying was still prominent in the back of their minds. As such, it evoked chronic anxiety that surfaced under certain conditions, for example when atrophying intruded into consciousness. With the exception of one participant and two borderline cases, images of becoming wheelchair bound were typically perceived as a horror scenario, or were repressed. It nevertheless evoked intense fear, despite people having passed through the three stages of *engaging with CMT*.

Even though *engaging with CMT* did not fully succeed in eliminating stress about undue deterioration, it nevertheless had definite positive consequences as well. In fact, it proved to have been a constructive endeavour. The core strategy especially enabled CMT-affected individuals to experience some degree of control over the disease, including the course thereof. The control aspect was present in all three stages, even in optimising, where cognitive-emotional strategies prevailed. This perceived control indeed resulted in some reduction in anxiety, although the exact amount cannot be established with qualitative data.

In addition, at the end of the *fighting back* stage participants did accept the reality that their strategies of resisting and combatting have not been successful in halting the weakening process, suggesting that they have to a limited extent come to terms with the atrophying process. The fact remains that it still evoked undue anxiety. Two interview snippets are presented to illustrate a negative outcome (no. 1) and a positive one (no. 2) in the context of deterioration:

1. *I just worry if I live too long, what will happen then. Hopefully God will spare me that one. I may have to walk with sticks and worry about things. When you get old, your kids don't want to look after you any more. I don't want to think about that too much, but that's how I feel about it. To have to be dependent on someone.... I prefer not to think about a wheelchair. Hopefully I will die before then.*

2. I can't actually visualise that I would be in a wheelchair. I can't actually picture it, it's like that would never happen to me, whereas realistically it could. And I think the possibility with me is increased. If it happens, it happens. Look, if I am in a wheelchair it will probably be the most high tech one (laughs). It will have to be.

Regarding the uncertainty about the CMT status of descendants, the indicators of a successful outcome corresponded with those discussed in deteriorating. They were "manageable anxiety" and "coming to terms". Whereas most strategies of *engaging with CMT* somehow had a bearing on deterioration, it was a different matter with the inheritance concerns. Not many strategies bar those discussed under *dealing with descendant's CMT status* addressed the inheritance concerns directly. Examples of those that directly or indirectly did, were investigating the basics, empowering the self, empowering the spiritual aspect and the cognitive strategy of broadening one's perspective. Having fewer strategies did not mean that the strategies were particularly ineffective. On the contrary, in about half of the participants, these strategies did reduce subjective anxiety about the possibility of the children having inherited CMT.

Naturally, the latter statement also means that, in the other half of the cases, the parent's strategies and actions were not successful. Once again, two interview excerpts to illustrate, first a positive outcome and second a negative one:

1. Ja, ek het vrede gemaak daarmee as hy CMT het. Ek het dit en ek meen, het dit my nou 'n slegter persoon gemaak? Hy sal dalk probleme op skool hê dat hy nie aan sport kan deelneem nie, maar dis al.

Yes, I have made peace with it if he (the child) has CMT. I have it and I mean did it make me a worse person? He may have problems at school because he cannot participate in sport, but that is all.

2. No. I won't have any more children. I'll stop it here. It's not nice to see this kind of thing. When I see my child, I feel for him, I really feel for him. He doesn't talk about this thing so much. When I bring it up, he brushes it off. I don't know how he'll deal with somebody else. I would like to know what is going on in his mind. I love babies, I love children, but I'll stop it right here.

An interesting phenomenon could be detected in instances where parents knew for sure that their descendants, both children and grandchildren, had inherited CMT from them. Under these conditions, all parents in this study to a lesser or larger extent were tormented by feelings of guilt. Partially because their options for helping the CMT-affected child were rather limited (there is, for example, no known cure for the disease) parents re-entered the process of *engaging with CMT* by employing and managing the process in the life of the child. The intensity of the involvement was, however, not necessarily as high as their own.

Under these conditions, and having gone through the process themselves, most sub strategies of *orientating* were either skipped or very briefly engaged by parents. Actually, only *digesting the diagnosis* were of prominence and took the form of information provider and general support to the affected child and his or her parent if it was a grandchild. The sub strategy of *fighting back* was engaged almost in its entirety with differences in accent here and there. *Optimising*, in particular *optimising own interest* (the child's) also featured, notwithstanding the child's young age. The following interview passage illustrates a CMT-mother applying *empowering the body* in her children:

Dr .....(name of doctor) felt that all my children have it (CMT) in different degrees. My one child plays a lot of sport and loves his soccer, the other one does karate. It is important; I need them to keep up the physical activity. Unfortunately, I did not do the same with all my children.

General involvement in the life of an affected grandchild, as well as the experience of pain, is illustrated by the following grandfather:

My granddaughter participated in dancing, but she got kicked out of the dance group recently because of her CMT. Other parents had the complaint that she allegedly was holding the group back. I am so very, very sorry for her; really, she enjoyed it so much. Then the group did not win the competition anyway.

To summarise, the core strategy of *engaging with CMT* that affected people employed to resolve their main concern, *dealing with unpredictable CMT manifestations*, did not produce one cut and dried positive outcome, but instead mixed consequences. Launching and managing strategies to persevere and deal with the disease's practical manifestations, for the most part, had a positive outcome and resulted in wellness. The vast majority of participants managed to subjectively live with the disease rather efficiently in this context, by which it is not implied that it was easy going. In this way the core strategy of engaging with CMT did indeed enable the CMT-affected to achieve and maintain an acceptable, if not above average, degree of well-being/quality of life.

On the other hand, coming to terms with the threat of profound deterioration, in particular to the extent where independent functioning was seriously threatened, was not obtained by the majority of participants by way of *engaging with CMT*. Regarding the inheritance concerns, only about 50% of people with CMT came to terms with it by way of the core strategy engaging with CMT. The rest still experienced fear and/or agony. In view of these two negative outcomes, wellness as an outcome was adjusted to "qualified wellness". These outcomes, as well as the resultant cyclic re-entering of *engaging with CMT,* are depicted in **Figure 4.1**.

Before embarking upon the contextualisation and integration of the identified grounded theory with the relevant literature (the next chapter), the results pertaining to verification measures will be discussed.

PART 2

VERIFICATION

Overview

As discussed in Chapter 3, Glaser (1978; 1992) is decisive that verification has no place in grounded theory. In fact, he finds it unacceptable that Strauss and Corbin (1990) attempt to apply the canons for judging quantitative research, such as reproducibility and generalisability, to grounded theory. He directs our attention to the dynamic nature of theory, the fact that it is fluid and changeable in time and space. He maintains that the criteria provided in Discovery of Grounded Theory (1967) are all that are necessary. These include fit, relevance, and that the theory must work, as well as be modifiable (Glaser, 1978; 1992).

Despite Glaser's strong feelings in this regard, I decided, nevertheless, to verify my findings. The first reason for my desire to verify stems from my own position in this study, in which I occupy the dual role of researcher and participant. Being both researcher and participant simultaneously, opens up the possibility of negative bias. The second reason is the fact that the present grounded theory is entirely new, and verifying my findings, mainly by triangulation with other methods and means, may be meaningful. Since verification is not the prime focus of this study, these measures are only applied to a limited degree, mainly in the light of Glaser's position with regard to my need to verify my newly formulated grounded theory on *engaging with CMT*.

One influential model that addresses verification in qualitative research was developed in the 1980s by Guba and Lincoln, who substituted the quantitative measures of reliability and validity with trustworthiness, a new formulation more applicable to qualitative research. Trustworthiness comprises four subdivisions or criteria, namely credibility, transferability, dependability, and confirmability (Guba & Lincoln, 1994; Krefting, 1991; Lincoln & Guba, 1985; Morse et al., 2002). Even though formulated in order to move away from quantitative criteria, Lincoln and Guba (1985, p.300), for explanatory purposes, state that credibility,

transferability, dependability, and confirmability are the qualitative equivalents to quantitative research's internal validity, external validity, reliability and objectivity respectively. Lincoln and Guba (1985) subsequently formulated a plethora of strategies, which may be employed to satisfy each of their four criteria. Only a limited number of these were applied to this research, the most prominent being triangulation of methods.

Concerning *credibility*, or truth-value, of this research project, three of the strategies formulated by Lincoln and Guba (1985) were focused on. They are triangulation, member checking, and enhancement of researcher skills.

Enhancement of researcher skills

Beginning with the latter, researcher skills were enhanced by way of an orientation session regarding data coding skills. In this session, I coded the first interview script together with an outsider in order to eliminate weaknesses and discrepancies as far as possible. It almost goes beyond saying that I also studied numerous examples of coding and categorising in grounded theory studies. Another strategy, perhaps with a different accent, is that most coding, especially open coding, was checked by a psychometrist doing qualitative content analysis, with similar data coding techniques.

Member checking

The strategy of member checking "whereby data, analytic categories, interpretations and conclusions are tested with members of those stake holding groups from whom the data were originally collected" (Lincoln & Guba, 1985, p.314), were implemented as far as possible, notwithstanding the vast geographical distances between me and most of the participants. According to Lincoln and Guba (1985) member checking is both informal and formal, and occurs on a continuous basis. All data- analysis constructions, such as interview transcripts, summary sheets and even the final report should ideally be taken back to all participants in order to establish to what extent they regard the researcher's reconstructions as "representative", as well as to react to them if desired. Due to the already mentioned geographical distances, member checking was only applied

partially. The interview transcripts were only submitted to the Cape Town participants. All signed the transcripts as being a true reflection of their discourse. A copy of the results chapter, excluding the verification part, was mailed to approximately 50% of the participants. They were not entirely selected at random due to the ethical aspect reported by Krefting (1991, p.219): "Researchers must be selective about which informants are involved in member checking. Often, informants are not conscious of the information discovered by the researcher and may become troubled if made aware of it". She continues that anything potentially harmful to their well-being, be withheld. All participants who read the results chapter indicated that they identified with the contents of the chapter, including the formulated grounded theory, and that it is a true reflection of their realities as shared with me.

Triangulation

Explaining *triangulation* in social research, Neuman (2000, p.124) states: "...it is better to look at something from several angles than to look at it mainly one-way", and Krefting (1991) regards it as a powerful strategy to enhance credibility and quality of research in general. Denzin (1978), in Lincoln and Guba (1985), identified four main types of triangulation: sources, methods, investigators and theories. In this project, only triangulation of methods, "in which data collected by various means are compared" (Krefting, 1991, p.219), was used. Five psychometric questionnaires and one biographical questionnaire, described in the methods chapter, were completed by all participants.. They are the Brief Cope (assesses coping strategies), the WHOQOL-BREF (assesses quality of life, including health facets), the Beck Depression Inventory (BDI) (subjective level of depression), the Perceived Stress Scale (PSS) (subjective appraisal of general life stress) and the Rosenberg Self Esteem Scale (RSE) (self-esteem).

Juxtaposing the quantitative, psychometric findings to the BSPP, or core process of *engaging with CMT* proved to be problematic. The most salient problem amounted to the fact that participants were in different stages of the core process, either *orientating, fighting back* or *optimising*. In addition, some participants had also temporarily revisited previous stages due to contingencies in the disease's

trajectory. It is likely that the prominence of the constructs assessed by the psychometric instruments may vary between stages. For example, coping strategies such as planning, acceptance and the use of instrumental support, assessed by the Brief Cope, psychological well-being (BREF), perceived depression (Beck) and so forth might come to the fore differently in the three stages. In addition, it cannot be established with certainty which stage each participant was referring to while answering the questionnaires. True scientific comparisons between the two types of data are therefore not attainable.

If the questionnaires had been administered separately for each stage of the core process after having established the exact stage each participant was in at the time, one could have perhaps compared the two types of data more precisely and comprehensively. Since that, however, was not the case, average scores for the 11 participants would be used to compare the data from the questionnaires with the core process *engaging with CMT*. A summary of the averages of all participants' questionnaire results are shown in **Table 4.1.** A comparison between the questionnaire results and the core process of *engaging with CMT* will susequently be discussed.

Explanation of the psychometric scores in Table 4.1

WHOQOL-BREF

Domain 1 (Physical health)

The fact that participants scored the lowest in domain 1 (physical health) may be interpreted as supporting certain qualitative findings. CMT primarily affects the body, for instance cramps, muscle weakness, pain, fatigue, impaired balance, numbness and so forth. However, other than these disease specific manifestations, general health may actually be good, which may explain the average score of almost 60.

Domain 2 (Psychological well-being)

The second lowest average score was obtained in domain 2 (psychological). Well-being in the psychological domain of people with CMT may be adversely

Table 4.1: Summary of psychometric scores (averages)

	Average score (N=11)	Theoretical maximum
WHOQOL-BREF		
Domain 1 Physical health	59,9	100
Domain 2 Psychological	67,0	100
Domain 3 Social Relations	72,6	100
Domain 4 Environmental	73,3	100
General Health facet	3,3	5
Overall Quality of Life	3,1	5
BDI	5,6	-
PSS	15,7	40
RSE	20,9	30
BRIEF COPE		
Self distraction	4,0	8
Active coping	6,5	8
Denial	2,2	8
Substance use	2,0	8
Use of emotional support	5,3	8
Use of instrumental support	4,5	8
Behavioural disengagement	2,3	8
Venting	4,4	8
Positive reframing	6,1	8
Planning	6,0	8
Humour	4.6	8
Acceptance	7,7	8
Religion	6,5	8
Self blame	4,0	8

Key: WHOQOL-BREF = World Health Organisation Quality of Life Assessment - Short Form; BDI = Beck Depression Inventory; PSS = Perceived Stress Scale; RSE = Rosenberg Self Esteem Scale; Brief Cope = Carver Brief Cope

affected by experiences of uncertainty, frustrations, anxieties, impaired body image and self-esteem, to mention a few. However, participants had been living with the disease for decades and many probably would have adapted better to it, generally speaking, than recently diagnosed people, which may partly explain the relatively high average score of 67,0. In addition, all participants felt that they were faring well in terms of managing the practical effects and manifestations of the disease, an aspect of the core concern to which they allocated a high priority in terms of subjective well-being (as delineated earlier in this chapter).

Domain 3 (Social Relations)

The average score of 72, 6 in domain 3 (social) indicate that CMT did not markedly impair social relations at that stage of their lives (the WHOQOL-BREF assesses quality of life during the two weeks prior to the test date), which largely corresponds with the qualitative findings. Most participants did not report the experiencing of serious, debilitating social problems at the time of the interview.

Domain 4 (Environmental Aspects)

The environmental domain yielding the highest score is also in agreement with *engaging with CMT*. One reason for this finding may be a lack of enough participants from traditionally marginalised communities, who may be particularly vulnerable to the typical problems relating to this domain, such as transport problems, diminished safety and security, poverty and poor accessibility to health services.

General health

Participants rarely discussed serious health problems of a general (non-CMT related) nature during the qualitative interviews. A possible explanation for the above average general health score of 3, 1 may therefore be that, in responding to this item, participants' focus was not on their CMT per se, but instead on their health in general.

Overall quality of life

The overall quality of life score is based on a single question that asks people to rate their quality of life. In view of participant's rather high age, which ranged from 38 to 70 (average 48,6) and the many years of engaging with the disease (in most cases since early chidhood), the slightly above average score may reflect general life experience and experience in dealing with CMT. All people except one (a widow) were married at the time of the interview with, in general, adequate support systems. It may also be that many participants are in the later stages of engaging with CMT. The stage of optimising is, amongst others, characterised by a "making the most of" inclination where the focus is on maximising ones quality of life with the disease. The explanation in domain 2 regarding the management of the practical effects and manifestations of the disease (see above), is also applicable here.

Even though it is impossible to establish with certainty to what extent participant's grappling with deterioration and inheritance issues (the two least successful outcomes of engaging with CMT) is reflected in the psychological well-being and quality of life scores, the possibility exist that they may be largely responsible for these scores not being higher than those obtained.

Beck depression inventory (BDI)

Both the average BDI and PSS scores do not reflect the presence of undue quantities of the constructs that they assess. In fact, a BDI score ranging between five and nine, as is the case here, is considered normal fluctuations in mood (Devilly, 2005). Only one participant resorted in a higher category, which is the next one up (mild to moderate depression). The low BDI levels of depression are in line with the grounded theory findings. Serious manifestations of depression, as well as a related concept of helplessness, did not emerge prominently in this study.

However, this is not to say that feelings of depression and helplessness were entirely absent; on the contrary, participants did experience these moods to some

extent from time to time. It was however more a case of mild, transient episodes as people adjust, or re-adjust, to the disease's manifestations. Said differently, these moods were generally not of a chronic nature and occurred mainly in response to salient developments/changes in the disease's trajectory, including its direct and indirect effects. Examples from the qualitative data include noticing profound wasting away of muscles, as well as reduced ability to execute motor tasks such as writing, typing, playing musical instruments, manipulating eating utensils, playing ball games, driving a vehicle and walking long distances. In addition, as was mentioned elsewhere, many of these reactions may be explained from the perspective of normal mourning that people go through in response to loss, rather than being serious depression per se. For many, the core strategy of *engaging with CMT* very much amounted to an action orientation, rather than passivity/unmotivatedness, one of the profound symptoms of depression (Nevid, Rathus & Greene, 1997).

Perceived stress scale (PSS)

Like the BDI, the relatively low PSS score also supports the qualitative findings of this research. On rare occasion did participants report excessive and debilitating levels of global, general stress, at least not in the one month prior to the test's administration, which is what the PSS assesses. A degree of stress was indeed present in the lives of participants, which is perhaps to be expected. In the words of Northern (2000, p.30) "CMT can be a stressor, either at the time it is first diagnosed, or later, when particular aspects of the condition begin to affect the individual". Stress related to specific stressors, however, such as atrophying and the CMT status of one's descendants, is indeed encountered in the qualitative data and was not detected by the PSS. This may be understandable, since the PSS does not purport to assess specific stressors.

Rosenberg self esteem inventory (RSE)

At first glance, some may regard the RSE score of 20,9 out of 30, which is the theoretical maximum that is possible to obtain, as rather high, especially in view of those qualitative findings that demonstrated CMT-affected people's self to be

vulnerable to aversive social conditions such as public ridicule and stigmatisation. These negative influences are not limited to childhood (for example debilitating school experiences), but also to adulthood in the appropriate aversive contexts, such as the work environment. In addition, self-concept is intrinsically related to body image (Livneh & Antonak, 1997) and, with bodies atrophying, pressure on the self may resurface continuously, given that neuromuscular conditions such as CMT have a high visibility.

One possible explanation for the self-esteem score of 20, 9 concerns the measuring instrument. Typically, measures of self-esteem reflect a strong trend towards positive self-evaluation, that is to say, scores cluster on the positive side (above the theoretical mean of 15 out of 30) (Schmitt & Allik, 2005). Schmitt and Allik (2005) found that this phenomenon is rather universal: in their study they, for example, encountered it to a lesser or larger extent in 53 diverse cultural settings.

However, another possible and seemingly valid explanation could be the fact that CMT-affected people undertook considerable work on their self-concepts throughout the entire process of *engaging with CMT*, even though most work took place during the stage of fighting back. In other words, participants' persistent efforts in reorganising their self-concepts have been largely successful, not only in the short term, but indeed also over the long-term.

Linked to above-mentioned, but also an explanation in its own right, concerns the rather high age of participants (average 48, 6 years, range 38-70). This amounts to the fact that, as the years go by and people grow older, their maturity levels and coping strategies might have improved to such an extent that they are able to better endure or counter potential debilitating influences to their self-esteem, such as ridicule, stigmatisation and poor body image. In addition to this enhanced hardiness and better coping skills against the debilitating influences, more effective coping usually results in more coping successes, thereby enhancing self-image and self-efficacy.

It is equally possible that many, if not most, participants are in the stage of optimising (the last stage of *engaging with CMT*), where the broadening of perspective and concomitant insight regarding their lives with CMT are focused on. Activities that reflect existential values, such as contributing and appreciating loved ones, are encountered here more than elsewhere in *engaging with CM*T. People in optimising have accepted the reality that their fighting the disease has resulted in rather limited success (in other words, it might have been rather futile), and that optimising their life with the disease in the years ahead is the proper way to proceed. It is probably not unreasonable to expect that the enhanced insight and greater inner peace, as described in this paragraph, may be conducive to a more "healthy" self-esteem than before (in the two earlier stages). In fact, there may even be rather drastic gains in self-esteem, because, amongst others, CMT-affected people in this advanced stage often realise that ill or disabled people are by no means inferior humans.

To conclude then, the abovementioned delineation indicates that it is likely that the RSE score of 20, 9 is indeed an accurate assessment of participant's self-esteem and thus support the qualitative results.

Brief Cope Questionnaire

For practical purposes, as well as to ease explanation, the 14 Cope scales will be regarded as coping responses or strategies. In this section, average scores for each of the scales will be used. The theoretical maximum attainable on each scale or strategy is 8.

Active coping

The active coping score of 6, 5 is, together with religion, the second-highest score reflected in the Brief Cope results. According to Carver et al. (1989), active coping entails increasing one's action taking and related efforts; it resembles problem focused coping as described by Lazarus and Folkman (1984). The high active coping score supports a fundamental finding obtained in the grounded theory analysis. This amounts to participants' inclination not to rest on their laurels, but instead to act and engage with the disease, to fight its manifestations

and to make the most of their situation. Amongst the myriad of action strategies are exercising, taking medication, practising religion, gathering information and self-development. The Brief Cope's high active coping score confirms these action-taking behaviours. In addition, the active coping orientation is intertwined with the strategy to fight back and to never surrender to CMT.

Behavioural disengagement

Behavioural disengagement amounts to giving up trying to cope (Carver, 1997). The low Brief Cope score (2, 3) confirms the qualitative finding that participants tenaciously persevered with a strategy to never surrender to the disease. It also links to the high active coping score discussed above.

Planning

The rather high planning score of 6, 0 corresponds to the grounded theory data, where a planning for the future-focus emerged rather strongly, especially in the context of efforts to gain control. Participants typically planned ahead in order to provide for the possibility of profound deterioration, which includes plans for retirement. Once again, the high planning score confirms the problem focused orientation that was explained above under active coping.

Acceptance and denial

Acceptance is the highest Brief Cope score (7, 7), and should be read in conjunction with the low denial score (2, 2). These two scores correspond fully to the qualitative findings. Perhaps to be expected in congenital, progressive neuromuscular diseases such as CMT, and indeed reflected in the qualitative data, substantial denial of the disease's existence by the affected is unlikely. The opposite, acceptance, is likely, because there is no real escape; CMT is incurable.

Religion

The Brief Cope score of 6,5 for religion is the second-highest score (shared with active coping) to acceptance (7,7). In the qualitative investigation, religion emerged as equally prominent, that is to say, as a very important factor and key

strategy in the lives of people with CMT. Religion as a strategy particularly came to the fore during the stages of *fighting back* and *optimising*.

Humour

Even though the humour score of 4, 6 are just above the average of 4, 0, it may nevertheless be seen as corresponding to humour as encountered in the qualitative data. Humour surfaced strongest in the later stages of the core process and were the most noticeable in the stage of optimising.

Self-distraction

This strategy yielded an average score of 4, 0. Self-distraction refers to actions that people employ in order to take their mind off stressors (for example, disease manifestations). It includes behaviours such as daydreaming, escaping through sleep and immersion in television (Carver, 1997; Carver et al., 1989). The Brief Cope score of four indicates that participants employed these strategies to an average extent. Although these behaviour patterns did emerge in the qualitative data, it cannot be established with certainty whether they were present to an average extent. Examples of daydreaming are where an affected individual dreamt about climbing a mountain with friends like in the old days, being able to dance with one's partner again and vividly dreaming about (impossible) sport achievements.

Self-blame

The self-blame score on the Brief Cope is an average of 4, 0. In the grounded theory findings, self-blame was reported mostly, but not exclusively, by those people who had passed the CMT gene to their children, or who suspected that they had done so. Like in self-distraction, it cannot be established with certainty whether it was present to an average extent or not in the qualitative data.

Use of instrumental support

Instrumental support [seeking advice, information and assistance (Carver et at., 1989)] yielded a score of 4, 5, which is just above average. The qualitative findings indicate that the utilising of this type of support tended to occur more, but

by no means exclusively, in the earlier stages of *engaging with CMT*. Since most participants have lived with CMT for many years since diagnosis and have probably adapted to it to some extent (as described elsewhere), their use of instrumental support might have gradually receded over the years. Nevertheless, in view of the myriad problems engendered by living with CMT, it is probably unlikely that the use of instrumental support will recede below average levels, even in the later stages of *engaging with CMT*. In view of this aspect, as well as the fact the Brief Cope assessed coping in the three months prior to the date of testing, the score of 4, 5 is considered to be supportive of the qualitative results.

Use of emotional support

The Brief Cope score for the use of emotional support (getting moral support, understanding and sympathy [Carver et at., 1989]) is 5, 3, which is slightly higher than instrumental support (4, 5). This tendency also appears to manifest in the qualitative data, especially regarding the first two (moral support and understanding). It was extremely difficult for me to establish in the qualitative data whether participants (in the applicable context) were seeking sympathy or recounting reality as they experienced it. Corresponding with the use of instrumental support, emotional support appears to have receded gradually over the years. It was most prominent in the time period following diagnosis, in other words, during the stage of orientating. Seeking emotional support also occurred more prominently following turning points or crises in the disease's trajectory, such as profound atrophy, an increase in pain or cramps, and so forth.

Positive reframing

The rather high Brief Cope score of 6, 1 is in line with the qualitative finding regarding this strategy. The cognitive reframing of stressful events in more positive terms is encountered throughout engaging with CMT. However, it is encountered more in the later stages of the core process than earlier, probably because of greater insight as people gradually "matured" into the acceptance of reality, the critical junction at the end of the stage of fighting back.

Venting

The Brief Cope score of 4, 4 indicate that participants more or less to an average extent (slightly above) ventilated their feelings when they experienced distress, which is what venting in the Brief Cope assesses (Carver et al., 1989). Evidence, or indicators, of this behaviour are present in the qualitative data, but the exact extent thereof could not be determined by studying the verbal communications contained in the transcribed interviews.

Substance use

The substance use score of 2, 0 is the lowest of all on the Brief Cope. This corresponds strongly to the qualitative findings. None of the participants engaged in debilitating substance use and moderation was the norm.

As the abovementioned delineation indicates, the vast majority of the Cope's 14 strategies remarkably correspond to the qualitative "equivalent" of those particular strategies. However, considerably more (different) strategies than those measured by the Brief Cope, emerged in the grounded theory analysis.

In sum, it has been shown that the results of triangulation via psychometric questionnaires largely corresponded to and thus supported the qualitative results, as far as the particular constructs that they assess are concerned. These instruments, however, assessed a very limited scope of variables operating in the grounded theory, but the sample they did assess, proved helpful for triangulation purposes, that is to say, to view the qualitative findings from a different perspective with a view of enhancing quality and credibility.

CHAPTER 5

DISCUSSION

This study culminates in the formulation of a substantive grounded theory about how people with the neuromuscular condition, CMT, manage the effects of the disease in order to continuously adapt to it and augment their well-being. The core strategy through which this is accomplished, emerged as *engaging with Charcot-Marie-Tooth disease*. This, in turn, comprises three mostly sequential stages that CMT-affected people move through as they continuously adapt to and manage the manifestations of the disease. The three stages, namely *orientating, fighting back* and *optimising*, comprise various sub-strategies and actions of the core. By way of these formulations, the two research questions are answered: (1) in what areas, and how, does CMT affect the lives of those who have the disease, according to their own frame of reference? (2) how do people who have CMT manage the manifestations, effects, implications and challenges that stem from this disease, in order to augment their well-being?

In this chapter, the qualitative results will be contextualised within the applicable scholarly literature. This task will be accomplished in two sections. In Part one, *engaging with CMT* will be compared to a synthesised stage-model of adaptation to chronic illness and disability (CID). Part 2, which comprises the larger part of the chapter, will focus on contextualising the three stages of *engaging with CMT* within the existing scholarly literature. The chapter ends with a conclusion, reflection and recommendations for future research.

PART ONE

COMPARING ENGAGING WITH CMT TO A SYNTHESISED STAGE MODEL OF ADAPTATION TO CHRONIC ILLNESS AND DISABILITY

BACKGROUND

Because CMT is a medical condition, chronic and not acute in nature, the formulated theory of this study may be positioned in the broad research realm of psychosocial adaptation to chronic illness and disability.

Research on the adaptation to chronic illness and disability (CID) has predominantly focused on acquired and adventitious illnesses and disabilities. Vast arrays of conditions were studied, for example, life threatening diseases (e.g. cancer, heart disease) and non-life threatening conditions (e.g. multiple sclerosis, spinal cord injury, rheumatoid arthritis, diabetes). Certain ones are of sudden onset (e.g. spinal cord injury, amputation, myocardial infarction), whilst other, more chronic conditions have a gradual, insidious onset (e.g. neurological conditions, rheumatoid arthritis). Furthermore, certain conditions have a relatively stable course following initial trauma or onset, whilst others have an unstable course, characterised by exacerbation and remission over time (Livneh et al., 2004; Sidell, 1997).

Considerable uncertainty prevails regarding the etiology of many, if not most, of these chronic conditions. Often the cause is unknown and the exact role of inheritance is unclear. Some researchers reason that a genetic predisposition for particular diseases is inherited, which is then activated by adverse conditions. Examples are chronic conditions such as rheumatoid arthritis, asthma, diabetes and multiple sclerosis (Youngson, 1995).

In contrast to the large body of research on adaptation to acquired conditions, psychosocial adaptation to congenital diseases and disabilities, and in particular,

neuromuscular conditions, which includes CMT, have not even remotely received the same amount of research attention. Less than five qualitative research studies on the experiences and management of CMT and muscular dystrophy respectively could be traced. They mostly comprised interviews and the extraction of themes. No grounded theory study pertaining to living with congenital neuromuscular diseases and CMT in particular could be traced. The paucity of research was also encountered on the more practical level of the coping strategies or styles that people adopt to deal with the effects of their congenital neuromuscular diseases.

Livneh et al. (2004, p.413) refers to the coping strategies mentioned above, as "Coping strategies in the service of adaptation to CID". People adopt these strategies in order to cope with stress that is engendered by their medical condition: effects such as pain, handicap and discomfort. As was expounded in Chapter 1, a number of coping ways and styles have been studied. The two major categories are: (1) problem (or task) orientated and approach strategies on the one hand, and (2) emotion orientated and avoidant strategies on the other. According to various studies (De Ridder & Scheurs, 2001; De Ridder, Scheurs & Bensing, 1998; Livneh et al., 2004) the former tends to be more adaptive in the long run than the latter. An example of an exception is uncontrollable diseases. Here, certain variants of emotion/cognitive type coping, for example suppression, may be the better route, rather than persisting with futile attempts to exercise control in the situation.

Getting back to the broader adaptation to CID, Livneh and Antonak (1997, p.424), having reviewed the seminal works on this topic, describe the process as follows: "… adaptation to chronic illness and disability is a dynamic, gradually unfolding, and progressive process through which the individual strives to reach an optimal state of person-environment congruence referred to as adjustment." Adjustment is conceptualised as the final phase of the adaptation process, characterised by optimal adaptive levels of functioning. There is no uniformity in the use of this terminology, though, and some researchers, for instance Kendall and Buys (1998), refer to the process itself as adjustment.

Disagreement is not limited to terminology; it also exists regarding what exactly the adaptation process itself consists of. Over the years, psychosocial adaptation to CID has come to be conceptualised by theoreticians and researchers in one of two divergent ways: (1) the chronically ill's reactions to their disease follows a stable, linear sequence of phases, or stages, that occur over time, and (2) adaptation to CID amounts to a non-sequential and independent pattern of behaviour, which does not reflect stages or phases (Livneh & Antonak, 1997). A rather well-known stage model, adapted for use in CID, is the one by Elizabeth Kübler-Ross (1969), which she had formulated to depict the stages, in the context of grief, that one passes through in the face of your imminent death or the loss of a loved one (Kendall & Buys,1998; Kübler-Ross, 1969; Loggenberg, 2006).

The grounded theory, which emerged from this study, is characterised by the basic social and psychological process (BSPP) of e*ngaging with CMT.* It resorts under the first grouping, and is therefore a stage model. It is a model depicting the (mostly) sequential stages that people with CMT pass through as they, on a continuous basis, adapt to and manage the effects of their disease. Technically speaking, it would be more correct to say that *engaging with CMT* entails phases and not stages. Some writers see the former as pertaining to partially overlapping, non-exclusive groupings of reactions and the latter to discrete, exclusive groupings. In this regard, I will follow the views of Parker, Schaller & Hansmann (2003, p.235), by stating that this distinction is largely inconsequential to this study, and that the two terms will therefore be used interchangeably.

No stage theory that was constructed using grounded theory methodology on the adaptation to and/or management of congenital peripheral neuropathy, including CMT, could be traced. There exist more general "stage grounded theory studies" on adaptation to CID, though. One example is by Charmaz (1995), who identified three stages that affected people proceed through as they adapt to impairment. These are experiencing an altered body, changing one's future identity and surrendering to the sick body. A second, more implicit, example is by Corbin and Strauss (1988), who identified illness trajectory phases, depending on the particular case, as acute, comeback, stable, unstable, deteriorating and dying. Their

delineation of biographical processes to be completed after disruption of the biography by a CID, may also amount to phases. They are contextualising, coming to terms, reconstituting identity and recasting biography.

THE STAGES

Regarding the quest for a seminal work on adaptation to CID, Parker et al. (2003, p. 235) states: "by far the most comprehensive effort to review the literature on the psychosocial adaptation to CID and develop a model incorporating research findings has been accomplished by Livneh and Antonak (1997)". The latter work will therefore be the main source for Part one of this chapter.

In their comprehensive review of the research literature on psychosocial adaptation to CID, Livneh and Antonak (1997) presented an extensive synthesis of the different stages, as described by various researchers over many decades, which people with CID pass through in their adaptation to their conditions. A few of the researchers' work that they studied include Shontz (1965); Weller and Miller (1977); Devins and Seland (1987) and Antonak and Livneh (1995). They synthesised the stages most commonly encountered in the literature as *shock, anxiety, denial, depression, internalised anger, externalised hostility, acknowledgement* and, finally, *adjustment.*

In order to meaningfully compare the present identified theory to these stages, they will be briefly discussed. This will take the format of a short theoretical description in italics, based on Livneh and Antonak (1997), followed by a brief discussion and comparison to the present identified theory. For convenience sake, these stages will be called the synthesised stages.

Shock

Shock is the initial reaction to the onset of a sudden, severe physical injury (for example, spinal cord) or psychological trauma (e.g. the diagnosis of a life threatening disease such as cancer, or a neurological disorder). Feelings of being

overwhelmed, disbelief and cognitive disorganisation are characteristic (Livneh & Antonak, 1997).

Emery (1994) states that affected people, (and/or parents if it is a child) will inevitably experience reactions of shock and disbelief when they are informed of a diagnosis of muscular dystrophy. Questions such as "why me", "this cannot be true" and "no one else in the family has it" will typically surface. According to him, the shock reaction will still occur, even though they might have been suspicious that something has been seriously wrong for a very long time.

A shock reaction to the extent as described in the two preceding paragraphs was not encountered commonly in participants with CMT; in fact, only about one third reacted with shock. An equal amount of people experienced diagnosis as a relief, and the rest were concerned, but said that they would not describe their feelings at the time of diagnosis as shock in the true sense. Livneh and Antonak (1997, p.25) substantiate this finding: "shock may not be experienced at all, however, by people with a gradually deteriorating and uncertain medical condition". CMT is such a condition. They conclude that reactions of shock apply more to the sudden onset of traumatic conditions, such as myocardial infarction and life-threatening disease.

Another factor may be that the element of shock was dramatically diminished by the fact that participants had lived with the disease for many years before being officially diagnosed (average age of diagnosis = 29, median = 28). The disruptive nature of their symptoms over many years had sensitised them for the possibility of a medical condition. Said rather plaintively, they suspected that there might be a problem with their health because something had been causing serious problems for such a long time. An additional problem identified by Robinson (1988), also applies to CMT affected people. These individuals face the dilemma of other people not believing that something is medically wrong with them, and that they are "putting it on", hence the relief of discovering a legitimate medical reason for their symptoms (Robinson, 1988).

Anxiety

According to Livneh and Antonak (1997), anxiety is the typical reaction when the individual, and/or parents if it is a child, initially realise the reality of the magnitude of the traumatic event. Other sources of anxiety include uncertainty about the future, the unpredictability of the illness, fear of recurrence of symptoms, the threat of future lifestyle changes and anticipation of invasive procedures

Anxiety is commonly encountered in the data and findings of this research. It is, however, not a distinct stage as such, but instead occurs in varying degrees throughout all three stages, and even beyond in instances where resolution is not attained. All the above-mentioned sources of anxiety emerged during the grounded theory research. Anxiety about the uncertain future, which embraces many of the above sources, was a very common finding amongst people with CMT. Although this was rather more prominent at the time following diagnosis, it remained important throughout the entire process, especially in view of the unpredictable course of the disease. The specific sources of anxiety with regard to CMT include the following: (1) anxiety about the CMT status of children and grandchildren, (2) anxiety about becoming dependent on others and (3) anxiety about losing the ability to walk and become wheelchair bound.

The above findings are largely in agreement with Livneh and Antonak's (1997) view that anxiety and depression in chronic illness appear to be more future orientated, whilst traumatic event-related anxiety is more past orientated. The latter typically involve grieving for the loss of body parts or functions and cognitive abilities. The former largely entails fear of the unknown, that is to say, possible future suffering, which was clearly encountered in the findings of this study. In fact, the main sources of anxiety can be summarised as fear that the worst-case scenarios may become a reality. This amounts to fear that they may lose the ability to walk, end up sitting in a wheelchair full time, and fear whether their descendants did inherit CMT from them.

A useful addition to the above distinction may very well be that anxiety in chronic diseases, such as CMT, is of a more long-term and chronic nature, whereas

traumatic event related anxiety appears to be more acute, receding gradually after the incident. In CMT, any physical deterioration that people become aware of also causes escalation in anxiety, which is not encountered in the synthesised model.

Denial

Denial is a defence mechanism that the individual employs to protect him or herself against the grim reality of having an incurable, chronic condition, with all the concomitant negative implications. Minimisation of the disease is typical (Livneh & Antonak, 1997).

Denial, insofar as it is a designated stage on its own, is not encountered in participants with CMT. As a stage, it appears to be easier identifiable as a rather early reaction following disability. In so doing, the person typically endeavours to conceal his or her disability. Wright (1988) attributes people's efforts to conceal mainly to stemming "from the belief that having a disability makes the person less desirable, less good" (p. 118).

Elizabeth Kübler-Ross (1969) contends that denial may also serve a constructive purpose, in the sense that it functions as a buffer after traumatic news, allowing the person to collect him or herself and then to gradually implement other coping mechanisms. Denial is therefore not merely used to avoid the reality of the situation; it also serves the function of protecting the individual. It (denial) may take on many forms, for example believing that their case will be different, that they won't deteriorate as rapidly as others, rejecting the diagnosis, arguing with professionals, lack of cooperation and even a futile search for a miraculous cure (Sowell, 2000b; Loggenberg, 2006).

With the exception of rejecting the diagnosis, which did not occur at all, the above manifestations were to a limited degree encountered in a few participants with CMT. This applies to both past and present functioning. Examples are arguing with medical professionals and believing their case will be different. Only one person had denied his condition in the past when he was engaging with the stages of *orientating* and *fighting back*. At present, he has fully integrated the disease

into his self-concept. Minimisation of the seriousness of CMT was encountered in two people, although it is possible that they could have stated the realistic position, in other words that their condition was indeed not very serious.

An explanation for these findings may be that people with congenital conditions had been living with their disease since birth or the early onset thereof, which in the case of CMT is mostly during the first or second decades of life (Chance, 2001; Kedlaya, 2007; Northern, 2000). A lifetime of being subjected to the disease's manifestations resulted in most people realising that something was wrong. Under these conditions, the disease has been part of their self-concept since a very early age (Wright, 1983) and denial in the true sense of the word is therefore a rare occurrence. Charmaz (1991) points out that people who cannot define their condition as temporary and who actively "experience their altered bodies in their own worlds"(p.20), are unlikely to deny their chronic condition.

Depression

Upon realisation of the full impact, that is to say, the seriousness and implications of the CID, feelings of depression, helplessness and hopelessness are typically experienced. This reaction is often encountered amongst adventitiously impaired individuals, as opposed to congenital diseases. Depression is a reactive response of bereavement for lost body parts or functions (Livneh & Antonak, 1997).

Emery (1994), as well as Livneh and Antonak (1997), consider depression to be the typical reaction upon initial realisation of the implications of the disease, more or less at the time of diagnosis or soon thereafter. To a lesser extent, this is also the position regarding many of the participants in this study, especially regarding feelings of helplessness.

Depression, which at any stage during the adaptation process is of such a serious nature that it warrants clinical diagnosis, was encountered in one participant. He claimed that his depression existed for many years, and that he has had CMT superimposed upon his depression. However, he vividly remembers grappling with feelings of inferiority due to being physically weak, and failing in sport at

school (since his earliest years in primary school). He also struggled with poor body image, once again since his very early years. Since he was only diagnosed with CMT in his late-20s, the possibility exists that his claimed long-standing depression might have been related to feeling deficient due to CMT without realising it (having not received a diagnosis yet).

Another participant also experienced rather serious depression and feelings of hopelessness. Aside from these two rather serious cases, helplessness and depressive reactions of lesser dimensions are encountered throughout all the stages of *engaging with CMT*. Depression and anxiety are intertwined in the findings; the numerous uncertain implications regarding the course of the disease generated both anxiety and depression, as well as helplessness. Based on the findings, I agree with Livneh and Antonak (1997) when they say: "where a chronic illness is involved, anxiety and depression often share common ingredients…" (p.25).

Another facet that the above two researchers discuss under the depression stage, is bereavement, which is commonly encountered throughout the BSPP of *engaging with CMT*. They aptly state: "It (depression) is regarded as a reactive response of bereavement for the lost body part or function or of impending death or suffering" (p.21).

Sowell (2000b) states that **loss** is inherent to neuromuscular diseases. There are three types of losses that people with these diseases have to cope with: (1) physical losses, such as strength, balance and ability to walk, (2) lifestyle losses, including work or aspects of work, independency, hobbies and diverse pleasurable activities, and (3) interpersonal losses, such as crumbling relationships due to the disease. Sidell (1997) adds another important one, namely loss of perceived control. The realisation that the course of the disease is out of one's control, in many instances leads to feelings of powerlessness.

People grieve after most significant losses, not only health related ones. Many health experts believe that it is necessary to grieve after these losses in order to successfully adapt to them. Knowing in advance that loss of abilities will occur, as

in neuromuscular disease, does not protect the person from feelings of grief when the loss actually occurs. The losses due to neuromuscular disease are akin to losing a part of yourself, that is to say, your identity. In order to survive, the self must be redefined, which is partly accomplished via the grieving process (Sowell, 2000b).

In CMT, sorrow, helplessness and depression are also observed in parents who had passed the CMT gene on to their children or grandchildren in further generations.

Internalised anger

Anger in this context refers to anger directed at the self and includes self-blame, self-directed resentment and bitterness. Often, the person attributes the disease to own "wrong doing.", for example, wrong health practices, past transgressions, and so forth (Livneh & Antonak, 1997).

Anger directed internally does not strongly feature in the findings of this study. Self-blame and feelings of guilt can be detected in some parents who had passed the CMT gene to their descendants, as well as in a few cases where people felt guilty, in retrospect, about not having informed stakeholders such as the employer, about their CMT. One explanation for the low incidence of guilt feelings may be that people with congenital conditions feel that they had no control in acquiring the disease, or in passing it on to their offspring, since they were unaware that they had the disease when the child was conceived.

Externalised hostility

According to Livneh and Antonak (1997), this type of anger is directed at other people or environmental components that subjectively relates to the onset of the CID or the rehabilitation process. Blaming others, feelings of antagonism, passive-aggressive behaviour by obstructing treatment and general acts of aggression are examples. Another example is anger due to lack of applicable treatment for the CID.

Although, once again, not a dedicated stage as such, externalised anger is commonly encountered in the findings, especially in the earlier stages of adaptation. The anger is primarily directed at: (1) parents who withheld information about their own CMT, (2) the medical fraternity, for supplying inadequate information and for perceived poor performance, (3) significant others, social contacts, schools (in particular teachers), for injustices, unfairness, subjective personal degradation and lack of understanding, (4) the effects of the disease, e.g. painful falls to the ground, especially in public, and for lost abilities and opportunities, and, (5) God, (one participant only) for, in his words, "this gene combination that He gave me". Anger and bitterness of serious proportions were rarely encountered in the stage of optimising, if at all.

Sowell (2000b) see anger as an integral part of mourning. In this context, it refers to the experiencing of unfairness where questions such as "why me" are common. Loggenberg (2006) also mentions a source of anger that is very prevalent in people with CMT, namely frustration. The causes of frustration amongst people with CMT are legion. Struggling with tasks that require finger dexterity, inability to do things alone, falling, cramps, lack of understanding from others, dealing with perceived inefficient healthcare professionals are but a few examples.

It almost goes beyond saying that excessive anger and hostility may be debilitating. It may also generate friction and interpersonal tension, exacerbate stress and alienate crucial support people (Doka, 1993).

The two final stages, acknowledgement and adjustment, will be dealt with together.

Acknowledgement

Acknowledgement refers to a cognitive process, namely reconciliation with and acceptance of the condition, together with all the implications such as functional limitations, as well as integration thereof into the self-concept. A person in this stage displays four characteristics: (1) He or she accepts him or herself as a person with a disability, (2), gains a new sense of self-concept as a person with the

condition, (3)re-appraises life values, and (4) looks for new meanings and goals in life (Livneh & Antonak, 1997).

Adjustment

According to Livneh and Antonak (1997), adjustment is the theoretical final stage of the adaptation process. It refers to emotional acceptance and integrating of the disease, as well as the concomitant limitations caused by it, into the self-concept. There is also enhanced adjustment and reintegration to the outside world. Once again, there are four characteristics of a person in this stage: (1) he or she re-establishes a positive self worth, (2) re-discovers and realises the existence of both remaining and new potentialities, (3) actively pursues and implements social and, where applicable, vocational goals, and (4), successfully overcomes all obstacles encountered whilst pursuing these goals.

With regard to cognitive acceptance and reconciliation, *engaging with CMT* contained an intermediate step that is not encountered in the synthesised model. This is the transition point at the end of the stage of *orientating*, namely *realising the salient implications*. Insofar as the CMT-affected's rather limited disease related knowledge at this early stage allow for, cognitive recognition and acceptance manifested. This acceptance and insight, perhaps not based on comprehensive information as yet, was crucial for the commitment to fight back, in other words for the next stage to emerge.

The entire stage of *fighting back* encompasses efforts to accommodate, cope with and overcome the effects of the disease. In this process, the energy is directed at mastery and survival, in other words overcoming, the effects of the disease to as large an extent as possible. Non-acceptance of their CMT is not the important issue. However, to the extent that acceptance of a disease implies not fighting its manifestations, non-acceptance surfaced.

The finding that those with CMT resisted taking on the identity of a disabled person involves different issues and reasons than acceptance of their condition per se. Although these will be expounded in Part 2 of this chapter (under the sub-

strategy of persevering), a salient one will be introduced here. For many, flinching from the disabled identity amount to resisting the stigma and negative connotations, as well as the threatening implications, which are often associated with being disabled. Intertwined with this is the fact that many equate (rightly or wrongly) the disabled role with the most disastrous outcome of being wheelchair-bound and entirely dependent upon others. Put differently, they did not accept the potential *effects* of CMT, recoiling back from the stigmatised stereotype of the disabled being entirely dependent people. This does *not* mean that they did not accept the fact that they *have* CMT.

A great deal of, what Corbin and Strauss (1988) call "biographical work" (p.10**)** on the self in relation to the disease, is undertaken during this stage, as delineated in the sub-strategy *empowering self*. This process leads to, or aids, acceptance, if the latter had not already been attained earlier. The findings indicate that, overall, acceptance of CMT, both cognitively and emotionally, were not as problematic as for those having to adapt to an acquired condition such as loss of a limb, myocardial infarction and cancer. The possible reasons for this are reflected in the two studies below.

Li and Moore (1998), whose study on the acceptance of disability included participants with congenital disabilities (23% of N), also found that these individuals adjusted and accepted their condition better than those who acquired it later in life. Two reasons are furnished for this: (1) people with acquired disabilities experience severe psychological trauma due to loss. They require greater effort and more time to adapt, and (2) later onset acquired disabilities are more difficult for family and friends to accept. Additional factors aiding acceptance of congenital neuromuscular disease may include (a) better adjustment to the disease, with its slow deteriorating course, is possible because of the long time period that people have been living with it (Nätterlund, 2001), and (b) an appreciation that CMT is not life-threatening, and that there is no cure, in other words there is "no way out".

Whereas cognitive acceptance largely resulted from work during the stage of fighting back, emotional acceptance and adjustment, as described above, corresponds more with: (1) the turning point at the end of the fighting back stage, namely *accepting reality*, and (2) most of the stage of *optimising*. It is however very difficult to isolate cognitive and emotional acceptance in the findings with any measure of certainty. The two processes are closely intertwined, but the overall balance tips towards the one or the other as discussed above.

THE PROS AND CONS OF STAGE MODELS

Stage models are rather controversial, as pointed out by Livneh and Antonak (1997, p.23): "The controversy surrounding the existence of an orderly process composed of phases of adaptation to chronic illness and disability has been raging for many years in the field of disability studies". They continue by stating the positions for and against these models. Those in favour maintain that adaptation to the onset of CID amounts to a gradual process of psychosocial assimilation of changes in the body, self-concept and environmental interactions. This process takes place in a linear fashion via orderly reaction stages, as already described. An implication in these models is that later stages are dependent upon resolution of earlier stages (Kendall & Buys, 1998).

Opponents of stage models criticise it on various grounds. They maintain that people respond to CID in diverse ways and that reaction to these conditions are not universally experienced. A universal process therefore does not exist and psychological recovery does not follow linear, orderly stages. Another disagreement entails the last stage, adjustment, which they claim is not achieved by all people adapting to CID. There is also criticism from a rehabilitation perspective, probably more applicable to disabilities and conditions requiring rehabilitation, such as loss of a limb or function. Nonetheless, the existence of unavoidable stages may encourage passivity in the sense of rehabilitation workers sitting back and waiting for their patients to move through the stages. Stage models also normalise debilitating responses such as denial, anxiety and distress.

Professionals who do not observe the stages, may even regard their patients or clients as abnormal! (Kendall & Buys, 1998; Livneh & Antonak, 1997).

The aim of the present identified stage theory is not to propose a rigid process, neither is the sequencing of the stages absolute. My view corresponds with that of Charmaz (1995, p.661). She writes: "I provide a stage analysis of adapting to impairment as a heuristic device to understand experiencing illness, not as an ultimate truth or as a prescriptive tool for practitioners and patients, as Elizabeth Kübler-Ross's (1969) stage analysis proved to be. Depending on their physical condition and social resources, individuals may tumble through the stages rapidly and repeatedly, or they may plateau for years before moving into the next phase of adapting". In the present theory, however, re-entering of the first stage is unlikely because most people progress in one direction after this basic, orientation phase.

DISCUSSION

The juxtaposition of *engaging with CMT* and the synthesised stage model reflected both agreements and disagreements between the two, with the latter in the majority. In fact, it was only the last two stages of that model that resembled the present identified theory's latter stages. The resemblance was remarkably striking, though. On the other hand, the stages leading up to this point differed just as dramatically, as was described above.

Except for the obvious difference between the number of stages (eight versus three), the most salient difference concerns the beginning and end "points" of the adaptation process. Although both these differences will be elucidated in Part 2, they will nevertheless be briefly illuminated here as well.

Concerning the first, the synthesised model is not entirely explicit as to when the adaptation to CID commences. In general, the beginning of adaptation is taken to be either the onset of a sudden, physical injury/event (e.g. spinal cord injury, myocardial infarction), or the diagnosis of a life-threatening or other serious disorder (e.g. certain neurological conditions). However, people experienced CMT long before diagnosis, although it had not yet been diagnosed.

Even though Livneh and Antonak (1997) and others, such as Wright (1983), do mention that adaptation to congenital conditions differ from acquired ones, in the sense of the former being part of the individual's self-concept since very early childhood, they do not delineate these early processes. Neither are they included in the stages of adaptation as such. Shontz (1975) does include a pre-diagnostic stage, which he calls "The pre-impact phase" (p.160), but does not address the child's subjective experiences in the context of his or her condition.

In contrast, via grounded theory methodology, it was clearly identified that these early experiences are of crucial importance to the adaptation process. Considering that the vast majority of CMT cases have an onset in the first two decades of life (Kedlaya, 2007; Chance, 2001) and that the average age of diagnosis in this study is 29 (median age 28), affected people's disease related experiences stretched over many years and included a great many different contexts, such as the school, early working life and so forth. It follows that these experiences, memories and coping efforts may have a profound influence on the later adaptation to and management of the disease (after diagnosis).

From a theoretical viewpoint, the commencement of *engaging with CMT* is therefore simply as soon as the manifestations of the disease become a stressor to the child or, in the case of late onset types, the adult. More probable than not, it will not coincide with the biological beginning. Commencement, or onset, is also not a starting point as such, but instead a gradual process. A large proportion of the orientating stage of the present theory is devoted to these pre-diagnostic manifestations.

Moving to the opposite side, the last stage of the adaptation process, the synthesised model concludes with adjustment, which is depicted as being the final phase where optimum adaptation has been attained, both intrapsychic and concerning congruence with the environment. Provision is made for individuals not being capable of acceptance of their condition, as well as for the possibility of regression to earlier stages. Emery (1994, p.66), who calls this stage "homeostasis", also concedes that the last stage is an elusive "point".

The identified theory on the management of CMT does not have a final, optimum outcome; there is no finality to the process. In this theory, which encompasses an inherited disease, where the disease has been part of the individual's self-concept from the beginning (Grzesiak & Hicok, 1994; Wright, 1983), acceptance of the condition had rather different nuances. Two identifiable "clusters" of acceptance emerged. First, realisation and acceptance of their lot, in other words, *of the salient implications of having CMT*, emerged relatively early after diagnosis, at the end of the orientating phase.

The second time that acceptance emerges, is at the end of the stage of *fighting back*. Here, it emerged as acceptance *of reality*, meaning recognising that their struggle against the disease over a long period had not altered the course of the disease dramatically. Then follows another stage that has not been encountered in the extant literature, at least not in this format, namely *optimising*. In this stage, the nature of management changes from struggling against the disease to a focus of "making the most of". This process has no finality, but many reach a stage where they subjectively feel that they had "done all that they could", or where active optimising efforts markedly slow, signalling a variant of an outcome. This can however never amount to total finality to the process of handling the disease, because it is life-long and incurable! Yet, as will be shown, by applying the core strategy of *engaging with CMT,* participants to a remarkable extent succeeded in resolving the main concern, namely dealing with *unpredictable CMT manifestations*.

Optimising therefore flows into this variant, a tentative outcome called "*qualified wellness*". This concept amounts to the fact that the core process, enacted through the strategies of *orientating, fighting back and optimising*, only succeeds in eliminating distress regarding certain aspects in certain individuals. Subsequently, some people for whom the core process is unsuccessful in fully resolving their main problem re-enter the stages of *fighting back* or *optimising* so that they can continue to engage with CMT with a view of (subjectively) obtaining a better outcome. Re-entering the *orientating* stage/phase is rare because most had engaged with the disease for many years and their knowledge base about CMT had

progressed beyond the basic level. In cases where people know for certain that they have passed the disease to their descendants, some get deeply involved in the lives of their affected descendants (both children and grandchildren); they in effect re-enter the core process of engaging with CMT in the lives of their affected descendants. Typically, they will then employ the strategies of *fighting back* and *optimising*, or some of them, in the life of the child.

The difference between the two stage models is also encountered in their focus. To begin with, the synthesised model focuses on adaptation to CID. The stages are typically described as being the individual's reaction to the disease, for example the reactions of shock, anxiety and denial. The eventual aim of the individual's reactive behaviour is to successfully adapt, meaning to accept the disease and attain adjustment to it. How this is to be accomplished is not the focus.

In contrast, engaging with CMT has a much broader aim. The focus is more on the management of the disease's manifestations, taking into account past, present and future coping and management activities. Management of CMT includes a strong future orientation, in other words, not merely reactive but also pro-active. The focus is also on strategies directed at making provision for the uncertain disease course over the long-term, and for the future in general. Adaptation as encountered in the synthesised model is part of it, but the identified theory goes beyond merely describing reacting type stages. Engaging with CMT is not only about what CMT affected people do, but also *how* they do it, in other words, the strategies that they employ in order to resolve the main problems emanating from the disease as they subjectively experience it.

PART 2

CONTEXTUALISING THE THREE STAGES

In this section, the three stages of the identified theory will be contextualised within the scholarly literature. Throughout, the elucidations will focus on adaptation to CMT, although the broader field (substantive area) of adaptation to chronic illnesses and disabilities, under which CMT resorts, may also be referred to where applicable. To the best of my knowledge, no grounded theory study has ever been done on the psychosocial aspects of adaptation to CMT. This aspect, together with the paucity of research regarding adaptation to CMT in general (as was pointed out in Chapter 1), means that the discussion may range from rather basic issues to more comprehensive phenomena, including this research's unique findings and contributions. As already implied in Part one, the most outstanding contribution to the knowledge base of engaging with CMT concerns the first stage: orientating and the last stage: optimising.

STAGE 1: ORIENTATING

Temporally speaking, the stage of *orientating* encompasses the earlier experiences with CMT, stretching from the initial encounters with the disease, usually during early childhood, through diagnosis to the "point" where affected individuals grasped the broad, foremost implications of having the disease. The three sub processes, or strategies, of orientating are *dealing with pre-diagnostic manifestations, digesting the diagnosis* and *investigating the basics.*

Dealing with pre-diagnostic manifestations

The literature reflects a tendency for stage models of chronic illness and disability (CID) to focus on adapting to these conditions only after diagnosis thereof. What happened before diagnosis is hardly ever elaborated on, at least not in terms of ill people's disease related experiences. Diagnosis is typically regarded as a significant and often traumatic event, which leads to a set of adaptive reactions by

way of, for example, orderly and sequential stages as the individual adapts to the disease. The first stage in the adaptation process following diagnosis is usually described as shock (Corbin & Strauss, 1988; Livneh & Antonak, 1997; Parker et al., 2003). In addition, researchers on CID are either inclined to focus on conditions with a sudden onset, such as acute cardiac disease, or they assume that the pre-diagnostic phase is of rather short duration, or at least not lasting many decades, which is the typical position encountered in CMT. In fact, the pre-diagnostic stage is hardly ever focused on in the existing research.

In contrast, the present theory identifies the pre-diagnostic stage as embracing very important dynamics in the lives of those affected by the disease. Many, even at an advanced age, vividly remember their childhood hardships because of CMT (Beyer & Daino, 1990). The age of diagnosis of CMT participants in this study ranges from 16 to 47 years, with most participants' diagnoses made around 29 years of age. Since CMT is an inherited disease, the majority had to deal with the disease during almost their entire childhood, although the disease had not yet been diagnosed. In view of the gradual, deteriorating nature of the disease (Carter et al., 1995), symptoms during early childhood are of course not necessarily as serious as in later years.

Leaving the matter of not having received an official diagnosis of CMT out of the equation for the time being, the question arises whether it is easier to adapt to CID if the onset is during childhood instead of adulthood. Overall, Wright (1983) maintains that research has not shown with consistency that it is. Some reason that adults adapt better because of greater maturity, whilst others feel that the child, "being more plastic than the adult" (p.235), can more easily accommodate the changes in self-perception due to their condition. In the case of congenital disabilities, of which CMT is an example, she states that the person will not have to cope with alterations to the self-concept, and that adjustment is therefore facilitated. Better adjustment by those with congenital conditions, as opposed to those with later onset conditions, is also reported by Li and Moore (1998) and Strauss et al., (1984).

Continuing on this note, Wright (1983) maintains that there are many psychological differences concerning adaptation between those who have inherited conditions and those who acquire it later on. Fundamentally, children with congenital disabilities are only aware of their already impaired bodies; they never had a "normal" body. Lussier, (1980) states that these children's processes of body image, ego and self-identity development are more likely than not to proceed in the same way as that of non-disabled children.

To what extent above assertions are unconditionally applicable to children with CMT is not clear. The effects of this disease, like many other neuromuscular diseases, such as muscular dystrophy, are highly visible in the public eye (Livneh & Antonak, 1997). The child is therefore, more than non- affected children, exposed and vulnerable to debilitating social comments and behaviour from others, in particular significant others. Examples are mockery, stigmatisation and marginalisation.

In the *orientating* stage of this research, and in particular the sub-strategy *dealing with pre-diagnostic manifestations*, a great many problems were exposed that CMT-affected children encountered on a broad frontier, especially in the school situation. As mentioned, the impact of this was so severe that many adults vividly remember their trauma as children in this context. Whereas their body image and self-image development may very well have proceeded along similar routes than non-affected children, as mentioned above, the real threat to children with CMT was the potential damage to their self-esteem and self-efficacy.

Being physically weak, accompanied by failure in public, having poor balance, walking rather strangely, having poor handwriting and falling frequently are typical manifestations that more often than not resulted in negative self-related consequences, especially reduced self-esteem and self-efficacy. Further examples are public humiliation, unfair punishment, (punishment for poor performance where physical impairment prohibited the desired performance), unrealistic expectations, ridicule and stigmatisation.

Mynhardt (2002) defines self-efficacy as: "a person's belief in his or her ability to perform a task, reach a goal, or overcome an obstacle" (p.35). The negative effect of abovementioned and other failures, as well as continued experiences of non-mastery, on CMT-affected people's self-efficacy and subsequent performance may be extensive. In the words of Bandura (1982, p.123): "Judgements [own] of self-efficacy also determine how much effort people will expend and how long they will persist in the face of obstacles or aversive experiences" (parenthesis added). In the case of a child, these negative effects may be even greater still, because the child's cognitive and other coping skills are generally not as well developed as an adult's.

Self-esteem, as described by Rosenberg in the mid sixties, refers to an individual's sense of his or her value or worth, encompassing the attitude towards the self. This can either be favourable (the person likes himself or herself) or the opposite (Martín-Albo et al., 2007). The development of self-awareness during early childhood is closely intertwined with the child's perception of his or her physical being. If the individual has negative feelings about the body, this more often than not will result in low self-esteem (Naudé & Bodibe, 1990).

Many authors, amongst them Doka, (1993), Livneh and Antonak, (1997) and Wright, (1983) highlight the crucial role that body image, which is one's internalised mental image of your physique, plays in self concept and self esteem, even to the extent of it being regarded as the root thereof. The subjective component in body image is important, even more so with illness and disability, as explained by Doka (1993): "… body image has a subjective reality. One's body image may plummet even when objective changes seem only minor" (p.10). To what extent CMT-related bodily changes will impact upon body image will therefore depend on the subjective experience of these changes and symptoms. It would also be reasonable to expect bodily factors to increase in prominence as the child grows older and moves into adolescence.

In order to further explore how the myriad CMT- related stressors affect the child at various ages, a fruitful option to pursue is to contextualise it within an applicable

model of child development. Following Northern (2000), the challenges that CMT children face, especially regarding their self-esteem, may aptly be depicted using Erikson's (1963, 1980) stages of psychosocial development.

Erikson (1963, 1980) formulated eight developmental stages that people pass through from infancy to death. For the sake of relevance, only the stages up to young adulthood will be dealt with. The purpose is not to discuss Erikson's theory in detail, but instead to briefly indicate how CMT affects the child at each stage. Each stage is characterised by a psychosocial type of crisis, comprising two opposing qualities that the individual must resolve. The aim is to find a balance between them; if the positive quality dominates, further development follows and the individual progresses to the next stage. Mastery of each crisis enhances self-esteem (Erikson, 1963, 1980).

During the first stage, **infancy** (birth - two years), the two opposites to be resolved are trust versus mistrust. If, especially parents, lovingly fulfil the infant's needs, he or she develops trust. Northern (2000) indicates that CMT is usually not obvious at this stage. A close observer may perhaps recognise early signs, especially if the person is aware of a positive DNA finding. The threat to the infant in this stage is that parents may be experiencing guilt feelings for having passed the gene on to the child, and that this may negatively influence their relationship with the baby.

During the **toddler stage** (1 -3 years), the crisis that the child must resolve is called "autonomy versus shame and doubt". During this stage, motor development begins to gather more momentum; in the words of Erikson (1963, p.251): "Muscular maturation sets the stage for experimentation with two simultaneous senses of social modalities: holding on and letting go". The child learns to walk, as well as many other motor skills, for example to manipulate eating utensils and toys. Talking and bowel control are also usually mastered. In view of the motoric importance, many children now begin to show clear signs of CMT, for instance by being slower to start walking or walking clumsily. Falling more than usual and general diminished fine motoric ability are also expected to surface (Erikson, 1963; Northern, 2000; Naudé & Bodibe, 1990).

It is crucial for children with CMT to be encouraged and praised by parents, both for successes achieved and for trying. This will provide the early constructive beginnings of self-esteem and aid the development of autonomy and control over their bodies. Ridicule and criticism will impair their sense of autonomy, as well as promote doubt and shame (Northern, 2000). Most adults participating in this study had little recollection of their experiences during those very early toddler years. A few people mentioned that their parents told them that they started to walk late.

During **early childhood,** (about 3-6 years), also known as the preschool period, considerable motor skills development takes place, including fine motoric skills, such as manipulating eating utensils, tying of shoe laces and so forth. Children are energetic and are almost constantly on the move while exploring the environment. Psychosocially, children must find a synthesis between initiative and guilt. A clearer image, or concept, of himself or herself begins to emerge. Initially, these amount to "generalised attitudes about themselves", for example a sense that they are "slow or bratty" (Craig & Baucum, 2002, p.296). Self-descriptions in terms of good versus bad, slow versus fast and so forth, have important implications for their self-esteem, which is the evaluative aspect of the self. Parents are still the most influential significant other and providing feedback to their children is crucial in order to motivate them and not induce negative feelings about the self (Craig & Baucum, 2002; Erikson, 1963).

Children may become aware of their CMT during this phase (Northern, 2000). Once again, the motoric aspects may reveal or accentuate the effects of CMT. Since social relationships also expand, increasingly involving other children, for instance nursery school involvement, comparisons with other children also become more prominent.

Even though the onset of the most prevalent types of CMT (particularly type 1) is usually in the first or second decades of life (Young & Suter, 2003), it nevertheless emerged that not many participants had clear recollections pertaining to pre-school CMT manifestations. A few of the younger participants who had attended crèches mentioned that they had difficulty with tasks such as drawing and colouring in.

One participant said that she fell rather often (informed by her mother), and another, that he had broken his ankle before age six. In the latter case, the person was not sure if it was CMT related.

Childhood (age 6-12) encompasses the school going years. The psychosocial crisis that the child must resolve is industry versus inferiority. Physical development takes place at a slower rate, but the motoric skills and abilities are nevertheless still expanding. There is also increased participation in play. The child develops a wide range of skills and competencies at school. He or she learns the reward of task completion and perseverance. Success and praise leads to a sense of industry, resulting in high self-esteem; failure and criticism to feelings of inferiority. At this stage, significant others now include teachers and peers in addition to parents and siblings. Once again, it is crucial for self-esteem development that significant others offer genuine praise and recognition (Erikson, 1963; 1980). The latter refers to both efforts (in other words, for trying), and real successes.

The school situation may exert a profound influence on the development of self-esteem during childhood. The importance of this aspect is aptly summarised by Naudé and Bodibe (1990). They write: "His experiences in the school situation will lead the child to develop certain ideas about his abilities, competence and worth. Western society is generally achievement orientated and the school is an important area where failure and success are experienced. The child now sees himself through the eyes of his teachers and classmates as well, and for the first time he has to compare himself and his abilities with those of his peer group in a competitive situation" (p.29).

The above quote underlines the crucial role of the school in the self-esteem development of children with CMT. The findings of this research confirm this. Irrespective of the fact that children might not yet have been diagnosed with CMT, they nevertheless experienced the physical and other effects of the disease. As are reflected in the findings, the school culture with its emphasis on achievement in sport was a nightmare for affected children. The public humiliation, the

experience of failure (non-mastery) and criticism was detrimental to self-esteem and self worth, which is in line with self-esteem theory. Naturally, negative impact differed between children, depending on the seriousness of their symptoms, the nature of the particular institution, the people involved and the disposition/ personal characteristics of the child.

Northern (2000) mentions another important threat: children with CMT may become afraid to take chances, in other words to engage in healthy risk-taking behaviour, because of repeated physical failure and/or mocking by other children. He continues: "if their sense of discovery, adventure and willingness to try something new is thwarted by feelings of failure, they may enter adolescence with an inferiority complex and feel guilty for not 'trying harder'" (p.46). Beyer and Daino (1990) report still another problem that children with CMT have to contend with, namely that school personnel often treat children with physical disabilities as though they are also intellectually impaired. The latter is also encountered amongst participants in this study, although in a minority of cases.

To conclude, Erikson (1980, p.92), underlines the fact that teachers are in a critical position to potentially influence the self-concepts and identity of children. He therefore writes: "The selection and training of teachers, then, is vital for the avoidance of the dangers which can befall the individual at this stage" [childhood] (p.92, parenthesis added).

Adolescence (age 12 to 20) embraces the life task of identity formation. If unsuccessful, role confusion will be the outcome. Puberty is characterised by sexual maturation and concomitant bodily changes, for instance rapid growth and change in body proportions. Socially, the peer group now becomes increasingly important. Many of the processes and manifestations typical of adolescence may render teenagers with CMT who display the characteristic symptoms, vulnerable to debilitating influences to their self-concepts. In this stage, for example, teenagers are extremely concerned about their body image. In addition, social comparisons, which began gathering momentum in childhood, now escalate sky high. By way of these processes, they compare their abilities, traits and so forth with these

constructs in others (Craig & Baucum, 2002). In addition to this, "research indicates that adolescents are far more concerned than younger children about having their inadequacies revealed to others" (Elkind & Bowen, 1979, quoted by Craig & Baucum, 2002, p.400), and one realises just how vulnerable CMT affected teenagers are in terms of their self-concepts. Wright (1983) underlines the fact that a physical disability or impaired body does have a profound influence on self-related aspects, including identity.

In the quest for identity, adolescents tend to test limits by rebellion and risk-taking. Northern (2000, p.47) applies this to children with CMT: "adolescents tend to be risk takers who feel nothing can stand in their way. This powerful feeling does not correlate with having a potentially limiting neurological condition". He continues by stating that adolescents may engage in unsafe, unrealistic or unacceptable behaviour. Children who may have acknowledged CMT in the past may even seem to deny it at this stage, for example in their choice of physical activities.

The majority of my participants had clear recollections pertaining to life with CMT during adolescence. In the school context problems similar to the previous stage continued, but they additionally had a stronger social component due to the greater importance of peer groups. Fear of negative social scrutiny was so important that a few people admitted to staying away without permission from physical activities such as gymnastics, athletics and even school cadets. They rather faced disciplinary measures! With rare exception, females in particular remembered having difficulties with fashion clothing, for instance shoes, as well as other items that potentially directed attention to physical deficiencies. Matters pertaining to the self will be discussed again in the next stage when empowering of the self receives attention.

Young adulthood (age 20 - mid 20s) contains Erikson's identified life task, intimacy versus isolation, that needs to be resolved. Issues to emerge during this phase are largely about partners and careers. A positive self-esteem will aid both. Northern (2000, p.48) elaborates: "this stage is problematical for all young adults, not just for those with CMT. And choosing a career can be just as tricky as

choosing a mate, especially when weighing and balancing the possible effects of a progressive neuropathy". The biological aspects of CMT, such as drop foot, weakness, poor balance and decreasing stamina may necessitate early career changes.

Most of the above problems listed by Northern (2000) were, according to participants, present to some extent in their young adulthood years. Two people remembered changing occupation more or less during this time, although one of them was a lateral transfer within the same company. Problems with the opposite sex, even though not commonly reported, manifested more in social activities such as dancing.

Digesting the diagnoses

Even though the views of Corbin and Strauss (1988) on diagnosis and other CID related aspects do not focus on long-standing congenital conditions per se, it is nevertheless a very useful work for the purposes of this project. This research duo calls the search for the meaning of symptoms the "diagnostic quest" (p.24). It encompasses three phases, namely pre-diagnostic, diagnoses and post-diagnostic, or filling in phases. The filling in stage is in effect still an integral part of the diagnostic process because diagnostic ambiguities are still being clarified.

Above-mentioned researchers list five possible reasons as to why there is such vast variation in the *duration* of the pre-diagnostic phase. These are the nature of the symptoms, when and how the symptoms are reported, the skill and knowledge of the physician, technological factors and lastly, organisational factors.

With reference to the **first two**, they point out that symptoms are not always alarming, serious or intense. They are in fact very often elusive and are simply not noticed by the affected individual. Doka (1993, p.59) aptly uses Gestalt psychology to illustrate this point: "a basic gestalt premise is that one often ignores the ground and focuses on the figure. In health-seeking this means that one tends to ignore internal states until conditions force one to respond to them either because the internal stimulus, such as pain, becomes so intense that it mandates a

response or because circumstances in the external environment continuously redirect one's attention to the internal state". Often, symptoms are interpreted as normal or, due to lack of information, people follow the strategy to wait and see.

The behaviours described above were typical and indeed often observed in people with CMT. Symptoms were, especially during earlier stages of the disease, not alarming or prominent enough for parents/affected individuals to take action. An additional factor is lack of information, which resulted in wrong attributions, such as "just a clumsy child" and ultimately leading to wait-and-see strategies. As time went on, symptoms escalated and the effects of the disease took its toll. Examples abound in the data, for instance poor performance in sport, difficulty in driving a car, deteriorating handwriting, balance problems, fatigue and falls. At some stage, these became serious enough to prompt diagnosis-seeking behaviours (Doka, 1993).

Another reason for delays and problems with diagnosis concerns the **physician**, and more or less amounts to inadequacies on this frontier. Corbin and Strauss (1988) point out that some physicians simply may be better diagnosticians than others. Additionally, insufficient data, misinterpretation of symptoms and so forth all contribute to diagnostic error, resulting in pursuing of the wrong paths and concomitant lost time.

Perhaps to be expected with CMT, described by Linda Crabtree (2000) as an unknown disease although not a rare one, the results of this research indeed dramatically underline these physicians related problems. Diagnosis was an uphill struggle for most; being sent from pillar to post was almost the norm. Frustration abounded. These experiences substantially influenced subsequent interaction with the medical fraternity. Many people became wary of uninformed medical professionals and either avoided further contact or became very selective in their choice. Physician related inadequacies not only caused considerable frustration, but actually hindered adaptation after diagnosis.

Doka (1993) adds another perspective to this matter by pointing out that diagnosis is as much an art as a science. An element of uncertainty applies to most diseases, and many diagnostic tests merely point at probabilities. Diagnosis amounts to hypothesis testing, becoming more certain at each successive level of the testing process. By way of example of these uncertainty dynamics, a letter from a specialist to a CMT-affected individual may be viewed in **Appendix F.**

The last two of Corbin and Strauss's (1988) reasons, **technological** and **organisational** factors were not found to account significantly for variation in the length of the pre-diagnostic stage in people with CMT. The first entails delays due to technology being unsophisticated, with tests typically rendering false positives and negatives or inconclusive results. Although the possibility of this problem regarding CMT exists, none of the participants recounted this to have been an important factor in their CMT diagnosis. They possessed very little knowledge of the disease and its diagnosis at that stage, though. The fact that genetic testing is only readily available for a very limited number of CMT types, may perhaps hint in the direction of technological problems. Organisational factors, which include matters such as difficulties in getting appointments, lost records and time conflicts, cause frustration, but do not account significantly for the long pre-diagnostic phase in this study.

Possible underlying reasons as to why ill people, or people who suspect that they may have some disease, do not seek diagnosis and medical treatment as soon as possible, will be discussed in the next section, under *handling the CMT status of descendants*. The reason for this is that it relates perhaps better to a phenomenon encountered in that context, namely that parents failed (not in the negative, judgemental sense!) to take their children for official medical diagnosis in order to establish if they have CMT.

The **diagnostic event** itself, described by Nätterlund (2001, p. 35) as "learning of the diagnosis" and by Corbin and Strauss (1988, p.28) as "the announcement" has always been regarded as being one of the most significant events in the entire process of living with a serious disease or disability. Doka (1993, p.63) puts it as

follows: "…diagnosis is often described as a turning point, a time of crisis when one's whole orientation toward life changes". In studies on living with muscular dystrophy, Nätterlund (2001) and Nätterlund, Sjöden and Ahlström (2001) determined that all participants, irrespective of type of muscular dystrophy, experienced learning of the diagnosis as very traumatic.

The way in which the diagnosis is revealed depends on many factors, such as the certainty of the diagnosis, the nature of the disease, the prognosis and the physician's communication skills. The latter, in conjunction with the doctor's sensitivity, play an important role in how the announcement is made; either bluntly and with little compassion, or gently and understanding (Corbin & Strauss, 1988). Wright (1983) advises that medical professionals who are concerned about losing objectivity by being warm and friendly, should consider that objectivity versus non objectivity represents one continuum or dimension and warmth versus coldness/aloofness, another. In other words, they can be warm and interested in the client as a person without sacrificing objectivity. The verbal diagnosis is usually confirmed in writing; often a letter from a neurologist/specialist to the referring doctor (please see **Appendix D** for an example of such a letter).

Having learnt the diagnosis, people respond to it in diverse ways. Two stand out, namely shock/disbelief and relief (Corbin & Strauss, 1988). About one third of participants in this study experienced relief, another third shock and the rest were concerned or had no particularly strong emotions, especially if they were diagnosed very young.

Possible reasons for experiencing relief upon being informed about the CMT diagnosis were elucidated in Part one. To recap, the main reason amounted to the fact that the many years that they had lived with the symptoms, had sensitised the particular individuals for the diagnosis. In other words, notwithstanding being rather puzzled by their symptoms, they suspected that something substantial was wrong with their health. There was also the matter of receiving a legitimate diagnosis in the face of many accusations that their symptoms were not valid and even neurotic.

Responding to a diagnosis with relief is not unique to CMT. Robinson (1988), for example, in a rather large survey (N=826) amongst people with multiple sclerosis, found that 50% of the participants were relieved upon hearing the diagnosis. Essentially, he furnishes the same reasons for the phenomenon as discussed above and states that relief is a perfectly understandable response in the circumstances. He maintains however, that the relief has a different quality by stating: "Even where it (relief) is felt, it may be qualified by shock, anxiety, or be short lived, when a strategic view is taken on how best to manage the future" (p.49). This view of relief corresponds with this study's findings; when relief was experienced, it was not particularly long lasting in the face of future concerns.

More or less one third of this study's participants responded with shock upon learning of their CMT diagnosis. Actually, the traumatic impact of being diagnosed is dramatically illustrated by the fact that so many people with CMT did experience shock (and concomitant negative emotions) despite the fact that they had been living with the symptoms for many decades. However, suspecting that something was physically wrong was not enough to prepare these individuals for being confronted by the magnitude and permanence of a CMT diagnosis. Typical conditions that prevailed here is a scenario where their parents did not know either they had CMT, or chose not to inform their children about the possibility.

Investigating the basics

Doka (1993) mentions that there are several tasks that await a patient and family after the diagnosis has been revealed. The first of these is what he calls "understanding the disease" (p.67), which corresponds partly to investigating the basics as formulated in this study. Understanding is considerably promoted by the furnishing of quality information by the particular medical professional. Although certain information can be acquired elsewhere, the applicable medical professionals are the main source, at least at this stage (Doka, 1993; Wright, 1983). In a study on muscular dystrophy, Nätterlund (2001) found that several of her participants felt that they had not been given sufficient information on the disease. This corresponds with the present findings of this project where all the participants except one felt this way.

Inadequacies in communication, as well as information deficiencies, created unnecessary dissatisfaction, uncertainty, confusion and anxiety that could perhaps have been easily avoided. Doka (1993) highlights two of the social barriers in this situation: (1) the diagnosed individual may be intimidated by the social status and authority of the physician and is therefore hesitant to ask questions, and (2) educational barriers between the two parties exist, resulting in the individual not understanding the physician's language.

Excessive anxiety and other psychological barriers such as an introverted disposition, also work against absorbing and understanding the information. Doka (1993) advises people in this situation to write down questions before their next visit, to inquire about contact particulars of those with similar diseases and so forth. Naturally, with CMT being relatively unknown (Crabtree, 2000), it would be unreasonable to expect general practitioners and specialists in non-related fields to possess the same level of knowledge about this neuropathy than neurologists do.

Even though the gathering of disease related information is important, *investigating the basics* of the identified theory has another equally important component, namely *tracking down one's own inheritance*. The quest for knowledge regarding the origins of one's own disease ultimately served the same purpose as gathering disease-related information, which amounts to enhanced feelings of control in the face of uncertainty, as well as empowerment in order to be able to decide what to do. The strategy of tracking down one's own inheritance, as formulated here, applies only to conditions with inheritance as the only etiology. In this rather limited context, tracking down one's own inheritance is an addition to the existing literature.

Realising the salient implications

The critical junction at the end of the stage of orientating, realising the salient implications, which enabled the affected individuals to decide on the way forward, namely to fight the disease's manifestations, has also not been encountered in previous research. The formulation of this intermediate step is therefore an addition to existing theories on dealing with CID.

The component of *orientating* relating to the period prior to diagnosis, as it emerged in this study, may be regarded as a unique formulation. As delineated in Part one, diagnosis is usually taken as the commencement of the adaptation process to CID. In this study, the influences and dynamics of the early years, by way of the strategy *dealing with pre-diagnostic manifestations*, was found to be an integral part of people's adaptation to CMT. This aspect not only played an important role in the past, but also markedly influences present and future management strategies, and for this reason should be included in a theory on how affected people engage with the disease. Naturally, since pre-diagnostic events occurred in the past, the role of these dynamics will be retrospective.

All participants in this research had been diagnosed a long time ago and nobody was judged to be presently in the stage of *orientating*. *Orientating* nevertheless clearly emerged as a distinctive stage that all had passed through. Some also revisited the stage in connection with, for example, past problems and strategies that they were not entirely happy with. Examples of these are issues regarding their own inheritance, such as unresolved CMT-related issues with their parents or grandparents, as well as ambiguities pertaining to diagnosis. However, these revisits to *orientating* are transient and focus on specific issues. In view of the rather basic nature of most strategies in this stage, comprehensive re-entering of the stage is unlikely.

STAGE 2: FIGHTING BACK

Fighting back emerged as the clearest demarcated stage, especially in the sense that it was easiest to identify which specific action strategies addressed each of the three sub-problems of the main concern, *unpredictable CMT manifestations.* In general, participants dealt with the second and third sub-problems of the main concern, namely the threat of *deterioration* and *the CMT status of descendants,* by way of the strategy *wrestling with worst-case scenarios*, whilst *persevering* and *empowering* comprised the main strategies employed in order to deal with the stressors engendered by the first sub-problem, *symptoms and effects*. Even though the three stages of *engaging with CMT* carry the same weight, as already explained

elsewhere, *fighting back* is the stage in which many, if not most, spent the longest time, exerted the most effort and, if they have progressed beyond it, revisited the most frequently.

Fighting CMT partly corresponds to the process of struggling against an illness, a formulation encountered in the writings of Charmaz (1991, 1995). Her description (1991, p.46) of people busy struggling in this context, encapsulates the essence of the endeavour: "...he vowed that he would struggle (against his disease) until death" and "(his condition) became his personal enemy to confront, to challenge and to conquer" (parenthesis added). *Never surrendering* is the specific sub strategy of fighting *back* that corresponds closest with struggling.

In the context of acceptance of their condition, Charmaz (1991, 1995) actually states that struggling is only one of four ways in which people may respond to their chronic illnesses. Specifically, chronically ill people may: 1) ignore their illness, or 2) they may struggle against it, or 3) reconcile themselves to it, or 4) accept it. Although these ways of dealing with illness focus more on acquired chronic illnesses and not on congenital conditions such as CMT per se, in many instances it may equally apply to the latter. Ignoring CMT did not emerge as a significant factor in the findings of this research. This may be because the disease's manifestations are mostly intrusive and visible, rendering ignorance unlikely. Of the remaining three, struggling and never surrendering will be discussed first, whereas reconciliation and acceptance issues will be dealt with towards the end of fighting back. First, however, *wrestling with worst-case scenarios*, that is to say, strategies that are primarily aimed at resolving the unpredictable components of the main concern, will receive attention.

Wrestling with worst-case scenarios

Dealing with inheritance concerns

The focus in this discussion is on concerns about passing the CMT gene to descendants, not on issues about receiving the gene from parents. Issues about one's own inheritance (from parents) were dealt with elsewhere, mainly in

orientating. Said differently, at this point, we mainly focus on the CMT status of children and grandchildren.

Inheritance matters and one's genetic legacy are major issues, or pervasive conditions, for people with congenital conditions. The following quotations, taken from a qualitative study on the experiences of people with mytonic muscular dystrophy, illustrate this: "Many subjects (40% of the total to be exact) said that the worst aspect of muscular dystrophy is the risk of transmission to their children (p.1020)", and " subjects described guilt at their genetic legacy" (p.1024) (Faulkner & Kingston, 1998, parenthesis and accentuations added).

The expressed sentiments mirror the feelings of a great many participants with CMT; the high level of parent's anxiety about the CMT status of their descendants is one of the most salient findings of this research. CMT-affected parents disclosed at least three interrelated concerns (major stressors) regarding inheritance, which resulted in various action strategies in order to handle them. They are: 1) whether to have children at all, or to have additional ones, 2) if they have children already, anxiety if they had inherited CMT or not, and 3) extreme concern about the well-being of children who, for certain, had inherited the disease. There was also indirect pressure exerted by blood relations of the proband, for instance brothers and sisters, about their own CMT status, as well as that of their children.

Regarding the first stressor, participants of this study either already had children and were anxious about having further ones, or were concerned about whether their married children should have children of their own. Number 2 includes the rather puzzling finding that most affected parents did not take their children to have them officially diagnosed. The third concern mainly involved agonising for the affected child or grandchild. A few people did not fall into these three categories, for example those cases where the children were adopted. They nevertheless had valuable opinions to add on many inheritance matters, such as the merits of pre-natal termination of pregnancy if the fetus has CMT.

Aside from the oversimplification that parents normally want what is best for their children, which salient factors play a role in parents' fear that their children may have a neuromuscular disease, such as CMT? Given that certain conditions are more disabling than others are, why, in general, is having a child with a potentially disabling congenital condition perceived as such a big threat? Why avoid diagnosis and live with uncertainty instead? The answers to these questions are far reaching, embracing variables ranging from societal attitudes towards disabled people, which include formal laws and structures regarding the disabled, to the subjective envisaged hardship that awaits a child with a disability.

It is considered beyond the aim and scope of this project to delineate the vast number of laws, policies, employment practices and so forth that guide dealings with the disabled, as well as the various mechanisms and discourse practices that shape societal meanings and attitudes towards these people. Instead, deliberations at a workshop in this context will be presented to serve as an example of the type of dynamics that eventually, directly or indirectly, exerts influence on prospective parents with a congenital condition grappling with the issue of having children. Opposing views between two keynote speakers at a Disability and Reproductive Choice workshop in Manchester, England, in 2004, strikingly illustrate the conflicting societal views on the matter of people with a congenital disability having children. The discussion is taken from Rodgers (2005, p.15-16).

Dr Bill Albert from the British Council of Disabled People pointed out that the lack of social support to parents of disabled children and the numerous battles in order to gain adequate health, social care, education and so forth for the affected children are extremely depressing and discouraging. Disability is often stigmatised, disabled people are discriminated against and are seen as lesser beings by others. In almost every society, disabled people have remained the poorest of the poor. Disabled people, or many of them, in turn internalise this and think of themselves as unworthy. He asks what sort of message all this sends to prospective parents bringing a child with impairment into this world. There should be a greater emphasis on positively welcoming these individuals into our society.

In contrast, Professor John Harris from The University of Manchester (Bioethics) maintains that, although people with a disability are not unworthy or of a lesser value or status, he has concluded that society has moral reasons to select against disability. His discussion of the concept of harm and analysis of related arguments led him to this conclusion. He asks whether it is better to have a disability, or not to have a disability. Therefore, if one has a choice to create a person with a disability, the matter takes on a different perspective. Those who chose to have a disabled child when they could have prevented it, should accept moral responsibility for their choice.

Another salient factor in the context of having children that influences parents with CMT, encompasses the fact that most genetic diseases generate stress and place a considerable emotional, social and financial burden on the person and his or her family/support systems (Robins Wahlin, 2007). This also applies to chronic illnesses in general (Corbin & Strauss, 1988; Sidell, 1997), as well as to muscular dystrophy in particular (Nätterlund et al., 2001) and CMT (Arnold et al., 2005; Beyer & Daino, 1990). In view of the distress typically engendered by these conditions, it follows that avoidance rather then approach behaviour is understandable. In addition, negative societal attitudes, stigmatisation and discrimination against this group of people may contribute substantially to parents and other blood relatives of the proband's fear and avoidance behaviour.

It is clear from the above that reproductive choice for prospective parents with a confirmed CMT status is an unenviable one. This also applies to the decision to have further children. Indeed, for most parents who carry the CMT gene, and for that matter most other congenital neuromuscular diseases, this decision ranks as one of the most difficult, sensitive and personal issues in the context of living with the disease (Crabtree, 2001; Shapiro & Goldfarb, 1990). The former writer actually advises parents to follow their heart and points out that no CMT affected person (including her and me!!) would have been here had our mothers decided against having children because they could inherit CMT.

Other than to follow one's heart, what can affected parents, who desire to have children, do to manage the risks of having an offspring with CMT, and reduce their anxiety about the matter? Aside from strategies that did not dramatically reduce anxiety, such as meticulous self-monitoring of children, two strategies emerged in this study that added more value in this context. They are, first to acquire knowledge about the disease and second to embark upon genetic counselling, which may or may not include genetic DNA testing. The latter includes minimising the risk by way of prenatal diagnosis (PND) (Faulkner & Kingston, 1998); however, the strategy of PND, in this study, only emerged with regard to having grandchildren and not for having one's own children.

Except for the fact that information gathering here was specifically directed at inheritance and reproductive issues, not much can be added to what has been discussed elsewhere regarding this strategy. Genetic counselling and related aspects need however to be elaborated on. The same applies to the observed strategy of self-monitoring instead of medical diagnosis.

Essentially, *genetic counselling* is concerned with establishing the risk of inheriting a congenital disease, informing the legitimate parties who chose to be informed about this risk, the choices/options available to them, and assisting them in decision-making as to how to proceed. It is a process through which people who are at risk of passing on the mutant gene to children, as well as other at-risk parties, for example blood relatives who are anxious if they have the disease, can get information about the disease. This includes how it is inherited, the probability of transmitting and acquiring it, and if the applicants choose, establishing their own genetic status. The latter entails genetic DNA testing. Although couples and other parties should always make their own reproductive decisions, genetic counselling will contribute to informed decision-making. Both the supply of accurate information by genetic counsellors and good communication skills are crucial for the process to be successful (Emery, 1994; CMT United Kingdom, 2004b; Robins Wahlin, 2007).

In a study by Arnold et al. (2005), it emerged that people with CMT are not sufficiently referred for genetic counselling. Largely, this corresponds with my research findings. Not many participants of this study attended dedicated, separate genetic counselling sessions; in fact, neurologists (and/or other medical professionals involved in the diagnostic process) counselled the majority. Neither dedicated counselling nor counselling by the diagnostic team's medical professionals resulted in completely satisfied customers; the main complaints being that the information base of the medical professionals and counsellors subjectively seemed inadequate, or that their interpersonal communication skills towards their patients were lacking. This corresponds fully with the findings of Arnold et al. (2005).

Research done by Bottorff and colleagues in Canada as recent as 2005 (in Robins Wahlin, 2007) reported that 48% of physicians and 31% of nurses in Canada lacked formal education in genetics (p. 284). No comparative figures for South Africa could be traced. Nätterlund et al. (2001) recommend that genetic counselling be provided on a separate occasion soon after diagnosis, and not during the diagnostic process. They question the assumption that medical professionals always supply genetic information at the time of diagnosis and that it is sufficient: they established, amongst others, that their participants did not always adequately "take in" the information. Subsequently, they state decisively: "it is the task of health-care staff to provide knowledge and foster understanding concerning the hereditary nature of the disease on a suitable occasion before the persons plan to become parents" (p.796). Providing summary letters to patients, information leaflets, and continued follow-up are recommendations by Faulkner and Kingston (1998) to better the counselling process. They also allude to the education of medical professionals about CMT.

The accelerated advances in the field of *molecular genetics* have resulted in tests being available for an increasing number of genetic diseases, for example Huntington's disease, congenital Alzheimer's disease and certain cancers (Robins Wahlin, 2007). The availability of genetic tests for CMT differs. Tests for many types of CMT, especially the rarer types, are not necessarily available to most

people because they are not done at all laboratories, in all regions, and even in all countries (Northern, 2000). Others may perhaps be undertaken more for research purposes. Northern (2000) states in this regard: "There is a difference between what is theoretically possible and what is currently available in NHS laboratories" (p.19).

Further to the discussion in Chapter 2 regarding genetic testing in CMT, Bird (2006) and Kedlaya (2007) provides recent information regarding the CMT types that can be tested for. Genetic testing for CMT 1A, which is widely available, now detects more than 98% of people with this variant. Other types for which genetic tests are currently available are CMT 1B, 1C,1D, 1F, 2A, 2B, 2B1, 2D, 2E, 2F, 2I, 2J, 2K, CMTX, 4A, 4E and 4F. As indicated, these tests may however not be available at all laboratories. In South Africa, testing is only readily available for the PMP-22 mutant gene (mainly type 1A) (Dr. J. Heckman, Groote Schuur Hospital, personal e-mail communication, 2006/04/03).

Is the behaviour of parents with CMT who did not take their children for genetic testing, as well as blood relations of the proband who also did not undertake it, unique? The literature indicates that it may not be. This type of testing, that is to say, genetic testing of family members without signs of the disease in order to establish their chances of developing the disease in future, is called predictive testing. Leaving the matter of allowing minor children to be genetically tested out of the equation for the time being, requests to undergo genetic testing for late-onset congenital conditions by couples and other at risk people have proved to be not as popular as expected (Evers-Kiebooms, Welkenhuysen, Claes, Decruyenaere and Denayer, 2000; Lucassen & Clarke, 2006; Robins Wahlin, 2007). In Huntington's disease, a late-onset congenital neurogenetic condition, for example, only between five and twenty percent of at-risk persons worldwide choose to be tested and subsequently enter predictive testing programs (Evers-Kiebooms et al., 2000).

Although the reasons for this phenomenon differ from case to case, communalities exist. Many of these appear to be equally applicable to a finding of this research, namely that parents were reluctant to take the children even for a normal neurological diagnosis. Tibben et al. (1992), in Robins Wahlin (2007), concluded

that non-participants in predictive testing overemphasise the potential negative consequences of a positive result (that they do carry the mutant gene), for example depression, inability to cope with the results and lacking the courage to face the result. A negative result (non-carrier) also has ramifications; here those involved focus on guilt feelings, depression and fear of being banned from the family. Guilt in this context is referred to as survivor's guilt; the person feels guilty because he or she escaped. Robins Wahlin (2007) highlights a previous finding of her research team, namely that for many faced with the prospect of predictive testing "it is easier to bear uncertainty than certainty"(p.282).

Evers-Kiebooms et al., (2000) maintain that research done in 1996 by one of the researchers in this group, Decruyenaere and colleagues, identified pre-test personality characteristics and coping style as key factors in the decision to be tested. Tested people had significantly higher mean ego strength and better coping strategies than the general population. The better coping strategies amounted to more usage of active problem-solving strategies.

It should be said that predictive genetic testing for late onset genetic diseases, particularly those for which there is no cure, is highly controversial. These medical conditions include certain CMT types with known late onset, for instance a few type 1's, almost all the type 2's, certain type 4's as well as some of the rarer forms (Crabtree, 1997; Kedlaya, 2007). A positive test result means that the risk of developing the disease at a later stage escalates sky-high. The asymptomatic individual carrying the mutant gene may stay healthy for an unpredictable number of years. The implications of knowing one's status are far-reaching and may permeate many facets of life, for example work, marriage and reproductive decisions. To know or not to know one's status amounts to a decision only the affected person can make. It is indeed a decision with tremendous short-, mid- and long-term consequences and should always be a well-informed, free and personal decision without external pressure (Evers-Kiebooms et al., 2000).

One parent in this study who requested CMT predictive genetic testing for his child of minor age encountered resistance because of ethical reasons. The literature

indicates that this is in line with current practice worldwide. Predictive testing of minor children on a parent's request is regulated by internationally agreed guidelines and protocols. These amount to respecting the child's right not to know. Because of the far-reaching consequences of predictive testing, it should not be undertaken until the child has reached the legal majority, and is competent to make his or her own decision (Evers-Kiebooms et al., 2000; Lucassen & Clarke, 2006; CMT United Kingdom, 2004b). Lucassen and Clarke (2006) point out that the vast majority of people with Huntington's disease, when allowed to make informed choices as adults, choose not to know their genetic status. Testing during childhood therefore denies those children informed choices as adults.

When "competent" adolescents themselves request to be tested, a difficult dilemma results, the pros and cons of which is still being actively debated (Evers-Kiebooms et al., 2000). Crabtree (1997, p.298) states a reality regarding adolescents: "Anyone who has reached puberty can either father a child, or get pregnant". She holds the opinion that all young people in a family that has CMT should be tested, whether they show it or not.

The matter of prenatal testing had been dealt with in Chapter 2, under the heading of diagnosis. Regarding this issue, Arnold et al. (2005) reminds us that prenatal testing for any disorder that is not life threatening is still controversial. She and her co-workers discuss three studies relating to reproductive issues in CMT, which had yielded conflicting results. The first, by Macmillan and Harper (1992), found that those people, who expressed the intention to use prenatal testing and terminate an affected fetus, were more seriously affected by CMT than those who chose not to, or who were unsure. In contrast, Lebo (1998) found that mildly affected parents often chose to terminate an affected fetus. Harding's (1995) finding is that views regarding prenatal testing depend on the affected individual's personal experiences of the disease. Many people in her study felt that they would not terminate a fetus at risk of developing CMT 1A because it is a (usually) mild disability.

The latter finding is in agreement with the findings of this study. The vast majority in my study also felt that they would not support terminating a CMT- affected fetus, mainly because the disease is usually not extremely disabling. Harding's (1995) first finding also corresponds with Northern (2000), who states: "Inevitably, views on the subject of testing will be coloured by an individual's own experience of CMT" (p.20).

To conclude this section, a brief discussion of a theory formulated in 1953 by Barker (in Shontz, 1975), as to why people with illness symptoms do not seek medical care, namely diagnosis and treatment. Although formulated more for acute illnesses, elements of his theory may enhance insight into the behaviour of many CMT-affected parents who did not take their children for medical diagnosis, as well as adults in general who also avoided it. The theoretical explanation does not apply to cases where the affected individuals either are symptom free or are not aware of symptoms.

In people with illness symptoms, according to Barker (1953), there is overlap (tension) between forces inducing movement towards diagnosis and treatment, and forces inducing movement away from these actions. He describes five vectors or forces that are in operation. Two support the decision to seek diagnosis and treatment, and three the decision not to. Vectors have either positive (+) or negative (-) signs; the signs are determined by their emotional quality and not whether they induce movement towards or away from diagnoses/treatment.

The decision to seek medical help is influenced by the complex relationship between the positive vectors (where the positive emotional quality moves the individual towards a desired state) and the negative vectors (which moves the person away from suffering).

<u>Forces directed towards diagnosis/treatment.</u>

These are *symptom distress* (S-) and *expectation to return to health if treated* (RH+). The negative sign in the former is because people want to avoid pain and

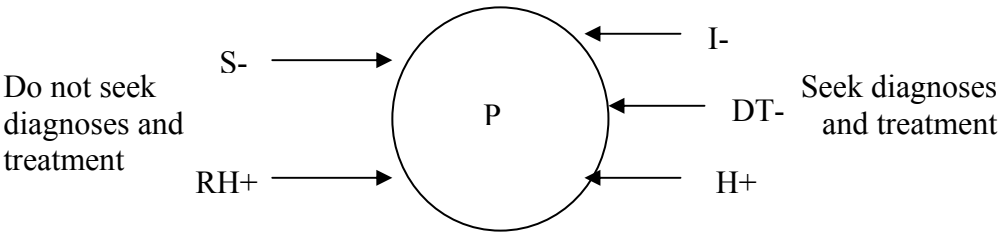
discomfort. The positive sign in the latter derives from the desire to approach a desired goal.

Forces directed away from diagnosis/treatment.

The three are *fear of discovery of serious illness* (I-), *fear of diagnostic and treatment procedures* (DT-) and *concept of self as one who is always healthy* (H+). The first two are negative because people normally avoid these states and the last one is positive because continued health is attractive.

The vectors are depicted in **Figure 5.1.** In this diagram, movement of the person (P) to the right represents his or her decision (and action) to consult a doctor or specialist. This happens because the combined force of S- and RH+ is greater than the combined force of I-, DT- and H+. Movement of the person (P) to the left means the opposite.

Figure 5.1: Forces influencing a person who is experiencing illness symptoms, as identified by Barker (1953) (adapted from Shontz, 1975, p.106)



To re-iterate, if the combined force of S- and RH+ is greater than the combined force of I-, DT- and H+, the individual will take action and seek medical diagnosis and treatment. If S- and RH+ are less than the combined forces of I-, DT- and H+, the individual decides not to seek diagnosis and medical care.

The latter scenario is the typical position in parents with CMT not taking their children for diagnosis, or adults with the disease acting the same way. Firstly,

RH+ is low, there is no expectation to return to normal health due to treatment (there is no cure for CMT), and the course of the disease is characterised by progressive weakening rather than improving (Northern, 2000). S- Also tends to be low because many children, especially at an early age, tend not to initially display serious symptoms, at least not physiological ones (Northern, 2000).

In fact, as was delineated in the previous section on diagnosis, symptoms tend to gradually become more prominent and therefore noticeable. Many parents simply do not notice them or attribute symptoms in their children to "normal" phenomena, for example that the child is clumsy. The very real possibility of parents denying symptoms in their children also has to be recognised. Because children's symptoms may become more serious over time and, additionally, they are faced with more and more challenges, vector S- increases. In instances where symptoms were clearly of a more serious nature, some parents did indeed have the children diagnosed.

As far as forces away from diagnosis are concerned, factor I-, which amounts to fear of a positive diagnosis of CMT, is considered to play the biggest role. DT- was observed in some parents, especially those who had recollections of painful diagnostic procedures, and particularly the electric nerve conductivity test, who wanted to spare their children the ordeal. H+ is regarded as average in the early to mid stages of the disease. Besides, although affected, most people were unaware that they had CMT in the early years because it had not yet been diagnosed.

According to Barker (1953), in Shontz (1975), certain combinations of vectors provide high levels of tension and others low levels. Maximum tension that is possible, is caused by a configuration in which S-, DT- and I- are at high-levels, whilst RH+ and H+ are low. Once again, this is the typical position encountered in this study: as children with CMT grow up, factors S- and I- increase, and RH+ and H+ plummet. It is therefore understandable, at least to some extent, that parents (and undiagnosed adults) reported high levels of subjective anxiety regarding these diagnostic issues.

To conclude this section on inheritance and reproductive matters, a brief look at the situation where children had indeed inherited the disease, in other words with positive CMT status. Under these conditions, enhanced, and in many cases excessive, involvement in the lives of affected children emerged in the findings of this research. In some instances, it approached a scenario of overprotection. Parents also typically responded with self-blame and guilt feelings for having passed the disease to their children.

Emery (1994) states: "The natural inclination for most parents is to shield their affected son or daughter from what they themselves perceive as a cruel blow (having muscular dystrophy) and they respond by being overprotective" (p.62, parenthesis added). He continues with advice that overprotection at any stage of the child's development should be avoided as far as possible and that feelings of independence be fostered instead. Independence will become especially important during adolescence; over-protected, dependent children with disabilities may experience much frustration during this stage, which leads to more stress within the family. Issit (2005) also cautions that: "A fine line exists between expecting too much from a child (with CMT) and being overprotective, both for well-intentioned parents and teachers alike "(p.29, parenthesis added).

Aside from emotional strain and special caring needs, having a child with a neuromuscular disease in the family may generate unique stressors. Professional counselling in order to deal with these stressors is an option for many. Examples of unique stressors are that certain siblings of the affected child being anxious about whether they will develop the disease or not, whilst others may feel isolated and ignored because, invariably, the affected child will warrant more attention. The latter may link to perceptions of overprotection: two mothers in this study were, for example, honest about their CMT-affected children receiving special, "extra" attention. Parents often blame themselves and experience guilt feelings for having passed the mutant gene to the child. Even the sexual relationship between the parents may be affected because they fear conceiving another child with the

disease (Emery, 1994). Indeed, in the words of Issit (2005, p.29) "Being a parent of a child with CMT can be challenging, particularly if you have CMT yourself".

Emery (1994) strongly advises open, honest communication between all parties. According to him, psychiatric interviews with parents of children with Duchenne muscular dystrophy revealed that many (a third to be precise) of them had problems even talking to one another about the disease. Affected children in particular should be encouraged to talk about their fears, frustrations and so forth. Alarmingly, two mothers, participants in this research, exposed that they often wonder how their CMT-affected children are experiencing the effects of the disease and what their feelings in this regard are.

Sowell (2000a) contributes to the subject of parenting a child with a neurological condition by presenting practical advice gathered from parents who have such a child. In agreement with Emery, discussed above, she is of the opinion that supportive behaviour, open communication and honesty are at the heart of dealing with these children. Instead of protecting the child by being euphemistic, the correct terminology should be used; this actually makes the terms less frightening to the children. It is also less disturbing to the child if sensitive and "difficult" questions, such as if they will get better or even be cured, be answered truthfully rather then evasively. Naturally, the language usage and level of communication should be appropriate to the child's age. It is also crucial that parents, from an early age, emphasise the child's abilities and talents.

Managing Deterioration

Fear of undue deterioration, or atrophying, and the likely loss of function/abilities resulting from it, emerged as one of the most formidable problems confronting people with CMT. Its importance as a sub- problem of the main concern, *dealing with unpredictable CMT manifestations*, is underlined by the fact that it probably accounts for most of the unpredictability component of the core. The fear of rampant deterioration, being a pervasive condition, results in the experience of much anxiety and stress, which is aggravated when the individual becomes aware that he or she has deteriorated further.

Ahlström and Sjöden (1996), in their study on muscular dystrophy, found deterioration to be an equally weighty problem in that disease. No less than 73% of their subjects identified it as a major problem, surpassed only by "difficulties experienced because of muscular weakness" (p.368). Arnold et al. (2005) reported similar findings, specifically regarding CMT. By way of example of the severe effect that deterioration is capable of having on a CMT-affected person, a neurologist's letter in this regard may be viewed in **Appendix E.** Fortunately, this particular individual has since remained on a plateau for many years, albeit at considerably reduced levels of physical functioning.

It emerged in this study that deterioration is not necessarily on the forefront of awareness at all times. In fact, many affected individuals reported not noticing atrophy for long periods, and only when it became very prominent. Despite people with many chronic diseases not always being aware of deterioration, looming atrophy is nevertheless an ever-present threat that can surface at any time.

It appears from the literature (Corbin & Strauss, 1988; Horowitz, Rein & Levinthal, 2004) that the phenomenon of not noticing particular manifestations of a disease, or making wrong attributions about them, is not unique to CMT. Even regarding a much more prevalent condition such as Congestive heart failure, Horowitz et al. (2004) found "A deterioration in their condition (short of extreme illness) did not usually trigger a meaningful response. They did not link perceptible cues to action" (p.635). The fact that CMT-affected people have been known to reach plateaus where no deterioration takes place for long periods (even for up to 15 to 20 years - Crabtree, 2001), makes it even more understandable that atrophying may escape attention. However, eventually the effects of atrophying are usually forced into awareness.

What causes an affected individual on a plateau to move from that plateau and start deteriorating again? Crabtree (2001) is of the opinion that sometimes, it is just time for one's body to change, in other words, there is no identifiable precipitating event. She continues that, in some instances, the ageing process, or events such as

menopause or childbirth may "restart" the process or accelerate it if it was very slow. The role of stress in exacerbating CMT or in restarting the atrophying process after a plateau is unclear, and there is a paucity of scholarly literature on this matter. Crabtree (2001, 7th last paragraph, [p. 11]), founder of CMT International, writes in this regard: "There isn't any literature on stress making CMT worse but we have heard from hundreds of people who have experienced exacerbation of their CMT after a major stressful event in their lives". She mentions examples such as death of a loved one, a bad case of flu, a car accident, losing one's job and a divorce.

For us who have CMT, the uncertainty as to how far and fast we will atrophy is very real and indeed a formidable, and for many an extremely anxiety provoking threat. Given vast individual differences, losses either in bodily functions or in skills/abilities due to deterioration become more and more profound as the years pass by. The finding that people with CMT grapple with the experience of **loss**, is not limited to this research. The importance of this aspect is also encountered in other research on CMT (Arnold et al., 2005; Beyer & Daino, 1990) and muscular dystrophy (Nätterlund et al., 2001). As is reflected in the results, and it probably applies to many other neuromuscular diseases and life as well, people with CMT employed the strategy of mourning in order to deal with losses, with a view of ultimately accommodating them into their self-concept.

Erdal and Zautra (1995) researched the unpredictability phenomenon of health downturns (deterioration) in arthritis and vision problems rather extensively. Only arthritis is the focus here, and especially their two experimental groups/conditions. The first group experienced arthritis for the first time (new health downturn group) and the second recurrent downturns (chronic health downturns). They hypothesised that the chronic health downturn group (the second group - stress is predictable) would experience less psychological distress and greater well-being than new health downturns (the first group) due to the predictability of the stress and the possibility of anticipatory coping. The latter entails the opportunity for coping before a stressor occurs; if one knows that it is going to occur, as well as what it entails.

They found the opposite. The chronic arthritis group with recurrent downturns experienced the most distress and the least well-being. Furthermore: "the illness downturns that provoked these reductions in psychological well-being were not catastrophic in magnitude. Most were mild to moderate setbacks in health" (Zautra, 1996, p.703). Erdal and Zautra (1995) explain that additivity theory, formulated by Seta, Seta and Erber, in the early 90s, rather than anticipatory coping, was more applicable to these findings. This theory holds that, over time, the stress of a multitude of negative events will add up to a cumulative negative effect on an individual's well-being. The effects of health downturns therefore compound with each event, resulting in a downward spiral.

With reference to the present study, the majority of participants were able only to provide rather broad, general information pertaining to deterioration; they could not accurately recollect specific information such as the duration (length) of plateaus, as well as the exact extent and duration of each downturn. Three people, two males and one female, reported experiencing rather regular downturns and, correspondingly, relatively shorter periods of plateau over the years. All three, including one person who was retired, were extremely alarmed and anxious about the past and present rate, as well as the extent, of their atrophying. The threat of the pattern continuing or even worsening in future caused formidable stress.

The rest of the participants, including myself, either tended to consistently atrophy very slowly and gradually, or they tended to plateau for long periods. Although this group was also stressed, the stress appeared to be less acute and amounted to a less serious manifestation of "a generalised state of apprehension or foreboding", to use a rather well-known description of anxiety (Nevid et al., 1997, p. 200). I want to point out though, that, for the purposes of the present delineation, I do not want to stray into the differences between stress and anxiety, choosing instead to follow Lazarus and Folkman (1984, p.5) who write: "If one recognizes that there is a heavy overlap between the concepts of anxiety and stress, and does not feel it necessary to quibble about which term is used.....". A related matter is that the

distress under discussion here pertains specifically to the deterioration stressor; it does not mean that the participants were (necessarily) highly stressed in general.

Given that this is a qualitative study where the exact size of variables, such as the strength of downturn-related stress/anxiety, as well as the rate and length of plateaus, were not determined, it nevertheless appears as if additivity and anticipatory coping apply, at least to some extent, to the above two groups of participants. To clarify, there are tentative indications that, in CMT cases where atrophying is more regular and the individual plateaus for shorter periods, additivity theory may apply, irrespective of the seriousness of the deterioration. When the individual plateaus for longer periods, anticipatory coping may be operative.

The effects of repeated losses of valuable abilities by people with progressive diseases are sometimes referred to in the literature as chronic or episodic sorrow. This entails periods of sorrow and stress alternating with periods of peace and happiness (Nätterlund et al., 2001). The latter research group's muscular dystrophy subjects reported losses concerning activities of daily living, leisure activities, independence and gainful employment. In the present study, these losses, as well as the three types identified by Sowell (2000b) and delineated in Part one (the depression stage), were also observed in people with CMT. However, profound manifestations of chronic sorrow did not clearly emerge in the findings of this study. Another finding of theirs that is also encountered in my findings, is that participants perceive the gradual progression of the disease as an advantage because it "gives them time to reflect on what is happening" (p.797). This is also in agreement with anticipatory coping, as described above.

With physiological factors not subject to voluntary control playing such a large causative role in bodily atrophying, it may rightfully be asked to what extent all the efforts and strategies employed by people with CMT (and other neuromuscular diseases where atrophying is distinctive) will make a difference? Can they influence the process of deterioration at all? The answer is twofold:

On the one hand, their efforts will not halt the process of deterioration (Crabtree, 2001). If we however approach the question from another angle of incidence, namely whether their actions can aggravate, or speed up the process, the answer from literature appears to be more confirmative. Put differently, lifestyle and related variables do appear to be able to exert some influence over deterioration, although in a negative way. If for example, weakened muscles are exercised too strenuously, a phenomenon called overwork weakness results, which may cause permanent damage to the muscles (Carter, 1997; Crabtree, 2001; Northern, 2000; Vinci, Esposito, Perelli, Antenor & Thomas, 2003). Crabtree (2001) states that the already malfunctioning peripheral nerves may also be permanently damaged. She is decisive in her recommendation that physical overexertion be avoided by faithfully following a strategy of pacing oneself.

Another example of lifestyle related choices aggravating CMT is medication. Certain drugs are toxic to the peripheral nervous system and may exacerbate or aggravate CMT (Crabtree, 2001; Northern, 2000). Lists of these potentially damaging drugs are available from most CMT organisations worldwide. An example, taken from Northern (2000, p. 35) is depicted below.

Table 5.1: Drugs that people with CMT should avoid
(Source: Northern, 2000, pp. 35-36)

Drugs used in the treatment of cancer
Doxorubicin/Adriamycin
Cisplatinum
Misomidazole (can be used with caution)
Suramin
Taxol
Vincristine

Drugs used in the treatment of tuberculosis
Ethionamide

Isoniazid (INH)

Drugs used in the treatment of heart conditions

Cordarone-X – for irregular heartbeat

Hydralazine (Apresoline) – for high blood pressure

Perhexiline (Pexid) – for angina

Anaesthetics

Nitrous oxide/ Entonox

Succinylcholine (suxamenthonium, Anectine)

Other drugs

Chloramphenicol – an antibiotic

Dapsone – for chronic and certain rare skin diseases

Phenytoin (Epanutin) – for seizures and pain

Disulfiram (Antabuse) – for alcoholics

Flagyl (Metronidazol) – for trichomonas infection

Glutethimide (Doriden) – a sleeping pill

Lithium – for manic depressive illness or headaches (use with caution)

Nitrofurantoin (Furadantin, Macrobid, Macrodantin) – for urinary tract infection

Penicillamine – for rheumatoid arthritis

Vitamins

Mega doses of Vitamins A, B6 (Pyridoxine) and D can be harmful. A mega dose is defined as ten times the recommended daily allowance.

Other substances

Alcohol

Tobacco

Other lifestyle related variables that may aggravate CMT include being overweight, mainly because it puts more strain on weakened muscles and joints, more pressure on the heart and lungs and cuts the amount of oxygen available to

the body. Heavy usage of alcohol can also damage nerves and should be avoided at all cost (Isitt, 2005, p.8). Tobacco is also considered detrimental (Northern, 2000).

Knowingly or unknowingly, many of the strategies employed by participants of this study to resolve the main concern, had at the heart of it the explicit or implicit motive to either pro-actively influence atrophying, or to make provision against it. Generally speaking, it appears from the above discussion that, from our current knowledge base, the best thing that people with CMT can probably do about physical deterioration, is to limit speeding it up by not engaging in known debilitating practices.

With rare exception, it is seen as beneficial for affected people to possess a sense of **control** over as many manifestations and related variables of their disease/disability as possible. According to Livneh et al. (2004), in their overview of various research endeavours between 1977 and 2000 on the topic of control beliefs in CID, findings indicate that high perceptions of control are associated with better psychological adjustment in a wide variety of diseases. These include cancer, cardiac conditions, diabetes mellitus, rheumatoid arthritis, chronic pain and various disabilities. The results of this research suggest that, in general, this may also be applicable to CMT. However, perceptions of control in chronic illnesses and disabilities embraces different facets that still needs to be elaborated on.

Research by Affleck, Tennen, Pfeiffer and Fifield (1987) has shown that three types of control perceptions may be distinguished, namely control over treatment, control over symptoms, and control over the course of the disease. These may also be regarded as **targets** of the control endeavour. In addition, two distinctions are made regarding the **source** of the control: personal and health care providers. The latter amounts to relinquishing control to a health care provider in the face of very limited opportunities for personal control (which was relevant in their study amongst rheumatoid arthritis subjects). This phenomenon is also encountered in other diseases such as cancer. It deviates somewhat from Rotter's mid-1960s theory of external locus of control, in that people with diseases can experience

enhanced control because they believe that both they (internal) and the doctor (external) are able to control the disease (Affleck et al., 1987; Link, Robbins, Mancuso & Charlson, 2004).

Relinquishing control to a health care provider did not emerge as particularly important amongst people with CMT, if at all. A possible explanation for this finding is the relatively low incidence of post-diagnostic doctor consultations for CMT- related manifestations (excluding the period following diagnosis). CMT is characterised by slow and gradual atrophying over many years (Carter et al., 1998). Although eventually "causing significant neuromuscular impairment" (Chance, 2001, p. 280), there is not necessarily frequent and dramatic setbacks necessitating medical consultation, as may be the case in diseases such as cancer. In sum, control perceptions in people with CMT amount largely to perceptions of personal control.

What compromises personal control? The activities of personal control mentioned by Affleck et al.'s (1987) subjects remarkably resemble some of the strategies of this study's stage of *fighting back*, and to a lesser extent, *optimising*. A few examples are: exercising, avoiding/controlling stress, praying, positive thinking, correct diet /nutrition, adequate resting, staying active, regulating of activity level, using medication and seeking/accepting support.

In cancer research, Link et al. (2004) distinguished between pro-active and reactive control strategies in their subjects. The former is intended to optimise well-being and the latter to maintain well-being. Examples of pro-active strategies are exercise, complementary therapies, having a positive attitude, diet and re-prioritising one's life. Reactive strategies include enjoying time now, efforts to decrease fear and anxiety, addressing the effects of treatment, stoic behaviour and planning for death. Clearly, there is a degree of overlap with *engaging with CMT,* regarding both pro-active and reactive strategies. They are distributed mainly throughout fighting back and optimising, with no clear pattern distinguishable.

Getting back to the three **targets** of control perceptions, all three listed by Affleck et al. (1987) are encountered in the findings of the present research project. In fact, two of the control targets, symptoms and disease course, resemble two divisions of the present grounded theory's core concern, namely symptoms/effects and deterioration (see Figure 4.1). Control over treatment overlaps with this two. Participants implemented a myriad of strategies to address these targets, as reported in the findings. In terms of treatment strategies, for example, participants exercised control in decisions such as what medical functionalists to consult, whether to undergo surgery or not (and when), whether to acquire orthoses, which alternative medications to use and what exercise programmes to embark upon, if any.

The subjects of Affleck et al.'s (1987) study felt that they had more control over the symptoms than over the course of the disease, which roughly is in agreement with this research's findings. All participants experienced physical deterioration, or atrophying, as unpredictable and largely uncontrollable, whereas some symptoms were overcome using alternative ways, helping aids and so forth.

Link et al. (2004) found that having perceived control had definite advantages for their subjects, who had cancer. Those who held the belief that they could exercise control over the disease were significantly younger, less depressed, more often employed, living with other(s) and more confident of being cured, than the control group. To what exact extent these benefits are applicable to an inherited neurological condition such as CMT or muscular dystrophy, could not be determined in this study and remains an intriguing question to be investigated using, perhaps, quantitative methodology.

On a more general note, Corbin and Strauss (1988) identify a number of important conditions that help people in downward illness trajectories, which are encountered in CMT, to keep going despite getting worse. Amongst them are:

1. Fluctuations in the downward spiral, which results in ups and downs, or good days and bad days. There are therefore still times of feeling relatively

well: "Moments of joy can temporarily render the illness relatively unimportant" (p.83).

2. The experience of success derived from particular activities.
3. Pursuing personal philosophies, such as "it is God's will", or "taking things as they come" (p.83).
4. Perceived success in influencing the effects of the disease.
5. Searching for cures amongst less traditional modes of medicine.
6. Hoping that a cure is around the corner, and
7. Making and living an intense commitment to keep standing for as long as possible.

In this study, some of the above-mentioned aspects emerged as action strategies rather than conditions, as stated by Corbin and Strauss (1988), for example numbers 3, 5, and 7. In this instance, there are resemblances not only to the stage of fighting back, but also to the stage of optimising (no.3 and, to a lesser extent, no.6).

An apt ending to this discussion on how people with CMT and other neuromuscular diseases deal with deterioration, is considered to be Taylor's (1983) thesis that people facing life-threatening events are unlikely to give up control when some of their avenues of mastery are blocked. Rather, they then actively search for opportunities to gain control in a selective and adaptive manner. If, for example, an individual's cognitions about him or her having control over a tumour (such as beliefs regarding the power of having a positive attitude) is disconfirmed by a second tumour, they are unlikely to give up control and may actively pursue other alternatives, such as dietary changes.

Even though CMT is very rarely a life-threatening disease, the findings of this study indeed reflect the content of Taylor's thesis. Examples in CMT include implementing alternatives (both cognitive and behavioural) when the effects of atrophying surfaces after a long plateau, or when beliefs about being competent in a role or task, for example a job, is shattered by contradictory evidence, such as a poor merit assessment. Practical examples of alternatives employed are getting helping aids, exercising, weight control, turning to religion, altering one's home

and trying treatment options such as physiotherapists and even the alternative route.

Persevering

Never surrendering

Never surrendering embraces the phenomena of fighting and combatting a disease, as well as displaying a fighting spirit. A fighting spirit is also one of the categories in a popular self-report questionnaire that is used to assess coping with Cancer, namely Greer and Watson's (1987) Mental Adjustment to Cancer (MAC) scale, which had been adapted for use in muscular dystrophy (Ahlström & Sjöden, 1994; Nätterlund, 2001).

Never surrendering entails a commitment to fight against the manifestations of a disease and to keep standing for as long as possible. Corbin and Strauss (1988, p.84) summarise it aptly: "An intense and deep commitment to oneself and others can keep one going too. Some people evince tenacity for life - a strong desire to live…" A strategy of non-surrendering does not appear to be uncommon among people living with chronic diseases. An example is epilepsy, where Hofgren, Chaplin, Norlin-Bagge, Carlsson and Malmgren (1998) found that "campaign against" (p.14), which resembles never surrendering, was the most commonly problem-focused strategy employed by their subjects. They continue: "This indicates that even people with a long history of epilepsy do not merely accept the limitations posed by epilepsy" (p.14).

Robinson (1988) obtained a similar finding amongst his subjects living with multiple sclerosis (MS). The majority of spouses of subjects with MS identified "fighting the disease" as the strategy that the affected person is engaging in. It is also encountered amongst people with CMT, as is reported in research by Shapiro and Goldfarb (1990), as well as Arnold et al. (2005) (and the present findings). People living with muscular dystrophy also display this inclination (Ahlström & Sjöden, 1994, 1996; Nätterlund, 2001).

A rather puzzling anomaly between this research's findings on a fighting spirit and that of Nätterlund (2001) is observed. She found that higher levels of a fighting spirit in her muscular dystrophy subjects, as assessed by the adapted Mental adjustment to Cancer (MAC) scale, correlated with poorer quality of life, also assessed by means of a psychometric instrument. The opposite, however, was found in cancer research, in other words, a higher fighting spirit correlated with enhanced quality of life. She reasons that: "patients with cancer can probably fight and obtain palliative treatment for tiredness, nausea and pain, whilst patients with muscular weakness cannot overcome their paralysis", and that "…fighting spirit may be a reflection of an unrealistic notion of what can be controlled in progressive diseases like muscular dystrophy" (p.59).

In this research, CMT participants' fighting their disease, as is most distinctly reflected in the strategy of *never surrendering*, did not emerge as resulting in diminished quality of life. In fact, the majority of people felt that fighting the disease was rather beneficial, in other words, the opposite is suggested by the findings. Arguably, the most salient and perhaps apparent reason for the discrepancy may be due to methodological differences. Quantitative, standardised psychometric testing was not the focus of this qualitative project and the findings largely reflect participant's subjective views. Comparisons with quantitative results are therefore not feasible.

Another possibility is that CMT may not be perceived by those who have it as necessarily being as severely paralysing and uncontrollable as muscular dystrophy. As reported in the "results" section, symptomatic treatment strategies such as helping aids, surgery and certain medications made a difference and in many cases added considerable value to CMT participants' lives. Perhaps people with the more serious forms of muscular dystrophy and other neuromuscular diseases may not experience improvement due to symptomatic interventions so dramatically, if at all. Persisting with efforts to take control of the disease is furthermore suggested by the fact that strategies such as *never surrendering* persisted, although in different and/or reduced ways, after participants have *accepted reality* at the end of

this stage of *fighting back.* It therefore continued in altered forms throughout the stage of *optimising*, and, in some cases, even beyond.

Finding alternative ways

Various studies on living with CMT underline the fact that the disease may, and in many cases does, cause serious physical impairment (Beyer & Daino, 1990; Pfeiffer et al., 2001; Redmond & Ouvrier, 2001; Shapiro & Goldfarb, 1990). A few examples from research are: (1) timed motor performance tests of CMT subjects indicate that affected people take three to fifteen times as long as control subjects to perform prescribed motor tasks (Carter et al., 1995), (2) manual dexterity is impaired in 52% of subjects with CMT, and 68% reported neuropathy related pain or cramps (Pfeiffer et al. 2001), and, most importantly, (3) more than 80% of CMT respondents experience muscle weakness and 75% balance problems (Redmond & Ouvrier, 2001).

Above-mentioned are just a few examples of the myriad of negative physical effects of CMT. It is not surprising then that these physical impairments affect the ability of individuals with CMT to perform their activities of daily living (ADL), such as moving, dressing, taking a bath and manipulating eating utensils, as well as executing their roles and responsibilities (Arnold et al., 2005; Beyer & Daino, 1990). These problems are contained in the leg, "symptoms and effects" of the triangular core problem (see Figure 4.1).

The degree to which symptoms, including physical impairments, interfere with affected people's lives, varies between individuals. The conditions that determine the degree of interference, as reflected in the findings, correspond largely with those identified by Strauss et al. (1984) and include: 1) the degree of seriousness of the symptoms, 2) their intrusiveness/disruptiveness, 3) whether symptoms are frequent, or occasional, 4) their predictability, 5) the degree of visibility and, 6) past experiences with the symptoms. Even though these conditions influence the *degree* of interference, it remains a reality for most people with CMT that their physical limitations *do* interfere with their ability to perform their activities of daily living, as well as to carry out their roles and responsibilities. In this research,

all participants except two felt that their symptoms substantially impair their functioning.

One of the most prominent strategies that the CMT-affected use in order to counter the limitations and impairments is to develop alternative ways and means to perform tasks and achieve goals. In the case of the major symptoms, effective symptom control, according to Strauss et al. (1984), often require redesigning or reshaping many important facets of the person's lifestyle. This process entails changing the way tasks are done, as well as devising alternative ways to do them. In turn, this may require considerable ingenuity and creativity; ingenuity more often than not through necessity and even out of desperation. Strauss et al. (1984) speculate that it may be lack of this inventativeness and social skills that prevent people from fully succeeding in the redesigning task.

Perhaps to be expected, searching for and applying alternative ways and means is also encountered in related neuromuscular diseases. Ahlström and Sjöden (1996) established that no less than 97% of their subjects with muscular dystrophy reported using this strategy. Nätterlund (2001) reports that "devices and tricks" (p.55) was the most often used problem focused coping strategy.

Resisting identity of being disabled

Upon reading *empowering*, the next sub strategy of *engaging with CMT,* (to be discussed in the next section), *resisting identity of being disabled* may at first glance appear to be out of place here, because empowering the self specifically receives attention in that discussion. However, refusing to accept the identity of a disabled person, as it emerged in this research, has more of an element of resistance (including fear) and a keep-standing-for-as-long-as-possible inclination to it than self-empowerment and development per se. As will be shown in *empowering*, all participants have to a lesser or larger extent succeeded in incorporating CMT into their self-concept. After all, the disease has been part of them since birth. However, to be labelled a disabled person went beyond acceptance of the disease for all participants except one. To reiterate, they firmly resisted it. Why is this so?

The reasons for this occurrence can be found in the meanings that CMT-affected people attach to the concept of disability, as well as in societal stigmatisations pertaining to it. One of these meanings has already been introduced in Part one of this chapter, under the heading of acceptance. It entails the phenomenon that many CMT-affected people equate disability with the worst-case scenario of losing the ability to walk and becoming dependent on others. This perception of *threat* (not necessarily conscious) is exacerbated by negative societal stereotyping of disabled people as, for example, being seriously impaired and helpless, and results in the CMT-affected recoiling back from disability.

Another factor is that membership of the disabled minority group is *unattractive* due to societal stigmatisation, stereotyping, negative attitudes and ignorance towards the disabled. The stereotype of a person with a disability is often that of an individual with great misfortune who is more or less permanently enmeshed with their tragic fate and struggling to survive. This stereotyped judgment and expectations typically reflect underlying devaluating attitudes. The disabled community worldwide are often seen as lesser beings by others and are discriminated against (Disabled People South Africa, 2001; Rodgers, 2005; Wright, 1983).

Disabled people are confronted by a phenomenon called "identity spread" (Strauss et al., 1984, p.81). Wright (1983) maintains that Dembo and co-workers introduced the concept of spread back in 1956. Spread occurs when an attribute, such as disability (a negative attribute) leads to negative associations and generalisations about the individual. The same holds for positive attributes, which leads to positive associations and generalisations. She explains that spread: "refers to the powers of a single characteristic to evoke inferences about a person" (p.32) and "not only are specific characteristics of the person inferred from physique, but the person as a whole is sometimes evaluated accordingly"(p.34). Even personality attributes may be ascribed to the person on the grounds of his or her disability.

A practical example is the case where disabled people's mental abilities are devaluated on the grounds of their physical impairments. Wright (1983) describes, by way of example, a situation of a blind person in a library, accompanied by a companion. The librarian addressed the companion as if the blind person was not present, asking "what does he want, does he want to sign it in his own name" and so forth. A participant of this study encountered the same problem. Their host ignored the person in the wheelchair and the companion was asked whether the disabled individual would prefer tea or coffee. Ridiculously, as in the library incident, the disabled person was forced to faithfully answer all questions via the accomplice. The questioner never grasped, or failed to show insight, that the disabled person's mental faculties, voice and so forth, are intact.

In addition to the many negatives associated with the disabled role influencing an individual, Charmaz (1983) highlights another angle of incidence. Many chronically ill people themselves hold ideologies and views about living with a chronic illness, which reflect residuals of the Protestant Ethic. These typically include values such as independence, hard work and individual responsibility. Dependency is viewed as negative; to maintain a normal life is desirable. In fact, the latter is a symbol of a valued self.

In view of the many factors that contribute to individuals with CMT and other medical conditions resisting the identity of being disabled, many employ tactics in an effort to normalise their situation. Normalisation is defined by Strauss et al. (1984, p.82) as: "the use of tactics to make or keep a symptom invisible or, if it is visible, to reduce it to a minor status". Said differently, intrusive symptoms are hidden and, if this is not possible, its impact is minimised by taking attention away from it. The aim is to "pass" as normal, or to do so as far as possible. An example is the occurrence of affected people hiding intrusive symptoms by covering them with clothes or by diverting attention away from them.

Normalising is not at all an uncommon strategy amongst people with chronic illnesses and disabilities. It is for example the third most prevalent strategy that subjects living with chronic epilepsy employed, as is reported in a qualitative study

by Hofgren et al. (1998). Interestingly, the first two positions of their ranking of strategies (in order of frequency of use by subjects) coincide loosely with this study's fighting back, although establishing rankings was never a focus in this research. Their first position is "campaigning against" which corresponds with *never surrendering/a fighting spirit* and second is "making practical adaptations", which resembles *finding alternative ways* (p.13).

In asthma, Van Mens-Verhulst, Radtke and Spence (2004) also established that normalising was prevalent amongst their subjects. And in this study? It was found to be indeed a rather common occurrence, although not the third most prevalent strategy as in the epilepsy study. It was nonetheless observable since childhood/adolescence and appears to be a strategy that is pursued by many people with CMT as they proceed on their way to *qualified wellness*. Even after accepting reality, some still grappled with concealment issues, for example towards the employer. This matter will be taken up again when *optimising* is discussed.

A related concept is Charmaz's (1991) formulation called "conceptual packaging" (p.68). Conceptual packaging is one of two interrelated strategies that people employ to keep their illness contained. The other one is passing, which bears resemblance to normalising discussed above. Conceptual packaging entails not defining illness as being an illness, thereby attempting to separate it from one's life and detach it from one's self concept. For example, physical status is not defined as an illness, but instead as a "condition". Others see themselves as relatively healthy, but with only a few problematic symptoms.

Even though Charmaz (1991, p.68) uses the terminology "illness", it is reasoned that "disability" and "disease" fall into the same category, and that her deliberations therefore apply to *resisting the identity of being disabled*. Many of this study's participants, including myself, conceptually ruled out disability and/or disease by, for instance, defining CMT as a condition, or that we are not abnormal in any way and only do things differently from non-affected people, or that CMT is not such a serious threat and it merely has a nuisance value.

Is concealing the symptoms always fruitful? The answer is no, according to Strauss et al. (1984, p.84), who warns: "Those who conceal illnesses, such as various neurological ones (CMT!), that they believe are discrediting or dishonouring have to carry the burden of anxiety that their concealment may be discovered. At the very best, that discovery may be embarrassing, but it could also be humiliating or destroy friendships or bring about unemployment" (parenthesis my own).

Empowering

The sub-category of empowering was initially called equipping. Although empowering was the preferred term for various reasons, equipping does reflect the essence of a very large component of this sub-category. The aim of empowering is to equip the body and mind with skills and other means in order to engage with the disease, as well as to increase perceived control over one's symptoms and the environment. There are three further sub processes, namely empowering self, empowering the body and empowering the spirit.

Empowering self

General background

Empowering self comprises three strategies, namely broadening of ones knowledge base, self-development and reorganising of the self-concept. The focus here will be on the last one. The other two will only be discussed briefly.

The gathering of **information** about a disease, which is described by Doka (1993, p.23) as "mastery and control behaviours", is a rather common strategy amongst people who have been diagnosed with a CID. Examples are rheumatoid arthritis (Iwasaki & Butcher, 2004) and epilepsy (Hofgren et al., 1998). All participants in the present study attempted to broaden their knowledge base. Although this process took place throughout engaging with CMT, there were two peaks. One occurred in the period following diagnosis, and the other here, in the earlier stages of fighting back.

It was rather surprising to find that approximately half of the participants had communicated with overseas CMT organisations. All felt that the fact sheets published by the Muscular Dystrophy Foundation of South Africa were a great help, and so was their MD magazine. The majority supported the idea of an online support group, where telephonic and e-mail communication will be the communication channels. It is my intention to follow up on this aspect in order to establish the viability of such a structure.

This discussion on the self will focus on how chronic illness and disability (CID), and in particular CMT, influence the self, as well as the affected individual's attempts to maintain integrity and empower his or her self in order to deal with the experienced disruptions. As a starting point, a few basic concepts will be briefly introduced in order to lay the foundation for the elucidations that follow.

The literature reflects a lack of uniformity regarding what parts constitute self, as well as the definitions of the various components. In addition, the self-concept and the different aspects related to it, is a comprehensive field of study, indeed an entire research area on its own. An in-depth discussion of this field is beyond the scope of this project and only those aspects of the self that are considered to be related to the research topic will be focused on. These are general self-concept, self-esteem, self-efficacy and body image.

Mynhardt (2002) defines self-concept tersely as: "… a basic schema consisting of an organised collection of beliefs and attitudes about oneself" (p.28). He describes self-esteem as: "… the self-evaluation made by each individual; one's attitude towards oneself along a positive-negative dimension" (p.31). Lastly, self-efficacy: "Self-efficacy is a person's belief in his or her ability to perform a task, reach a goal, or overcome an obstacle" (p.35). Wright (1983, p.217) explains body image: "That aspect of the self concept which pertains to attitudes, experiences, and functions involving the body is referred to as the *body image*".

The important role of body image and concomitant body experiences, so relevant in CMT and other diseases where body appearance is affected to a lesser or larger

extent, is underlined by Livneh and Antonak (1997) who state: " It (body image) is intimately tied to, and is often perceived to be the root of, one's self concept, self esteem, and, in general, one's personal and interpersonal identity" (p12). Body image has a substantial social component, in other words, it is influenced by social factors. Physical appearance is fundamental to many aspects of human interaction. It, for example, identifies sex, age, race and attractiveness. It is clearly observable and is the source of many judgments about a particular individual. Many of these are negative, as is often the case regarding people with disabilities (Shontz, 1975). In general, it may perhaps be expected that negative judgments may increase in line with increases in the degree of visible bodily impairment.

The self in chronic illness and disability

Livneh and Antonak (1997) allocate prime priority to the role of self in chronic illness and disability. They say: "The hallmark of successful psychological adaptation to a chronic illness or disability is the integration of imposed physical changes into one's reconstructed body image and self perception" (p.12). Based on the findings of this research, I also allocate the same high priority to the self. Arguably, it is in this area that CMT potentially has its most debilitating long-term intrapsychic effect, requiring essential "work" on the self concept, to use the wording of Corbin and Strauss (1988, p.9).

A general remark at this point is that types of work and subjective degrees of importance differ between individuals and problem areas. For many, managing the disease's disruptive physical symptoms and effects in order to survive in the environment, including the activities of daily living, requires an equal amount of work input, if not more, than does work on the self. Naturally, the type of work differs, complicating such comparisons.

A search of the literature reveals a dearth of research on the role and dynamics of the self in congenital neuromuscular diseases and **congenital** conditions in general. In terms of the latter, the work of Lussier (1980), working from the psychoanalytic perspective, is a rare if not the only example that could be traced. Grzesiak & Hicok (1994) and Wright (1983) do mention the issue, but do not really discuss it.

More or less the same situation applies to Strauss et al. (1984). In contrast, various salient works, for example Charmaz (1983, 1991, 1995), Corbin and Strauss (1988), Morse (1997) and Strauss et al. (1984) illuminate the position regarding the self in **acquired** diseases and disabilities intensively and more often than not at length.

A typical angle of incidence in the latter works is that acquired chronic illness and disability (CID) disrupts the integrity of self and that intensive work (Strauss et al., 1984) is required in order to come to terms with and integrate the changes into the self-concept. Corbin and Strauss (1988) write: "When a severe chronic illness comes crashing into someone's life, it cannot help but separate the person of the present from the person of the past and affect or even shatter any images of self held for the future… new conceptions of who and what I am - past, present, and future - must arise out of what remains" (p. 49).

Concerning development of the self in congenital conditions, in other words when a person is born with a particular condition, what is the position in this regard? According to Grzesiak and Hicok (1994), Lussier (1980) and Wright (1983), there are significant intrapsychic differences between individuals who have congenital physical conditions and those who acquire physical losses through trauma or illness. These researchers conclude that a person with a congenital physical disability or disease knows no other body, that is to say, they are only familiar with their already affected bodies. Livneh and Antonak (1997) pithily and accurately summarise Lussier's (1980) stance on children with congenital conditions: "…the process of their body image, ego, and self identity development is likely to follow similar psychological and social routes as that of children without disabilities" (p.4).

To what extent does this important stance by Lussier (1980) apply to the congenital condition of CMT? Before an answer is attempted, it is necessary to briefly look at the route of self-development in normal children, which is what he argues takes place in children with CMT. The delineation links to an earlier discussion (in the stage of *orientating*) of Erikson's developmental stages.

According to Wright (1983, p.218) psychologists agree that very young infants do not distinguish between what is the self and what is not. They, for example, pull their hair without realising that it is *their* hair that they are pulling. Gradually, with maturation and interaction with the environment, conception of self or "I" emerges. Learning about the self occurs via various routes, of which two are prominent at this stage. These are direct sensory experiences of the body and socially induced factors.

The latter entails assimilating of the leads and information provided by significant others, for instance, that they are a boy or a girl, that certain things are not becoming, their appearance, and so forth. The child will also develop feelings and attitudes about these facts pertaining to the self through interaction with significant others. By way of example, certain body parts begin to assume connotations such as good or bad and adequate or inadequate (Wright, 1983). As time passes, knowledge about the self is acquired via two basic processes: a) reflexivity, which entails introspection in the context of one's ability to reflect upon oneself, including as an object or actor, and b) interaction with other people and events (Foster, 1993).

The crucial influence that other people, and in particular significant others such as teachers, peers, and naturally, parents, exercise on the self concept is underlined by Wright (1983) when she writes: "…that the self concept is a social looking glass expresses the belief that ideas and feelings about the self emerges largely as a result of interaction with others (p.228). Foster (1993) is another author amongst many who believe that the self is essentially social in nature, in other words, it is constructed in and through interaction with others.

From this cursory discussion it is clear that children and adults with CMT, in terms of their self-concepts, and in particular self-esteem, are particularly vulnerable in the hands of teachers and significant others. Their typical physical weakness, clumsiness and manifestations such as poor handwriting stand out and are prone to ridicule and stigmatisation. Two closely related phenomena in the context of self-

formation are social comparisons on the one hand and effectiveness and self-attribution on the other (Crocker & Major, 1989, in Foster, 1993).

Social comparisons occur when we compare ourselves with other individuals or reference groups. Comparisons entail two dimensions: better or worse on the one hand, and the same or different on the other. The potential effect of comparisons on people with CMT, and in particular children, almost goes beyond saying. Irrespective of them not having being diagnosed with CMT yet, the fact remains that they typically perform poorly in activities requiring physical ability, such as running and gymnastics.

The tragedy is that children are very often forced into these activities. The fact that the disease is not widely known without doubt contributes to this. Teachers mean no harm, they are uninformed. It is exactly for this reason that schools should be informed about the more prevalent neuromuscular disorders. In this, CMT and other organisations for neuromuscular diseases can play a big role. Perhaps a short, to the point information paragraph on neuromuscular diseases in the manual for guidance and physical training teachers may serve a purpose. If parents know or strongly suspect that CMT is a factor, they should inform the school without hesitation (actually, this applies to all disabilities and conditions that cause impairment in children).

The second factor, *effectiveness and self-attribution*, also impacts greatly upon children and others with CMT. This stance holds that self-views are <u>not</u> exclusively formed by others, it also comes about through effective interaction with the environment. Self-concept is therefore also acquired by our own performance and effectiveness, and not merely by being passive victims of others. Self-attribution, as formulated by Bem in the 1960s, holds that people use their observable behaviour or outputs to deduce inner states and capabilities. For example: Academically, I consistently score the highest marks in my class, therefore I must be intelligent (Foster, 1993, p.801). One can only speculate about the attributions of an adolescent or young adult affected by CMT failing in physical activities such as skating, dancing and so forth.

The situation may not be so simple in the case of children with CMT, who do not necessarily posses the maturity to make meaningful attributions. More importantly, the attribution can hardly be positive if the observable behaviour is one of failed physical performance. In these circumstances, affected children may suffer in silence, and there may be damage to their self-concepts with repeated incidents of failure.

From this rather cursory discussion of the self, it is clear that, in the case of CMT, damage to the self- concept could have occurred irrespective of the fact that formation processes of the self followed the same route as in normal children, as claimed by Lussier (1980). Under certain conditions, children may indeed have grown up without, or with minimal CMT related damage to their self-concepts. For others the conditions were less benign. The conditions include the nature of the school and its culture, level of CMT-related information amongst the parties, quality of parenting and other support (for example, in my own case I was fortunate to have an older brother in the same school who protected me), educational staff's sensitivity, degree of seriousness of symptoms, and personality, as well as age, of the child.

The assertion that children and others with congenital conditions are only familiar with their already impaired bodies, signifying the absence of a causal crisis incident that may put pressure on the self, as it does in acquired conditions (Grzesiak & Hicok, 1994; Wright, 1983), does not necessarily apply to CMT. First, many congenital conditions, such as neurological diseases, of which CMT is an example, have a delayed and sometimes even a very late onset. This means that at some stage there is indeed a diagnostic incident that the individual has to deal with. In CMT type 1, by far the most prevalent type, onset is usually within the first or second decades of life. The onset in type 2 is usually much later, with certain forms even in late adulthood (Kedlaya, 2007; Reilly, 1998).

After diagnosis, as it emerged in the findings of this research, self-concept work came to the fore to a much larger extent. Reorganisation of the self-concept in

order to integrate, or at least reintegrate, the now identified disease, became more prominent. As was discussed in the previous section, even if people suspected that something is wrong, diagnosis was still experienced as traumatic by some.

Second, the above assertion does not fully take into account that affected people may have to deal, in many instances rather continuously, with losses that occur due to atrophying. These losses often involve body image, which is an integral part of the self-concept (Lussier, 1980). A participant of this study specifically mentioned how each loss put pressure on her and how she has to cope with each one anew. Strauss et al. (1984) almost reiterate her communication: "At every downward step in the trajectory- if it is the kind that does not simply remain on a plateau - the ill person must reassess where she is and therefore what social arrangements are necessary to manage effectively her symptoms…"(p.72).

Some may rightfully point out that in CMT, as in many other neurological diseases, the losses are of a gradual nature, which contrasts with the position in sudden onset CID. Affected people may therefore be aware of impending losses and make provision against it. Whereas the argument is valid in some cases, Beatrice Wright (1983) sketches a further scenario: "The change (in one's body) may occur gradually, as during childhood and adulthood, or more suddenly, as during adolescence or as a result of disability. Typically, even a gradual change is consciously first recognised, all at once, as a sudden and startling fact. This occurs when the meaning of the changing physical trait is of such a nature as to affect significant modifications in the self-concept. Thus the wrinkles and the greying hair that had been accumulating over a period of time may not even be noticed until the person suddenly realises,' I am becoming middle aged'" (p.235, parenthesis added).

Empowering the body

On a fundamental and observable level, CMT primarily affects the body. Because of this, it is perhaps to be expected, and was indeed reflected in the findings, that work in this area in order to counterbalance the foremost short and long-term effects of the disease will receive a high priority. This is not merely reactive work,

but indeed also proactive. The latter entails strategies, such as exercising, that is undertaken in order to strengthen the body as a safeguard against atrophying.

Empowering the body comprises three types or groups of strategies: (1) nutrition, exercising and weight control, which are largely aimed at *strengthening* the body and keeping it in good shape (fighting fit), (2) getting helping aids and undergoing surgery, which aim to *correct* certain bodily effects of CMT, and (3) medication, alternative treatment, pain management and (utilising) physiotherapists, generally aimed at the *management* of certain physiological effects. Since it did emerge in the findings, the fields of gene therapy and stem cell research will be introduced as a means of empowering the body.

NUTRITION, EXERCISE AND WEIGHT CONTROL

The *diet* related matters reported in the "results" chapter of this study, namely weight consciousness, as well as eating fresh, healthy foods, such as fresh fruit, vegetables, whole grains, nuts, fish and lean meat, are confirmed by Sennef (1999). Isitt (2005) also holds that a well balanced *diet*, high in fruit and vegetables and low in fatty and sugary food, together with weight control, can help minimize disability. Sennef (1999) additionally addresses the issue of a vegetarian diet. According to him, there exists at least one relatively recent published study, in which it was reported that neuropathy symptoms were entirely relieved in 85% of patients placed on a vegetarian diet and exercise. It should be mentioned however, that a small number of participants was used (N= 21).

To what extent vitamin and a dietary supplementations are beneficial, is still being debated. Passage et al. (2004) demonstrated that ascorbic acid (Vitamin C), a known promoter of myelation, reduced the expression of protein PMP 22 and resulted in significant myelation improvement in a CMT 1A mouse model. Crabtree (2001) claims that there is some preliminary evidence that vitamin CoQ10 (co-enzyme Q10) may be beneficial to some. Grandis and Shy (2005) caution that, although ascorbic acid treatment has potential, the dosage for humans is still unclear because the metabolism of vitamin C is different in mice than in humans.

They conclude: "Dietary supplements, including vitamins, creatine and co-enzyme Q have not been proven effective in treating CMT" (P.28).

Matters such as exercise and diet relating to CMT management should preferably be undertaken under the supervision of professional service providers, for example registered physiotherapists, dieticians, gym instructors and so forth. This also applies to the novel exercise programs Qigong and Tai Chi, which will be discussed below.

The value of *exercise* in the rehabilitation of people with neuromuscular disease (NMD) has been fiercely debated for many decades. One of the main concerns is the possibility of overwork weakness; that putting too much stress on the muscles will be harmful and may actually accelerate the disease process. There is also confusion as to what type of exercise is best. Frustratingly, on a fundamental level, conclusive scientific proof that exercise can indeed rehabilitate people with NMD is still lacking (Van Schalkwyk, 2003).

Abovementioned inconclusiveness does however not mean a total lack of positive findings. Carter (1997) discusses two well-controlled studies in this context. In the first, Aitkens, McCrory, Kilmer and Bernauer (1993) determined that, in slowly progressive neuromuscular diseases, including CMT, a moderate resistance exercise programme over 12 weeks resulted in a 4% to 20% strength gain in subjects without any negative effects. In contrast, the second study by Kilmer, McCrory, Wright, Aitkins and Bernauer (1994), in Carter (1997), determined that a high resistance exercise programme, also over 12 weeks, added no further benefits and in fact caused overwork injury in many subjects.

Nowadays, it is generally agreed that stretching and light, low impact exercise, either gymnasium based or aerobic, as well as activities such as swimming, walking and cycling, may be beneficial to people with NMD, and in particular to those with Charcot-Marie-Tooth disease (Carter, 1997; Isitt, 2005; MDF fact sheet #2, 2002; Van Schalkwyk, 2003). Extreme care should be taken at all times to avoid overworking of muscles.

One way to prevent overwork weakness is to look at new and perhaps even unconventional methods. An example is the rather novel exercise regimes Qigong and Tai Chi, both part of Traditional Chinese Medicine (TCM). Qigong encompasses gentle exercise regimes, aimed at both physical and mental components. Typical elements include posture training, deep breathing, repetitive movements and relaxation. When combined with the more active motions of Chinese martial arts, it becomes Tai Chi (Senneff, 1999; Wenneberg, Gunnarsson & Ahlström, a&b, 2004). According to Swanepoel (2006, p.125) people with medical conditions such as arthritis and injuries may benefit from Tai Chi. Possible benefits in general include better coordination, posture, balance and circulation. He also states that studies on older adults have indicated that Tai Chi may improve balance and reduce the risk of falling.

More than half of the participants in the qualitative study by Wenneberg et al. (2004a) amongst people with muscular dystrophy, reported an increase in mobility due do Qigong. In part II (Wenneberg et al., 2004b), the quantitative study, they found an improvement in participants' perception of their general health, as measured by the SF-36 scale. There was also a significant within-group difference regarding balance, attributable to this exercise regime. Wenneberg et al. (2004 a&b) recommend Qigong for use in muscular dystrophy rehabilitation and mentions that a 2000 study also found improved balance attributable to these methods amongst people with multiple sclerosis. It is regarded as two options to be seriously investigated for use in CMT, that is, Qigong for those more seriously affected by CMT and Tai Chi for the less seriously affected.

Senneff (1999, p.262-263) lists the beneficial physical effects of correct, regular exercising as the:

- improvement in blood circulation, thereby increasing oxygen supply to all tissue, including nerves
- loss of weight
- reduction in low density lipids (LDL) and triglycerides
- increase in high density lipids (HDL)

- lowering of blood pressure
- in many cases, boosting of the immune system

In addition to these physical benefits, he states that exercising improves quality of life. In his own words: "exercise make us feel better about ourselves, that perhaps we figure we are not quite as hobbled by our PN (peripheral neuropathy) as we previously thought, that we look better, have more energy, are generally healthier and happier, etc." (p. 264).

HELPING AIDS

Weakened limbs can be braced by means of helping aids called orthoses. These semi-permanent devices are mostly custom made of a strong, lightweight material, such as plastic, and include insoles, plastic splints, ankle-foot orthoses and wrist braces (please see **Appendix C** for an example of an ankle-foot orthosis). These are invaluable aids to help counteract the weakening effects of CMT.

Crabtree (2001) feels strongly about the value of orthoses and warns that people who should be using bracing but who choose not to do so are actually damaging their bodies through neglect. With bracing, body parts may last considerably longer. People whose self-image is threatened by orthoses may perhaps be rather sceptical of decisive viewpoints of this nature. Perhaps sympathetic discussion of the benefits and comfort of modern orthoses by medical support staff or medical professionals may go some way in persuading CMT-affected individuals to make use of them.

SURGERY

One of the more drastic treatment interventions for CMT is surgery: specialized orthopaedic and other surgical procedures to correct pes cavus, drop foot and other CMT manifestations. Foot surgery is fairly common in people with CMT. In their comprehensive CMT survey, Redmond and Ouvrier (2001) found that younger CMT affected people tended to have more tendon transfers as first surgery, whilst

arthrodesis (surgical fusion of bones on either side of a joint so that no movement is possible) were more prevalent in those with more profound weakness. Isitt (2005) cautions that the decision to embark upon the route of surgery should always be made with great care. Surgery should be seen as a very last resort.

MEDICATIONS

The strategy of using medications embraces both conventional medicine and alternative approaches. With reference to *pharmacological treatment*, there is as yet no cure for CMT on this frontier. Various medications are however used to treat muscle cramps, pain and other symptoms associated with CMT. **There are certain drugs with known neurotoxic effects that should be avoided by people with CMT because of the possibility that it may exacerbate their condition. A list of these medications is depicted in Table 5.1.**

A number of new pharmacological treatments are under investigation and some look promising. A particularly interesting one is the administration of a selective progesterone receptor antagonist (onapristone). Grandis and Shy (2005) discuss an experiment by Sereda et al. (2003) in which it was demonstrated that, in rats, administration of this progesterone blocker reduced the over-expression of protein PMP 22 and improved CMT symptoms without side effects. Although promising, especially for use in CMT 1A, they caution that the findings are still inconclusive and that many issues still need to be addressed.

PAIN

As described elsewhere, pain in CMT may take on serious proportions. Treatments for CMT- related pain include: (1) the use of helping aids or orthoses to give limbs more support, (2) movement retraining by a physiotherapist, (3) medication, (4) programs by specialized pain clinics and (5) alternative treatments, for example acupuncture and massage (Isitt, 2005).

ALTERNATIVE TREATMENTS

Even though some of the well-known alternative treatments such as acupuncture, reflexology and massage did not emerge very prominently in the findings, some participants did indeed experiment with alternative treatment. This encompassed mainly the taking of supplements and natural medications such as herbal extracts, as well as vitamins and tissue salts. A few had tried acupuncture without much success, and rubbing ointments and foot spas were popular.

The field of alternative treatments encompasses a wide range of techniques and remedies, ranging from the more well known ones such as physical therapy and acupuncture to extremely rare forms. There is little agreement what should be included in the field. Senneff's (1999) terse descriptions are regarded as sufficient. Alternative, or complementary, medicine is: "simply treatments out of medicine's mainstream" (p.112) and "unorthodox, non-traditional medicine (p.113)". A dissertation of this comprehensive field is beyond the scope of this project. There are many reliable sources on this topic. A good one that interested readers may consider is the main source that I used: *Numb toes and aching soles: coping with peripheral neuropathy* by *John A. Senneff*. Publication details can be found in the bibliography of this document.

Both Wenneberg et al. (2004 a&b) and Redmond and Ouvrier (2001) direct our attention to the popularity of alternative treatments amongst people with neuromuscular conditions. The former points out that the same popularity is encountered regarding other chronic conditions, such as multiple sclerosis. Senneff (1999) states that: "What some medical professionals may regard as quackery, in fact, appears to work for some people…. the bottom line is whether it gives relief to the individual who's trying it" (p.114). He continues that almost one third of American medical schools, including Harvard, Johns Hopkins and Yale, in 1999 offered coursework in alternative or complementary medicine.

Many participants of this study inquired about gene therapy and stem cell research; what it encompasses and whether there is hope on this frontier.

GENE THERAPY

Conventional gene therapy endeavours to correct a genetic defect by supplying healthy copies of the defective gene. This is extremely difficult to apply in CMT because the therapy will have to be tailored to each of the vast quantity of CMT genes (with new ones still being discovered). To overcome this, a new approach is being investigated, namely to deliver genes that encode neurotropic factors (naturally occurring proteins that stimulate the growth of nerve cells) to damaged nerves. It is possible that this approach will be used to treat all types of CMT (Stimson 2001).

STEM CELLS

The therapeutic use of stem cells is a very active field of research. Breakthroughs and developments are regularly reported in newspapers worldwide. Many people with neuromuscular disease regard it as one of the most promising directions in the search for cures. Fundamentally, stem cells are primitive, undifferentiated cells that can divide and differentiate in order to replace dead or dying body cells. There are many types, but for our purposes, only two need to be mentioned. The first is adult stem cells, which are present in the body cells of people at all ages. An example is those found in bone marrow, where they actively replenish red blood cells. Adult stem cells are mostly limited regarding the number of different cell types into which they can differentiate. Certain types, however are more versatile and do have the ability to differentiate into a variety of tissues (Giacomini, Baylis & Robert, 2007; Scuklenck & Lott, 2002).

The other important type of stem cells, namely embryonic stem cells, is the reason for much controversy and debate worldwide. Embryonic stem cells are found in the inner mass of the embryo at the very early stage, when it is only 100 to 200 cells large. To harvest them means destroying the embryo. These cells have the amazing ability to differentiate into any type of tissue and offer promising ways to cure a wide variety of diseases, for example Alzheimer's disease, cystic fibrosis and organ failure. In particular, they offer the exciting possibility to cure diseases where loss of specific cell types is involved (for example CMT). Typically, new,

healthy cells would be produced in the laboratory and then injected into the affected tissue to slow and eventually halt the progression of the disease. A positive development for CMT affected people is that scientists have recently found ways to convert stem cells into nerve cells and myelin producing cells. In future, CMT-damaged nerves may be repaired in this way (MDA USA, 2005; Scuklenck & Lott, 2002).

Giacomini et al. (2007) and Scuklenck and Lott (2002) directs our attention to the reality that stem cell research still faces formidable challenges because many fundamental issues are still unresolved. Needless to say, there are also the many moral and ethical issues surrounding the use of embryonic stem cells that needs to be sorted out, in particular the issue concerning destruction of the embryo. Even though embryonic stem cells can also be harvested from umbilical cord blood, it remains a very sensitive issue.

Empowering the spirit

A marked increase in religiosity, either gradually or more sudden, was a characteristic manifestation for most participants of this study. People with CMT typically engaged with the Almighty with a view of utilising His divine strength, assistance and guidance in their struggle against the symptoms and stress engendered by their disease. In the stage of *fighting back*, religious coping strategies amounted largely to the utilisation of God as a resource.

In his seminal book, *Psychology of religion and coping*, Kenneth Pargament (1997) postulated that people's religious beliefs are often translated into practical, specific strategies as a means to cope with stressors. This in turn more often than not influences mental and physical outcomes. In fact, there are indications that religion may be the most frequently used resource to cope with stressful events, such as illness and disability (Burker, Evon, Sedway & Egan, 2004).

The expansion in religiosity witnessed in the present study is therefore very far from unique. A few examples of conditions studied where religious coping contributed to positive health outcomes, are chronic lower respiratory disease and

cystic fibrosis (Burker et al., 2004), arthritis (Abraído-Lanza, Vásquez & Echeverría, 2004; Iwasaki & Butcher, 2004), Cancer (Laubmeier, Zakowski & Bair, 2004) and various studies on stroke/ blood pressure amongst the elderly (Senneff, 1999).

Concepts such as religiosity, spirituality, religious coping and spiritual coping have not been defined uniformly in the literature. The present discussion focuses more on religious coping and the description of this concept according to Abraído-Lanza et al. (2004) will suffice. They see religious coping tersely as strategies of an intrapsychic (mainly cognitive) and behavioural nature that are based on religious beliefs, values and practices. Paloutzian and Ellison (1982), in Laubmeier et al. (2004) regard the term spirituality as a broader concept that comprises both religious and existential dimensions. As already mentioned, in the stage of *fighting back* empowering the spiritual aspect had more to do with the religious component, whereas the existential component surfaced more in the stage of *optimising,* and will be discussed there.

Carver et al. (1989), who calls religious coping "turning to religion" (p.270), states that people under stress turn to religion for a variety of reasons. First, religion may serve the function of being a source of emotional support. Second, it may be a mechanism used to reinterpret stressors and grow by, for example, attributing ill health to God's will (in the positive sense). Lastly, it may be a tactic or strategy of active coping with the particular stressor. Interestingly enough, they, as well as Carver (1997), found that religion is neither problem focused nor emotion focused coping, but instead a separate factor on its own. Senneff (1999), perhaps rather idiosyncratically, regards prayer as a form of alternative treatment.

In their research on religious coping in end-stage pulmonary disease, Burker et al. (2004) adapted the RCOPE scale (Pargament, Koenig & Perez, 2000), and used only the appropriate 9 subscales (out of the full RCOPE's 21) which, according to them, have been identified by researchers such as Harrison, Koenig, Eme-Akwari and Pargament (2001) to be the most promising as reliable outcome predictors. The subscales of the RCOPE may be regarded as coping methods, which in turn

resort under various coping functions (Pargament et al., 2000). The nine coping methods (or strategies) utilised by Burker et al. (2004) were also applied in this study. They emerged clearly in the findings, that is to say, they were employed by many participants and will therefore be briefly discussed. The nine coping methods form four clusters. If a religious coping method is generally related to positive outcomes, a plus sign (+) is allocated. If it is generally associated with negative outcomes, a minus sign (-) is allocated and ± is used for mixed outcomes. The delineation is based on Burker et al. (2004, p. 182).

A. Religious coping methods to find meaning

1. (+) Benevolent *religious reappraisal*.

Redefining the stressor in religious terms as benevolent and even beneficial. For example, "God is trying to strengthen me".

2. (-) *Punishing God reappraisal*.

Redefining the stressor as God's punishment for one's sins.

B. Religious coping methods to gain control

3. (+) *Collaborative religious coping.*

Efforts to get control through problem solving in partnership with God.

4. (±) *Passive religious deferral*.

Passively waiting on God to act and control the situation.

5. (±) *Self-Directed religious coping.*

Attempts to gain control on own initiative without God.

C. Religious coping methods to gain comfort and closeness to God

6. (+) *Seeking spiritual support*. For example, "I looked to God for His support, guidance and strength".

7. (-) *Spiritual discontent.*

Confusion and dissatisfaction with God in the context of the stressor.

D. Religious coping to gain intimacy with others and closeness to God

8. (+) *Seeking support from the clergy or church members*.

For example: "I look for love and support from members of my church".

9. (-) *Interpersonal religious discontent*.

Confusion and dissatisfaction with the clergy or church members, disagreeing with what they want the affected individual to do or believe.

All the above methods of religious coping, with the exception of number nine, were to a larger or lesser extent employed by people with CMT. The positive and negative outcomes, as indicated by the +, - and ± signs, also crystallised accordingly, for example the individual who was dissatisfied with God and blamed him (*Spiritual discontent*), had a negative outcome. This person also displayed indications of number two and was so depressed that he had seriously attempted suicide at least once.

Those who saw God's role in their CMT as having a positive purpose (*Benevolent religious reappraisal)* and sought *spiritual support* were more at ease about their "lot" and displayed less anger and bitterness. *Collaborative religious coping*, in other words, coping with CMT in partnership with God, was the most prevalent strategy, although *passive religious deferral* was also encountered. Both had positive outcomes in terms of gaining (perceived) control over CMT-related stressors. Very few, if any, employed *self-directed religious coping*. Those actively involved in the church *(Seeking support)* tended to have a more positive outcome, not only in terms of the support and perceived contribution, but also because of the fact that they were practically *doing* something. The latter relates to the concept of problem-focused coping, as opposed to emotion focused coping (Lazarus & Folkman, 1984).

Burker et al. (2004) also makes a distinction between, what they call "global indicators of religiosity", and "the functionality of religion" (p.180). The former entails variables such as church attendance, religious involvement and strength of religious beliefs and practices. The functionality of religion has to do with how people use religion to make sense of and cope with the problems. In their study amongst 90 individuals with end stage pulmonary disease, they found that the two global indicators that they assessed (church attendance and religious involvement), were unrelated to outcome variables, which amounted to distress and disability.

This finding partially resembles the findings obtained in the present project. As far as church attendance is concerned, a few people reported that they avoided going

to church because of uneasiness and fear of falling on steps and uneven surfaces. Church attendance would therefore not be a good indicator of religiosity. However, many were involved in religious and church activities, which possibly also involved moving across uneven surfaces. Perhaps the activities took place later during the day when morning stiffness was not a problem. Unfortunately, I did not ask.

Another finding of Burker et al. (2004) is also contradictory to this study. It concerns the functionality of religious coping regarding cystic fibrosis (CF), which, like CMT, is a congenital condition. They found that CF patients engaged in a self-directed coping style (seeking control on own initiative without God), whereas the non-CF group to a much larger extent utilised religious coping, and specifically the strategy of passive religious deferral. Burker et al. (2004) attributes this finding to the fact that, because cystic fibrosis patients are born with the disease and are treated for it since a very early age, they become accustomed to their self-care regimens and limitations. Over many years, they have become aware of the influence of their own behaviour with regard to their condition.

These dynamics are not necessarily the case in people with CMT. Typically, they did not even know that they have a medical condition, as was explained elsewhere. Treatment, even if available, is therefore unlikely. Perhaps this explains the revealed discrepancy in coping between the two congenital conditions. People with CMT to a very large extent employed religious coping strategies, in fact, a great variety of them.

To conclude, Abraído-Lanza et al. (2004) criticises the coping literature for often suggesting that religious coping is a passive form of coping. In their study amongst Latinos with arthritis (N=200), they found that religious coping was positively correlated with active, but not with passive, coping. This finding largely corresponds with religious coping as it emerged in this study. In the stage of *fighting back,* religious coping was more active than passive. Passive elements were present, but active methods, such as the managing of CMT in partnership

with God, seeking His divine guidance in matters pertaining CMT and active involvement in church activities, were more prevalent.

Accepting reality

In this study, accepting reality emerged as comprising at least two intertwined facets, namely full acceptance of the condition in terms of having fully integrated it into the self-concept, and a realisation of the futility of undue struggling against the relentless *course* of the disease. The two processes occurred concurrently. The first was discussed at length under "empowering self" of the previous chapter (Results) and elsewhere; the essence being efforts aimed at the post-diagnostic integration (or, more likely, reintegration) of CMT into the self-concept. The second received attention under "accepting reality"of the same chapter. The latter does not refer to the myriad practical day-to-day effects and symptoms of CMT, where struggling/fighting in many cases was fruitful.

It emerged that, for many, work on the self concept continued to the end of the stage of *fighting back*; as is indicated by the discovery that certain individuals with this congenital disease were still grappling, even at this late stage, with issues such as concealing the disease, declining support, anger and so forth. However, eventually managing to fully (cognitively and emotionally) integrate the disease into the self-concept, aided the acceptance of reality in terms of insight that their energy sapping, high intensity efforts to alter the course of CMT were for the most part futile. The opposite scenario is equally possible; insight into the futility of their efforts could have aided full acceptance. Add to these dynamics the fact that there was an earlier, intermediate step of "preliminary" acceptance at the end of the stage of *orientating*, and it becomes clear that acceptance behaviour in the present project may not fully correspond to the extant literature on the topic.

The literature reflects rather diverse approaches to the question of what exactly the acceptance of a chronic medical condition or disability encompasses. The different foci and accompanying terminology include: "coming to terms" (Corbin & Strauss, 1988, p.76), "accepting intrusive illness" (Charmaz, 1991, p.46), "accepting disability" (Li & Moore, 1998, p.13), and "acceptance of loss" (Wright, 1983,

p.163). In addition, the accent appears to be on acquired, adventitious disabilities, such as amputations, spinal cord injuries and becoming quadriplegic, although certain diseases and illnesses are featured as well. Although CMT can be disabling, it is not necessarily the case (Crabtree, 2001); much of the disability literature may therefore not be directly applicable.

According to one fairly often encountered stance, acceptance of a CID means that the affected individual recognises and agrees to his or her status without rebelling, anger and other debilitating emotions. Pain and grief did not disappear altogether, but affected people are no longer overwhelmed by it or, for that matter, by their disease in the broader sense. They have succeeded in integrating it into their lives (Charmaz, 1991; Loggenberg, 2006).

A few salient approaches to the acceptance of CID, as encountered in the literature, will now be briefly introduced and compared to *accepting reality*.

The first approach had already been elucidated at length in Part one of this chapter. It encompasses the last two interlinked stages of the synthesised stages model (Livneh & Antonak, 1997). To briefly recap, the last two stages are *acknowledgement* (mainly the cognitive acceptance of the medical condition and integration thereof into the self-concept), and *adjustment* (mainly the emotional acceptance component, as well as reintegration into the outside environment).

In that discussion, it was indicated how *engaging with CMT* differs from the extant stages-models as summarised by Livneh and Antonak (1997). Suffice it to say here that it is difficult to pinpoint exactly where in the adaptation process acceptance "occurs". In addition, even acceptance is not cast in concrete; the degree of acceptance may fluctuate due to a variety of positive and negative conditions, such as atrophying, setbacks, mastery, symptomatic treatment successes and so forth. Charmaz (1991, p.47) summarises it accurately: "acceptance shifts and changes as experiences, plans and prospects change".

Despite the above difficulties, at least two rather condensed periods of acceptance emerged. The first is at the end of the *orientating*-stage, and is called "*realising the salient implications*". This acceptance is based on preliminary, and more often than not rather limited, disease-related knowledge and insight. It amounts to a general realisation of "what is in store" on the road ahead, that is to say, a tentative realisation of the salient implications of having CMT. This "preliminary acceptance" is crucial for the next step to emerge, which is the “decision” to fight back. Implicitly or explicitly, CMT-affected people realised that they have a fighting chance, in other words, that CMT does not necessarily result in disability and dependency.

A second cluster of acceptance behaviour manifested as "*accepting reality*" towards the end of the stage of *fighting back*. In this instance, CMT-affected people were confronted by, and ultimately accepted, a fundamental reality. This amounts to a gradual realisation that their *fight-back* strategies did not have a substantial effect on atrophying and related concerns, although they certainly have added value as far as countering the practical symptoms and effects of the disease are concerned. Many were also still grappling with concerns regarding the possibility of their descendants inheriting CMT. People affected by CMT have to accept this reality in order to shift the focus of their efforts from a struggling-against-orientation to a making-the-most-of-what-is-left-orientation. In so doing, they enter the stage of optimising. As mentioned in the two opening paragraphs of the present discussion, integration of the disease into the concept of self occurred concurrently.

The fact that degree of acceptance may fluctuate to some extent (rather than being a rigid end point of the adaptation endeavour) does not mean that it is characteristic for CMT-affected people to readily move from total acceptance to non- acceptance of their condition, if at all. Regressing back to non- acceptance is not characteristic of people who are in the stage of optimising.

As already noted earlier in this chapter, Charmaz (1991), in the context of acceptance, holds that people may respond to their intrusive illnesses in four

possible ways. They may ignore it, struggle against it, reconcile themselves to it or they may accept it. *Ignoring* an illness applies more to invisible conditions, or to vague or minimal symptoms. This largely excludes CMT, which is rather visible in terms of distinctive ambulation, clumsiness and so forth. Charmaz (1991) describes *struggling against* an illness as: "(the condition) becomes his personal enemy to confront, to challenge and to conquer" (p.46, parenthesis added). Some people struggling against their condition may focus their energy on hope for recovery, whilst others accept their disease and find a variety of ways to work around it. The latter corresponds largely with the position regarding struggling against and combatting CMT as encountered in this study.

At first glance, it may appear as if participant's persistent struggling against CMT amounts to low or even non-acceptance of their condition; yet it did not emerge as such. According to the findings, most participants felt that they have in due course accepted their condition and have made it part of themselves. The reasons for combatting CMT did not emerge as very strongly related to acceptance issues. The participants' discourses reveal that many were fighting the disease's manifestations for reasons that had little to do with acceptance or non-acceptance; mastering the environment, keeping standing for as long as possible, not succumbing to the disease's manifestations and experiencing some control in the face of uncertainty, were just a few apparent goals. Nevertheless, the possibility cannot be ruled out that, for a minority of people, persistent struggling against CMT contained an element of non-acceptance.

When people *reconcile* themselves to their diseases or disabilities, they acknowledge and accommodate them, and tolerate them within limits. Beyond these boundaries is seen as beyond human capacity to tolerate. An apt example in the case of CMT is most people finding sitting in a wheelchair, or losing independency, as outside their capacity to bear. This amounts to the *extent* of the disease being the problematic element, and acceptance is thus conditional. More or less all participants had in due course reconciled themselves to CMT.

In terms of *acceptance*, Charmaz's (1991) fourth way of responding to CID, the position is similar to *struggling*, discussed above. It amounts to the rather unique situation that, even though all participants had largely integrated CMT into their self-concepts, combatting CMT continued. Even though *fighting back* either receded into the background, or took on a different form, during the last stage of *optimising*, it never stopped. In fact, it amounted to more or less a lifelong commitment. A few reasons for this were discussed above, under *struggling*.

Continuing with the latter discussion, it appears as if acceptance of CMT has two separate facets that can hardly be separated meaningfully, namely acceptance of the disease as such and acceptance of its effects. To clarify, almost everybody in this study fully accepted that they have CMT and eventually succeeded in integrating it as part of themselves, yet they did not unconditionally accept all of the disease's potential short and long-term effects and implications, hence the combatting behaviour. Naturally, this reasoning is only applicable to the extent that fighting a disease's manifestations implies not fully accepting it.

It needs to be pointed out however, that these dynamics may simply be a case of affected people having reconciled (Charmaz, 1991) themselves with having the disease and that they have not fully accepted it. However, even those with very light symptoms and slow atrophying still combatted CMT. As already discussed, it emerged that the reasons for fighting back for the most part did not relate to acceptance issues. In addition, becoming aware of profound atrophying or loss of function, caused alarm and often triggered renewed fight-back behaviour. This also applies to observing even minor manifestations of the disease in descendants, especially those that have not yet been diagnosed.

Corbin and Strauss (1988) regard coming to terms with a CID as one of four overlapping and simultaneously occurring biographical processes. The four processes comprise "biographical work" (p.68) that needs to be done in order to put the individual's disrupted (by CID) biography back together again. In short, the processes are: 1) *contextualising* (incorporating the condition's trajectory into one's biography), 2) *coming to terms* (acquiring insight into and acceptance of the

biographical consequences of diminished and failed performances), 3) *reconstituting identity* (reintegration of identity around the limitations in performance into a new, unique wholeness of self, and 4) *recasting biography* (arriving at a new biographical scheme that will provide direction to the biography on the way forward).

In the formulated grounded theory of this project, most of the above biographical processes emerged, with different nuances and levels of intensity, under processes and sub-processes with different names, such as: *realising the salient implications, empowering self, accepting reality and optimising*. However, a question that may rightfully be asked is *to what extent the biographical disruption in CMT is as severe as in acquired, adventitious conditions.*

It has been argued elsewhere in this thesis that many people with CMT, especially the late onset types, experience a similar crisis (that requires adaptation to) upon diagnosis as their counterparts with acquired conditions. On the other hand, although not diagnosed, they had been living with the disease's manifestations, more often than not, for many years and with rare exception knew that something was drastically wrong. Furthermore, they had most probably adapted to the manifestations to some extent. This possibility of less severe biographical disruption may partly explain a finding encountered in the literature, namely that people with congenital conditions in general display better adjustment to their conditions than those with adventitious disabilities (Li & Moore, 1998; Wright, 1983).

However, it emerged in this study that, for many CMT-affected people, damage suffered to their self-image, which includes self-esteem, body image and self-efficacy, particularly during childhood, was experienced as traumatic and did indeed cause varying degrees of disruption to their biographies. It will have to be conceded however, that the debilitating effect of damage to the self could have receded over the years. Unfortunately then, the above-mentioned question cannot be answered in the present study and remains a task for future research that

specifically involves participants with various types of conditions, such as chronic, acquired and congenital diseases, as well as sudden onset types.

A last approach to the acceptance of a chronic medical condition or disability (Wright, 1983), views this process as a series of value changes or shifts that must take place for optimal acceptance of the condition to take place. This approach largely reflects values and behaviour typically encountered in the stage of *optimising*, which will be discussed in the next section. Besides, acceptance in this study emerged as a continuous process rather than an end-result, in other words, it continued in the stage of optimising.

STAGE 3: OPTIMISING

Introduction

The stage of optimising, like the first stage of orientating, is a new formulation that, to the best of my knowledge, is not encountered in the extant research literature on adaptation to chronic illnesses and disabilities (CID), at least not in the format as it emerged here. Even though people with CMT optimised resources and other related aspects throughout the entire process of *engaging with CMT*, optimising of their quality of life, or well-being, additionally and clearly emerged as a separate, distinct process, and concluding stage. It needs to be conceded, however, that this is the stage that overflows the most readily into other stages, particularly into the stage of fighting back.

In the context of CID, Livneh and Antonak (1997) states: "Adjustment, (is) theoretically conceptualised as the final phase of the adaptation process..." (p.22, parenthesis added). The fact that the process of optimising continued after acceptance of reality, amounts to an anomaly to views that regard acceptance to a CID, elusive as it may be, as the final stage, or theoretical endpoint, of the adaptation process to the condition. This includes similar formulations, such as Emery's (1994) "homeostasis" (p.66)".

The stage of optimising comprises both intrapsychic and practical, concrete strategies aimed specifically at enhancing quality of life for self and others. For a few, the former may entail a great deal of effort, or work, in order to broaden one's perspective on having and living with CMT. For most, however, a mature and balanced perspective has evolved gradually over many years of living with the disease, with relatively less intense work being required in the later years, which typically amounted to be in the optimising stage. Having a mature, balanced outlook contributes to wellness because it facilitates inner peace, reduces anger/bitterness, leads to improved interpersonal relations and generally aids stress reduction. Changes in the individual's value system, to be discussed below, are part of this process.

Practical strategies occur concurrently with the intrapsychic component and encompass actions that support the evolving balanced-life approach. The practical component includes steps aimed at looking after one's own interests in order to ensure self-preservation, as well as informing stakeholders about the status quo regarding one's CMT. In view of the reciprocal nature of outlook and practical actions, the two will be dealt with integrally.

Values and attitudes

Movement, or changes, in the CMT-affected's disease-related value systems coincides largely with the value changes that occur during the acceptance of a disability, as formulated by Wright (1983). She postulated that acceptance of loss that follows a disability, amounts to a series of value changes or shifts that must take place. They are:

1. *Enlargement of the scope of values.* The appreciating of values other than those that is in direct conflict with the disability.
2. *Subordination of the physique*. The deemphasising of the specific aspects of physical ability that contradicts his or her disability and situation. For example, physical sport if own legs are crippled.
3. *Containment of disability effects*. Not spreading or generalising his or her handicap beyond the actual physical impairment to other aspects of the self.

4. *Transformation from comparative values to asset values.* Not comparing self to others regarding areas of limitations, but rather emphasises own assets and liabilities.

All four of the above value changes, despite being formulated more for acquired disabilities, are encountered to a larger or lesser extent in *engaging with CMT's optimising* stage. The value changes represent a process that culminated in the critical junction of *accepting reality*, and it continues in *optimising*. It is not possible to establish exactly how far back the value changes started, but evidence of the process can be detected in *fighting back*, especially in the sub- process of *empowering self*. What clearly emerged however, is that the process gathered considerable momentum during the stage of *optimising*.

A large proportion of *optimising's* cognitive and behavioural strategies reflect values and attitudes typically encountered in Victor Frankl's existential theory that he formulated in the decades following World War II. Frankl (1967, 1969, 1975) postulated that people possess the spiritual ability to transcend themselves and their situation in order to search for logos, or meaning, in their lives. Typically, meaning in life may be found in four different endeavours or ways, namely through work, love, suffering and death. Meaning can also be found by way of three clusters of experiences for which people have shared meanings, and that constitutes three sets of values:

1. Creative values. These encompass those values that people experience by doing things, by contributing. It amounts to what we give, or contribute, to live.
2. Experiential values. This represents the experiencing/receiving component, that is to say, these values are realised by way of what we receive from life. Even the simplest and most basic things in life may be positively experienced and appreciated, for instance beauty, love, nature and so forth.
3. Attitude values. In situations where people have no options left, when creative actions or the experience of beauty are limited, meaning in life can still be acquired by way of the attitude that is adopted towards the suffering and

distress. According to Frankl (1969), these constructive attitudes towards unavoidable suffering constitute the highest of all values.

Examples of the above values and experiences abound in the stage of *optimising*. Although these are not limited to *optimising*, Frankl's (1967, 1969, 1975) existential concepts are encountered considerably more in *optimising* than in any other stage. A possible explanation for this is that, after they had accepted reality, people with CMT were left with very few alternatives, in terms of viable strategies, to pursue in the years ahead, in other words, that they still have to live with the disease. Finding meaning in their lives with the disease may therefore be seen as a natural progression that arises out of these circumstances.

In *optimising*, the greater part of the sub strategy *optimising of interests*, and in particular *improving the welfare of others*, reflects existential values. Here, involvement in community and religious activities are clear examples of creative values. Religion in particular emerged as very important to participants. Religiosity during *fighting back* had more of a "use-religion-as-a-resource"- focus, whereas in *optimising* the accent shifted to religious self-fulfilment and contributing. In the context of religion, Möller (1995) states: "With his view of personality centred on self-transcendence, Victor Frankl, as part of existential psychology, opened the door for a Christian perspective in psychology. The person who loses himself for the sake of Christ will be able to actualise himself and live out his life to the full" (p.254).

Contributing involves *informing others* about CMT, which includes sharing of experiences with them. It also includes relaying information to stakeholders, such as the employer and even own family and friends, about how the disease is progressing. One fundamental aim with this, although not always explicitly verbalised by all, is to enhance understanding towards individuals with CMT, as well as towards the disabled and the disadvantaged in general.

From a contributing perspective, it is my intention, after completion of this project, to follow up the matter of informing schools about neuromuscular diseases. The

aim is to involve various stakeholders, including the Muscular Dystrophy Foundation of South Africa. Another aspect that will be taken up is the possibility of a national telephonic support group. All participants expressed interest in such an undertaking, mainly because they felt a need for sharing experiences and mutual support.

Examples from the data regarding experiential values, that is to say, the experiencing of positive aspects that gives meaning to one's life, include the practising of a wide spectrum of hobbies, leisure activities and social relations by the CMT- affected, as was reported in the results chapter.

Much of the cognitive strategy of broadening perspectives encompasses Frankl's (1969) attitude values. Developing a mature, balanced outlook on life, and in particular on living with a progressive neuromuscular disease and even seeing the positive side to it, reflects the kind of attitude that Frankl says people choose to adopt in the face of unchangeable situations. So does being grateful for abilities that are still left, for CMT having taught them strength and coping skills (not all participants) and having enhanced empathy for the disabled and marginalised. Just getting on with one's life without excessive stress and worry about where the disease is taking one, and with a positive outlook, probably also resorts in this category.

Humour

The marked increase in the use of humour during this last stage is striking. By no coincidence, humour features strongly in logotherapy, the therapeutic leg of Frankl's existential theory. In paradoxical intent, the patient is encouraged to exaggerate the feared or stressful situation so that its ridiculousness is highlighted and it becomes humorous, resulting in a therapeutic effect (Frankl, 1967; 1969).

In some cases, the use of humour by participants reflects an ability to see a positive side in disease-related dilemmas, or stressors. The use of humour in this way may be seen as supporting a finding by Folkman and Moskovitz (2000), which was also discussed in Chapter 1, namely that many people see something

positive in a stressful encounter. Folkman and Moskovitz (2000) lament the fact that that the role of positive affect in stressfull encounters is neglected.

Humour is regarded as a cognitive-emotional phenomenon that amounts to the ability to focus on, appreciate or express things or situations that are funny or amusing. One of the most influential groups of humour theories is the so-called incongruity theories, which focuses on the cognitive aspects of humour. Proponents of this approach postulate that humour occurs when two divergent, incongruent ideas or situations are juxtaposed in an unforeseen, unexpected way. Laughter is the response to humour (Burkhead, Ebener & Marini, 1996).

In the above theories, humour is assumed a method of cognitive restructuring, a concept formulated by Meichenbaum (1977), and Burkhead et al. (1996) reason that these cognitive shifts may assist people in their adaptation to a disability. By way of humour, they may be able to change the perception of their situation from catastrophic to less frightening, enhancing a sense of mastery.

Doka (1993) feels strongly about the utility of humour and writes: "humour is one of our most basic coping mechanisms, one that can be very effective in times of crises since it can release tension, ease stress and strengthen social relationships" (p.20). Crabtree (1997) is on this wavelength when she recommends, "having a good laugh" (p.304) to teenagers and others with CMT who blunder during sexual activities because of their physical problems. In general, she feels that, for people affected by CMT, "a huge smile and a little wit will go a long way to winning friends…" (p.295).

The findings of this project tentatively support an interpretation that, amongst those with CMT, humour becomes a more prominent coping strategy after they had accepted reality, possibly because of enhanced inner peace and/or similar diminished stress experiences.

Social support

During optimising, individuals reflect back on the road they travelled with CMT, leading, amongst others, to an intense appreciation and gratitude towards their sources of social support. By far the most salient social support source to emerge is the spouse, or similar companion. Friends and community acquaintances also resort under this. In general, social support comprises a very important intervening variable between the disease's manifestations and subjectively experienced quality of life by those who have the disease. It amounts to a crucial contextual condition that substantially influences the way people with CMT adapt to the disease. This applies throughout their entire lives. In childhood, this role befalls parents.

By way of example of childhood support dynamics, herewith a short recount of my own situation (lived experience), which some may find interesting. I remember being very, very weak in all physical activities. I fractured my ankle a few times, as well as my arm. Like many with CMT, I was hurting, but trying to cope by pleasing others, clowning, studying excessively and so forth. I lacked the skills to make meaningful attributions regarding my limitations, as explained elsewhere. To make matters worse, my father divorced my mother and abandoned us when I was three years old. Compounding my dilemmas even more still, I had serious asthma since very young.

The only real factor that got me through, one could say my winning advantage, was a loving and supporting mother. Her kindness, empathy and support actually changed potential misery into worthwhile childhood years, with much more positives than negatives. Most people with congenital diseases and disabilities would support a notion that, without support, the obstacles would have been almost insurmountable. Social support during childhood, particularly parental support (or somebody fulfilling that role), is not an advantage, it is a crucial, life-giving necessity. Once again, a recommendation in this regard that may serve a useful purpose, is the implementation of a short information leaflet or even a program for parents who have a disabled child, or whose children have been diagnosed with a neuromuscular disease. Community health staff or even genetic counsellors may assist with this task.

Naturally, if the affected individual marries or lives with someone, the main support role shifts to the companion. The management of a chronic disease at home encompasses varying degrees of work that needs to be done by the affected individual and his or her companion, if any. Corbin and Strauss (1988) postulate that this usually requires a balanced division of work between the parties. There are three types of work: 1) illness related work, such as medication management, rehabilitation and various activities of daily living, 2) biographical work - work regarding reformulating and maintaining of an identity, and 3) everyday-life work - the daily tasks in order to keep their household going, such as housekeeping and child rearing. In the case of pronounced downward (deteriorating) trajectories, which may be encountered in CMT, an unbalanced workload typically results, in other words, the healthy partner ends up doing most of the work because the other cannot.

In addition to the extra physical work that befalls the spouse, his or her emotional experiences of the effects of the disease on the stricken partner, and on the marital relationship in general, may be intense. Robinson's (1988, p.50) findings suggest, for instance, that the immediate impact of a diagnosis of multiple sclerosis tends to be generally more negative for the spouse than for the affected individual! Naturally, the nature of the relationship prior to the diagnosis may influence this outcome. Another interesting finding of his, which he maintains is also encountered in other research, is that spouses perceive the level of functional disability in their affected partner as more severe than the affected person him or herself.

Corbin and Strauss (1988) maintain that, under conditions of profound deterioration, partners are often persistent in keeping themselves and each other going despite negatives such as overwork, lost dreams and so forth. They sustain hope and commitment to each other. They "sustain within themselves and each other cherished identities" (p.123). In order for a shared experience of where the disease is taking them to be maintained, communication and sharing of information are crucial.

What happens in cases where the course of the disease is relatively benign? This scenario, where deterioration may be slow and perhaps plateau for long periods, is likely in many cases of CMT. Robinson (1988) maintains that one of the most likely marital strategies under these conditions amounts to endeavours to continue with roles as before diagnosis, in other words, to keep the threatening social changes because of the disease at bay. He writes: "There is a pact between husband and wife to diminish the social, if not the physical, consequences of multiple sclerosis. Such a strategy is congruent with the idea of fighting the disease" (p.59). In other instances, living with the symptoms of a condition may result in enhanced appreciation of each other, in ways not known before. This, in turn, may actually lead to experiencing enhanced quality of life, particularly if it takes place in a religious context (Robinson, 1988).

All the above strategies described by Robinson (1988) and Corbin and Strauss (1988) are rather well represented in the stage of *optimising*. A quotation from Robinson (1988), a snippet from his dialogue with a participant, almost reflects the aspects of marital and religious deepening in vivo, and is considered an apt way to conclude this section: " My family accept me as I am. I believe we now have a better life than we did before, that may sound strange, but we appreciate each other more. With God's help we have gained from the MS. God has never let me down when I have confided in Him". This quote echoes words from many CMT affected people who are in the stage of optimising, including my own.

OUTCOMES/CONSEQUENCES

According to Glaser (1978), outcomes include anticipated and unanticipated consequences. Consequences in grounded theory tend to be of a temporal nature; a particular consequence or outcome often comprises the causal condition (cause) for the next action-strategy (Glaser, 1992).

In this study, participants' action strategies were ultimately directed at achieving as good a (subjectively experienced) quality of life, or wellness, under conditions of living with CMT as possible. This "global" outcome may be conceptualised as

being the culmination of many "intermediate" outcomes, which occurred throughout the entire process of *engaging with CMT*. Even though the many specific outcomes of the various action strategies are included in this, the major intermediate outcomes amount to the critical junctions, or turning points, at the end of each sub process (stages), of the core category.

In *orientating*, the turning point *realisation of the salient implications,* that is to say of being diagnosed with CMT, may be regarded as an outcome of the stage of *orientating*. In line with Glaser's (1992) assertion that consequences in one stage often comprise causes for the next stage, realisation of the salient implications was the causal condition for the strategy of *fighting back* to emerge. The same applies to *accepting reality* at the end of *fighting back,* which comprises the causal condition for the *optimising* of well-being.

Strauss and Corbin (1998) state about outcomes: "… explaining how they (consequences) alter the situation and affect the phenomenon in question, provides for more complete analysis" (p.134). It almost goes beyond saying that this was, and is, exactly the goal with the rather intensive elucidation in Chapter 4 (results) of the outcomes of this study. The stage of *optimising* has no theoretical end point; it is a continuous process that may diminish, but will probably never entirely cease, in the life of the affected person. A relative point of "saturation" amounts to a variant of an outcome, *qualified wellness.*

Continuing the brief recap, the core strategy of *engaging with CMT* resulted in the outcome of *qualified wellness*. The core strategy largely, but not entirely, succeeded in resolving participants' main concern, *dealing with unpredictable disease manifestations*. The component of the main concern that the core process succeeded in resolving, and for many the most important part, is overcoming and managing the many problematic, practical day-to-day effects and symptoms of the disease. The components that were <u>not</u> entirely resolved encompass the uncertainty/unpredictability facets of the main concern, namely atrophying (physical deterioration) and anxiety about the CMT status of descendants.

Regarding the latter, that is anxiety about the CMT status of descendants, the findings reveal that the core strategy, engaging *with CMT*, succeeded in resolving this concern of participants in only 50% of the cases.

In the case of the third leg of the main concern, namely deterioration, a rather unique outcome emerged. Participants *accepted the reality* that their extensive fighting efforts have not markedly altered the physical atrophying course of the disease. This coming to terms (Corbin & Strauss, 1988) is however, only partial, since the vast majority still feared the worst-case scenario of atrophying to the point of becoming dependent. They were experiencing considerable anxiety about the possibility of having to sit in a wheelchair, even if not full time.

In this specific context, *accepting reality* thus amounts to accepting the futility of fighting, but fails to reduce the high levels of chronic anxiety about becoming dependent. It needs to be said that there are researchers who maintain that failure in this context may be understandable, at least to some extent. Considering the large size of the threat in many chronic illnesses and disabilities, Glass (1994), in Kendall & Buys (1998) points out that many affected people believe that they will never be able to adjust to their disability, because it would require acceptance of a situation that is inherently unacceptable.

Livneh and Antonak (1997) concur when they point out that, in the case of many chronic diseases, as well as life-threatening ones: "… acknowledgement and acceptance would constitute some form of recognition or internalisation that the condition is likely to worsen or that death is imminent. Psychosocial adaptation under such conditions is almost inconceivable" (p.26). However, these strong words may not be directly applicable to CMT since *engaging with CMT was* indeed successful in aiding adaptation for many participants.

PART 3

CONCLUSION

This research project not only, in the broader sense, addresses the dearth of research on the psychosocial facets of living with CMT, but also specifically culminated in a substantive grounded theory on the disease-related problems and management strategies of CMT-affected people. As far as can be established, this is the first such grounded theory study.

My study contributes towards understanding the much-understudied area of living with CMT and congenital neuromuscular diseases in general. From the grounded theory approach a new dimension to the way adult people who have CMT manage the disease emerged and was formulated into a substantive grounded theory of *engaging with Charcot-Marie-Tooth (CMT) disease*. This theory comprises the way CMT-affected adults manage the effects of the disease in order to optimise their quality of life with the neuromuscular condition. The grounded theory approach proved invaluable in generating this theory in response to the research questions, which were thus satisfactorily answered.

The findings indicated that the main concern, or problem, with which CMT-affected people grappled, embraced a trifurcate of sub-concerns. The overall main problem emerged as *dealing with unpredictable CMT manifestations,* and the three sub-problems as how to handle the disease's *symptoms and effects*, the *CMT status of descendants* and physical *deterioration*. The main or core strategy employed to resolve the above-mentioned main concern of dealing with unpredictable CMT manifestations, emerged as the basic social psychological process of *engaging with CMT*, which, by coincidence, also comprised a trifurcate of sub-strategies. The three sub strategies represented overlapping sequential stages. They were *orientating, fighting back* and *optimising*. The entire process led to a dynamic outcome of *qualified wellness*, leaving most participants *faring well*, whilst others still experienced undue *fear and agony.*

Other unique contributions of this study comprise the following:

- A stage analysis regarding adaptation to a congenital neuromuscular disease with clear inherited etiology, such as CMT.
- A formulation of the stages of orientating and optimising.
- Fighting back is encountered in many studies, but does not comprise a distinctive stage, as it does in *engaging with CMT*.
- The importance of incorporating affected people's disease- related experiences and strategies prior to diagnosis, especially those during childhood, retrospectively into the analysis and eventual grounded theory, is illustrated.
- In congenital neuromuscular diseases with a gradual degenerating course, such as CMT, there is no theoretical end-point to the adjustment process, as is often the case in acquired conditions (Livneh & Antonak, 1997). Instead, a dynamic outcome that merely signifies a relative "point" of saturation is encountered.
- Light is thrown on the conditions, especially in the school context, which are detrimental to the self-concepts of children with high visibility neuromuscular diseases such as CMT, as well as on how these conditions affect their self-images.
- Identification of the phenomenon that some parents and/or grandparents may actively employ their own core strategy, which they employ to solve their main concern, in the lives of their affected descendants, either to help the child or as a way to deal with their own anxiety and guilt feelings, or both.

LIMITATIONS

As was conceded in the methodology chapter, theoretical sampling could not be fully implemented due to difficulties in tracing suitable participants, as well as their wide distribution and the vast geographical distances between them. Other shortcomings include:

- A rather skewed age range. Nine of the 11 participants were older than 40. This limitation is also reported in another CMT study (Arnold et al., 2005). Including the disease related experiences of younger people could have added more diverseness and heterogeneity.
- Very long passage of time (12 to 33 years) since diagnosis. Inclusion of more recently diagnosed cases could highlight the dimension of how future management of the disease is envisaged, in other words, prospectively.
- Lack of ethnic diversity. One of the outstanding limitations, and personal disappointment, of this research is the absence of participants from the black community, as well as other communities that were not represented. The reasons for this were highlighted in the methodology chapter. CMT affects all cultures and ethnic groups equally (CMT United Kingdom, 2004a). As was mentioned in Chapter 1, the demographics of South Africa's population suggest that there must be a great many people from ethnic groups other than white that have CMT. Most probably, they do not know that they have it and not only struggle physically, but also grapple with self-esteem issues. For most, it would add considerably to their quality of life by knowing what exactly is wrong with them, and by being empowered with proper information about the disease.

  The present efforts to obtain the desired participant diversity were unsuccessful and it remains a challenge for future projects. As it stands, this analysis and the substantive grounded theory may not be applicable to all cultural and ethnic groupings.

RECOMMENDATIONS

General recommendations

- From the perspective of avoiding, or at least substantially minimising, harm to the already fragile self concepts of children with disabilities and those who are physically weak, as is encountered in CMT, careful selection and proper training of sport/gymnastic teachers (and others involved in these activities) should receive very high priority. Humane educators who

display empathy towards these children, and who are able to unconditionally accept the possible weaknesses of others, will be much more effective and appear fairer, more compassionate and kinder to the learners.

- Linked to the above, is the active promotion of awareness for disabled and physically weak children amongst the schools' concerned parties, including their control boards and personnel, as well as in the broader community. Schools, for example, may market their proficiency in accommodating and educating physically disabled children as an asset in marketing campaigns. Naturally, their physical set-up should reflect this orientation; it should be disabled-friendly. If not the case already, the training programmes of sports and physical education teachers should include coursework on the needs and education of this group of children. The possibility of overwork weakness in particular should be addressed.

  Educators in general should be made aware of the needs of children with disabilities and impairments, including neuromuscular diseases. Higher awareness and training may assist them in handling these children in such a way that they experience dignity and believe in themselves, despite the physical limitations.

- The possibility of implementing dedicated exercise and sports programmes for physically impaired children should be investigated. To an extent, sport events such as the Para Olympics may very well signal that this is indeed the way forward. It need not necessarily involve vast amounts of additional capital, since any number of schools can group and coordinate in presenting these programmes. There, at any stage, may only be a handful, if any, physically impaired children in a given school.

- In addition to existing exercising regimes and techniques, less well-known and novel approaches such as Qigong and Tai Chi, which have been shown in research to be rather effective for people with neuromuscular diseases, may be a viable alternative for many individuals who cannot walk on uneven surfaces nor do gymnasium exercises.

- To an ever increasing extent, associations and organisations for the disabled and other chronic illnesses/impairments, such as muscular dystrophy and CMT associations, have the task (amongst many others) to promote greater awareness for the more positive attributes/skills (including those skills unaffected by the disease) of the people they represent. In the case of, for example, CMT where verbal communication skills are not usually affected, such strategies may assist in eliminating rather bizarre situations such as the one portrayed in Part two of this chapter where a disabled person is ignored and communicated with through an accomplice.

- A few participants expressed the need for a rather informal, conversation orientated support group for CMT-affected people. By way of this structure, affected people may be able to share experiences, information and be able to support each other. Because of practical realities, such as affected people being scattered across the country, it will most probably be based on, for example, telephonic, SMS, e-mail and Skype communications. However, participants in general did not express a strong view to officially break away from the Muscular Dystrophy Foundation of South Africa and to form a separate CMT organisation. Perhaps a different picture may emerge if more CMT affected people could be located and approached regarding this matter.

- All participants except one felt that many of the medical professionals dealing with their diagnosis and, to lesser extent treatment, did not instil confidence in terms of their knowledge about Charcot- Marie-Tooth disease (excluding neurologists). This state of affairs is rather disturbing in the light of the rather high prevalence rate of CMT and should receive attention. Molecular genetics and other research have uncovered many of the intricacies of this disease, and information about it is readily available, for example on the Internet and through various CMT organisations worldwide.

- Many participants expressed dissatisfaction with the interpersonal communication skills of medical professionals who handled their diagnosis,

as well as those who undertook the task of genetic counselling. If this complaint is persistently found in a large number of cases, as well as in other neuromuscular diseases, those responsible for medical training should take note and act accordingly to rectify the situation.

- Participants' enthusiastic interest in gene therapy and stem cell research underline the need for CMT and similar organisations to regularly inform their members regarding new developments and progress on these frontiers.

Recommendations for future research

In addition to eliminating some or all of the limitations and shortcomings discussed above, recommendations for future research are as follows:

- The paucity of research regarding the role of stress in the exacerbation of CMT in general, as well as in the commencement of atrophying after a plateau, needs to be addressed. Both qualitative and quantitative studies (and mixed methods designs), may add value in this regard.

- The confusion that exists regarding the role of exercising in neuromuscular diseases needs to be cleared up. Despite the reality of individual differences in the nature and seriousness of symptoms, burning issues such as what type and intensity of exercises are more suitable than others, as well as how to avoid overwork weakness, needs to be clarified by way of appropriate research. Regarding the type of exercise, the suitability for use in CMT of new and perhaps novel approaches, such as Qigong and Tai Chi, should be determined by way of research.

- Another recommendation involves the family system, within which the CMT affected individual functions. Exploring the impact of CMT on family members, how they deal with CMT in the family and their experiences in this regard, are considered fruitful endeavours for future research.

- The development of a disease specific quality of life instrument, for which a need exists (Shy & Rose, 2005), may be attempted; utilising, amongst others, qualitative data such as the present study's results.

- Lastly, replication (investigating the same research questions as in this study by way of Glaser's grounded theory approach), using participants with other congenital neuromuscular diseases and neuropathies, may establish to what extent *engaging with CMT* is applicable in those conditions.

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Appendix A

BIOGRAPHICAL DATA AND BACKGROUND INFORMATION

Date: _____

1. Name and Surname: _____
2. Tel: _____
3. Cell: _____
4. E-mail: _____
5. Gender: Male ☐ Female ☐
6. Date of birth: _____
7. Marital status: _____
8. Occupational status: Retired ☐
   Unemployed ☐
   Working from home ☐
   Employed fulltime ☐
   Employed casually ☐
9. Highest level of education
(highest school standard, diplomas, degrees, etc)

_____

10. Number of children, if applicable:

_____

11. Age at which you were first diagnosed with Charcot-Marie-Tooth disease (CMT) :

_____

12. Type of CMT (if known)

_____

An example of a classification of the different types is as follows:
Type I (1a, 1b, 1c, X- linked)
Type 2 (2a, 2b, 2c, 2d, 2e)
Type 3 (Dejerine-Sottas)
Type 4 (4a, 4b, 4c)
Type 5
Type 6 (e. Vizioli)

13. Perceived severity of CMT:
Please rate your current degree of physical impairment/disability due to CMT, as you experience it. Indicate with an X on the following scale:

Mildly affected						Very severely affected
1	2	3	4	5	6	7

14. Do you use any orthopaedic or other aids (e.g. braces, orthoses, walking stick etc)? If you do, please specify.

_____

15. Have you had any surgery because of CMT?

Yes ☐ No ☐
If you answered yes, please specify the approximate year and type of surgery.

_____

16. Has any medication been prescribed to you because of symptoms related to CMT, for example pain, cramps, etc.?

Yes ☐ No ☐

If you answered yes, please specify the symptoms and, if known, the name of the medication.

_____

_____

17. Have you tried alternative treatments, such as acupuncture, homeopathy etc.? Yes ☐ No ☐

If yes, please specify:

_____

_____

18. Do you engage in physical exercise? Yes ☐ No ☐
If yes, please elaborate.

_____

_____

19. What leisure activities to you participate in? (if any)

_____

_____

Appendix B

INFORMATION AND CONSENT DOCUMENT

Dear _____ (prospective participant)

(A) I hereby invite you to participate in my doctoral research study. I aim to investigate the experiences of adults with Charcot-Marie- Tooth disease (also known as Hereditary Motor and Sensory Neuropathy), for example how the condition affects their lives and how they are dealing with it.

You are required to engage in an interview with me and to complete a few questionnaires. There is a possibility that follow up interviews may be necessary.

Kindly read and sign part B below if you are willing to become a research participant.

Nicol Alberts
Researcher

(B) I _____, hereby give Nicol Alberts permission to use the information obtained during the interviews, as well as from the questionnaires and observations, for research purposes only. I understand that names and places will be changed to protect my identity and the identity of other relevant people. I understand that participation is voluntary and that I can withdraw at any time, should I wish to do so.

Signed _____ (participant), on

Date _____ at

Place _____

Researcher _____

Appendix C

Ankle- foot orthosis

http://www.manordrug4.com/servlet/the-256/ANKLE-FOOT-ORTHOSIS--fdsh-/Detail

Appendix D

Thank you for referring this patient who has been aware of a muscular problem since the age of 25. At that stage he noticed thinning out of the smaller muscles of his right hand. Since then, there has been a progressive loss of muscle affecting the lower extremities and to a lesser extent his other hand.

On specific questioning he states that his mother has flat feet and that his father has high-arched feet. His two brothers and a sister, according to him, are normal. He also has a son who is now aged 6 years who, he states, reminds him of himself when he was that age.

On examination he has the classical muscular atrophy of Charcot-Marie-Tooth disease and has the nerve thickening of the hypertrophic variety. The reflexes are absent, sensation is mildly impaired in the lower extremities so that vibration sense is missing in toes.

Electrophysiological Study

1. Electromyography

   Concentric needle electrodes were used to examine muscles of the upper and lower extremities. There was marked fall-out of motor-unit activity in the distal muscles and whatever motor-unit activity was there was of high-voltage and polyphasic in appearance.

2. Nerve Conduction Studies

   This was carried out on the left arm. The left median nerve conduction velocity was slow at 21 Meters/second, the ulnar 23.1 Meters/second.

3. Sensory Conduction Studies

   This was carried out on the right hand and the conduction velocity was slow at 16.3 Meters/second.

Opinion

The examination and electrophysiological study confirm the hypertrophic variety of Charcot-Marie-Tooth disease in this patient. I have explained the genetics of this situation to him and advised him to join the muscular

dystrophy foundation as they have a particular sub-group of sufferers with this same disorder.

With kindest regards to you.

Appendix E

FINAL DIAGNOSES: PERONEAL MUSCULAR ATROPHY (CHARCOT-MARIE-TOOTH DISEASE)
PROFOUND DEPRESSION

I last saw this man on _____and then again at follow up today _____. He suffers from a genetic inherited condition known as peroneal muscular atrophy which has deteriorated remarkably since I last saw him 3 years ago. It has deteriorated to the point where he is having great difficulty writing and his work is now being impaired. He is a qualified _____and has had to alter his work style and some of the work that he does because of his inability to write on the blackboard. This together with his progressive deterioration in gait over the period of the last 6-12 months particularly, has caused great consternation and anxiety for him. It has resulted in a profound depression manifesting as follows:

1. Inability to sleep - the patient has big large rings around his eyes
2. Marked deterioration in sexual function and libido
3. Marked decrease in appetite with significant weight loss
4. The patient has lost confidence completely at work and is actually considering asking them to demote him because he does not feel he is coping.
5. Significant memory problems and basic errors on simple work which is upsetting him greatly.
6. The patient has become forgetful and cannot even remember to keep appointments or meetings properly - this is worrying him greatly and perpetuating the cycle of depression. His extreme insomnia complicates the issue even further.

During the interview the patient appeared to have suicidal tendencies explaining to me that every man has his limit and he has apparently reached it and I am very concerned about his well-being. A referral to a psychiatrist is mandatory and anti-depressive therapy will be initiated immediately.

EXAMINATION He was awake and orientated. Mood was very depressed. His speech was normal. No temperature or neck

stiffness. Blood pressure 130/75 and pulse 77 regular. Rest of systemic examination normal.

NEUROLOGICAL EXAMINATION The pescavus is clearly noted worse on the right side. He has significant wasting of the intrinsic muscles of his hands and feet with absent reflexes at the knees and ankles and decreased supernator reflexes in the upper limbs. He has sensory deficit in glove and stocking distribution on his hands and feet from the wrist and ankles downwards. He has marked decreased function with dorsi-flexion of his ankles bilaterally at 0/5 and planti-flexion and all other movements of the ankle at about 3/5. He has weakness of the small muscles of his hands and to a lesser degree of his forearm. He is strong proximally in both upper and lower girdle. Sphincter function still normal.

He walks with a typical drop foot gait bilaterally, an orthopaedic appliances will be obtained for him so that we can support his foot in a brace so that he can walk more comfortably.

CONCLUSION This man has peroneal muscular atrophy. His syndrome is gradually progressive in nature and in the past 3 years he has deteriorated rather quite alarmingly. He is having major problems at work mainly due to his handwriting. He has developed a significant depression with resultant insomnia, eating disturbance, so-called memory problem and profound forgetfulness, all of which can be attributed indirectly to his reaction to his disease.

I have put him on Tryptanol 25mgs nocte for 7-14 days, then to be increased to 50mgs nocte. I will be referring him to a psychiatrist immediately. Should we have any further episodes to suggest suicidal tendencies it might be better to admit him to hospital.

Yours faithfully,

Appendix F

We apologise for the confusion that was caused by the previous correspondence but after reviewing the notes it is clear that we felt you had a familial sensory-motor neuropathy that could not clearly be delineated in either of the two major subgroups that are commonly seen. Furthermore, on examining the rest of your family (parents and 2 siblings) it appeared that everybody was normal clinically and electrophysiologically apart from your mother who despite being asymptomatic at the age of 51, did have minimal evidence of sensory conduction disturbance in all the sensory nerves tested.

This suggests that you probably inherited the diseased copy of a gene from your mother **but** it is not the usual pattern seen in autosomally inherited diseases (non-sex chromosomes); in the latter group the electrophysiological pattern is usually identical in parents and offspring even if the clinical picture is not so. This brings us to the presumption that in your case the particular diseased gene came from your mother's X-chromosome as this pattern is more consistent with an X-linked mode of inheritance. However, I must emphasize that although based on sound clinical data, it is still only a clinical presumption and not confirmed genetically. Unfortunately we (or elsewhere) cannot perform any further genetic tests on your family to answer clinical questions and it would be almost impossible to get a research team to examine the genetics of this very small family.

The practical implications of our clinical presumptions therefore suggests that you acquired the possible faulty X-chromosome from your mother and you cannot therefore pass this on to a male offspring as you would (and as you correctly understood in your letter) pass on a Y-

chromosome to your son. In other words, if our clinical presumption is correct, your son will not have this disease. However, if you should have a female child, she would have the faulty X-chromosome that she has inherited from you and she would inherit a normal copy from her mother. In effect it means that your female offspring would be in exactly the same clinical position as your mother in other words a carrier with possibly electrophysiological evidence of an abnormality but clinically completely asymptomatic. It is also important to realise that even though your daughter might be in a similar position to your mother with respect to the potential of passing the diseased copy of the gene on to offspring it may be very possible in the future, when she does come to the decision, to have a predictive laboratory test.

We trust that this answers your questions. Please let us know if anything is unclear.

Yours sincerely