



Report: International Conference On Birth Defects And Disabilities In The Developing World

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Vue d'ensemble

Ce rapport de la conférence Internationale sur des malformations et des déformations neonatales dans le monde en voie de développement donne l'opportunité aux infirmières et aux sages-femmes de savoir où obtenir l'information et les ressources sur des malformations et des déformations neonatates. Les infirmières et les sages-femmes sont souvent les premières professionnelles de santé à détecter n'importe quel malformation neonatale, ou n'importe quelle famille nécessitant la consultation génétique. Elles doivent, donc, être bien informées au sujet des développements internationaux dans ce domaine. Le rapporteur est conscient qu'aucun rapport ne peut faire justice aux résolutions de n'importe quelle conférence internationale. Cependant, il donne ce rapport dans l'esprit de partager certaines de ses expériences et perceptions sur les autres professionnel de la santé et chercheurs Durant les travaux de la conférence. Des références et les sites de Web sont donnés pour permettre aux lecteurs intéressés d'élargir leur connaissance au sujet de la génétique, et/ou au sujet des malformations et déformations neonatales.

Resumo

Este relatório sobre a 'Conferência Internacional sobre Defeitos e Deficiências Físicas de Nascimento nos Países em Desenvolvimento' providencia, ao pessoal de enfermagem e às parteiras, informação e recursos valiosos sobre onde obter informação vital sobre os defeitos e deficiências físicas de nascença. As enfermeiras e as parteiras são muitas vezes os primeiros profissionais na área de cuidados de saúde a detectarem qualquer deficiência de nascença, ou a identificar qualquer família que necessite de consulta de orientação genética. Assim, necessitam de estar adequadamente a par dos desenvolvimentos internacionais nesta área. O autor deste relatório está bem ciente do facto de que nenhum relatório pode, de uma forma justa, relatar os procedimentos de qualquer conferência internacional, mas providencia este relatório numa tentativa de partilhar algumas das suas experiências e percepções durante a conferência com outros profissionais e pesquisadores na área de cuidados de saúde. Oferecem-se aqui referências e detalhes de contacto na Internet para os leitores que estejam interessados em alargar os seus conhecimentos sobre genética, e/ou especificamente sobre defeitos e deficiências de nascença.

The International Conference on Birth Defects and Disabilities in The Developing World was hosted by the Department of Health: South Africa, with the support of the World Alliance of the Organisations for the Prevention of Birth Defects

Introduction

As nurses are often the first health care professionals to detect any birth defect, or any family in need of genetic counseling, they need to be knowledgeable about international developments in this field. No report can do justice to the proceedings of any international conference, this is merely an effort to share some of the author's experiences and perceptions during the conference with other health care workers and researchers. References and web sites are supplied for interested readers to broaden their knowledge about genetics, and/or about specific birth defects.

Purpose of the Conference

The conference was organised to address issues of:

- monitoring birth defects
- developing genetic services
- ethics in providing genetic services
- providing psycho-social support to people with birth defects and disabilities in the developing world.

Conference Programme

The conference was attended by approximately 200 delegates and presenters from South Africa, Australia, China, Cuba, India, Germany, Kenya, New Zealand, the Netherlands, Nigeria, the Philippines, the United Kingdom, the

United States of America and Zimbabwe.

The Minister of Health of South Africa, Dr Manto Tshabalala-Msimang, attended the official opening ceremony on 6 August 2001 and welcomed all national and international delegates. The Minister of Health also launched the Department of Health's Policy guidelines for the management and prevention of genetic disorders, birth defects and disabilities (published during 2001). The presence of the Minister of Health was much appreciated by all delegates. A number of overseas delegates mentioned that this was their first conference on genetics where the Minister of Health of the host country welcomed the delegates.

The plenary sessions addressed monitoring and surveillance, genetic services, congenital disability in the community as well as legal and ethical issues. Merely an overview of the major issues discussed at these plenary sessions can be provided. More information can be obtained by visiting the conference Web site at www.globalconf.co/birthdefects which will continue to be available incorporating new information as it becomes available.

Monitoring and Surveillance of Genetic Services

Since the 1990s research conducted in many parts of the world have repeatedly shown that a daily intake of 400mcg of folic acid reduces the incidence neural tube defects by up to 50%. Notwithstanding this scientific evidence, very few countries in the world fortify their food (such as bread) with folic acid additions, which could constitute a primary prevention public health measure - reducing the number of babies born with birth defects/disabilities. Research conducted in China indicated that subsequent to supplementing women's diets with folic acid,

the incidence of babies born with neural tube defects decreased by 40% in the southern and 85% in the northern parts of China respectively. Should folic acid be sufficiently utilised globally, the incidence of neural tube defects could decline globally in ways similar to the decline of birth defects attributable to maternal rubella.

The three major birth defects reported in South Africa, all of which could be addressed by means primary prevention measures, are

- fetal alcohol syndrome (FAS)
- neural tube defects (NTDs)
- congenital HIV.

Throughout the conference many delegates repeatedly emphasised that fetal alcohol syndrome (FAS) is a silent epidemic in many parts of the world, including South Africa where its prevalence has been estimated to be in excess of 30% in certain Western Cape areas, and that this birth defect is 100% preventable, making it the single most preventable cause of mental retardation. To complicate matters further, mothers who give birth to FAS babies, might also be HIV positive. Although congenital HIV is not necessarily regarded as a birth defect by researchers, it could be argued that if birth defects refer to any defect preceding birth, then both FAS and congenital HIV should be considered to be birth defects.

Although numerous sophisticated techniques are available to detect birth defects, not all countries could afford such high technology measures. Nevertheless, much could be achieved by specifying the minimum number of defects that each country should screen for, maintain accurate records and supply support to affected individuals and their families. The effective utilisation of information technology, such as South Africa's National Telemedicine Programme, can help to render effective health care (including prevention, diagnosis, treatment and care) to all people, including those living in outlying rural areas of the world. A demonstration was given where a radiographer from Zastron in the Free State Province, showed an x-ray to a medical doctor in Pietersburg, in the Northern Province, who diagnosed bronchitis.

Genetic Services

Presenters during these sessions emphasised that local problems need local solutions, but that solutions need to be based on statistics emanating from accurate statistics.

Such records could indicate that Sickle Cell Anaemia is a genetic condition occurring seldomly in Southern Africa, but commonly in Western and Central Africa. However, this condition also occurs in Cuba and in the Caribbean Islands. (Many children, and even adults, with Sickle Cell Anaemia die from malaria and

bacterial infections. More information about this condition can be obtained from www.sickelcellfoundation.com).

Not every primary health care worker could be expected to be an expert genetic counselor, but every one should be able to recognise and refer and suspected baby with a birth defect/disability. Genetic services should include effective preventive programmes, preferably implemented by a country's primary health care services, such as

- accessible and reliable family planning services
- adequate diets and micro nutrient supplements (especially folic acid to be taken by all women during their child bearing years, NOT only during pregnancies)
- educating communities about the relationships between maternal alcoholism and FAS and about the risks faced by babies born HIV positive
- avoiding known teratogenic agents in the environment such as radiation hazards; smoking, drugs
- identification and referral of persons at increased risk of having children with birth defects; educating communities about the risks concomitant with advanced maternal age during pregnancy
- technical support from genetic experts for all health facilities (such as utilising telemedicine facilities)
- providing genetic counseling services.

From many countries, including the USA, delegates emphasised the need for cultural sensitive genetic counseling, preferably to be done in the clients' home language and only by counselors who are proficient in that specific language.

Congenital Disability and The Community

Many communities continue to regard a child with a birth defect as punishment for some wrong doing. Persons with birth defects, and their families, struggle to lead normal lives in their communities. Blind and deaf persons could face huge challenges in striving to obtain minimum levels of education; and children whose mental capacities are affected (including FAS and Down's Syndrome children) might be unable to reach even their limited developmental potential because of lack of resources. Some communities could regard persons with albinism to be suffering from an infectious disease and might warn their children never to play with albino's lest they become infected with albinism themselves. Public transport (including taxis) might refuse to render any services to persons with birth defects, fearing that these

conditions might be infectious. This could cause serious problems to parents who might need to get their children with birth defects to clinics or hospitals.

Many persons with birth defects, and their families, live in social isolation. Voluntary societies can do much to bridge the gap between community ignorance and accessing resources for these persons and their families. Voluntary persons and organisations can help the health care professionals in numerous ways to achieve the ultimate aim of improving the functional abilities of affected children and their families. In some cases persons with birth defects might prove to be effective counselors, provided they have received sufficient training and provided they can render culture sensitive genetic counseling.

Legal and Ethical issues

Screening for and diagnoses of birth defects could be expensive procedures. In countries where pregnancies cannot be terminated legally, the ethical implications of antenatal screening pose numerous ethical problems because the mother has no legal choice but to carry the pregnancy to full term. In countries where pregnancies can be terminated legally, genetic counselors have a great responsibility NOT to impose the counselors' preconceived decisions on any clients but merely to provide the required information to enable the parents to make informed decisions.

Persons with birth defects could struggle to access health care services, and to obtain such services. It is known that benefits resulting from expensive medical procedures such as open heart surgery should be weighed against the expenses, according to national, regional and institutional priorities and policies. However, the ethics of refusing open heart surgery to children with Down's Syndrome could be questioned. Decision-making processes should be open to public scrutiny and procedures for lodging appeals against decisions made should also be in place, and should be known to any stake holders, especially parents whose children are affected by decisions granting them or refusing to grant them expensive medical treatments and/or surgery.

Many citizens of developing countries continue to make use of the services of traditional healers. Parents of children with birth defects are no exception. It is vital that mutual trust and referral systems between traditional and western health care sectors should be established, especially in developing countries, so that the prevalence of specific conditions in specific areas can be more accurate, and so that treatments can be provided wherever possible.

All health care professionals should be alert to the possible uses and abuses of genetic knowl-

edge. With the advances in information and biotechnology, this information could be processed and used in numerous ways - not necessarily to the person's benefit. The WHO Expert Consultation on New Developments in Human Genetics warned in Geneva on 1 July 2000 that "... many individuals, groups and nations have concerns about the use and exploitation of genetic data and genome technology ... and genetic advances will only be acceptable if their application is carried out ethically, with due regard to autonomy, justice, education and the beliefs and resources of each nation and community". One such implication could be that parents who know the sex of their unborn child might prefer to have a pregnancy terminated in the hope of having a boy with the next pregnancy. Besides all the ethical questions that could be raised by such a decision, the male-female constitution of future generations might become displaced with many more men than women in such societies.

With the completion of the Human Genome Project (HGP) the practice of medicine could be revolutionised, but the gaps between medical care available to the rich and the poor could also be widened thereby. Should genetic information become available about a specific person's potential development of diabetes or breast cancer, this information could be used to selectively exclude these individuals from insurance programmes - and the ethics of decisions based on such information need to be scrutinised.

Conclusion

The major emphasis in developed countries appears to be on preventing birth defects, whilst the major emphasis in developing countries remains to provide care for the persons with birth defects, and for their families. Despite the potential revolution in medical care which might arise from the knowledge obtained during and after the Human Genome Project, health care providers need to remain alert to potential ethical issues which might not be to their patients' benefit.

Many birth defects can be prevented by combating alcoholism, malnutrition and HIV amongst all women until the end of their child bearing years - not only during their pregnancies but also prior to and after their pregnancies. These efforts will only meet with success if they are directed at entire communities - including all men as well. Enabling children, including girls to attend school, and helping communities to overcome poverty and to eat balanced diets could render inestimable contributions towards reducing the incidence of birth defects in many developing countries of the world. 

References For Further

Reading

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World Health Organization. 2000. *Primary health care approaches for prevention and control of congenital and genetic disorders. Report of a WHO meeting, Cairo, Egypt, 6-8 December 1999*. Geneva.

Web sites which could provide further information

<http://www.globalconf.co/birthdefects>

<http://www.birthdefects@healthnet.org.za>

<http://www.haemophilia.org.za>

<http://www.hst.org.za> (South African report on saving babies)

<http://ibis-birthdefects.org> (International Births Defects Information System)

<http://pandorawordbox.com> (Medical Etymology and Mythology)

<http://www.sickelcellfoundation.com>

<http://www.waopbd.org> (World Alliance of Organizations for the Prevention of Birth Defects)

<http://sunsite.wits.ac.za/saida> (Southern African Inherited Disorders Association)

Support groups for persons with birth defects/disabilities

Albinism: Tel: 011-838-6529

Attention Deficit Syndrome: www.poshad.co.za (poshad@iafrica.com)

Brittle Bone Group: Tel: 031-332-9724

Cri du Chat Syndrome: Tel: 011-795-1647

Cystic fibrosis: Tel: 031-765-6687; 021-557-0323; 011-867-5538 (cfa@iafrica.com)

Down Syndrome South Africa: Tel: 012-460-0165; 031-28-7338 (downskzn@iafrica.com); dssa@icon.co.za)

FAS: SANCA: Tel: 011-482-1070

Fragile X Syndrome: Tel: 082-718-4967

Haemophilia: Tel: 011-787-6710; 083-225-9850; 083-896-3833

HIV/AIDS: AIDS Helpline: Tel: 0800-0123-22

Huntington Disease: Tel: 021-448-8766; 011-

865-1100

Marfan Syndrome: PO Box 7294, Pretoria. 0001. Tel: 012-333-9366

Muscular Dystrophy Foundation: Tel: 011-789-7653 (national@mdsa.org.za)

Noonan Syndrome: TNSNG, INC. PO Box 145, Upperco, MD21155, USA

Pader-Willi Syndrome: Tel: 011-442-5700

Porphyria: Tel: 011-894-2936

Southern African Inherited Disorders Association (SAIDA): 031-205-6915; 011-489-9213; (saida@mail.saimr.wits.ac.za)

Spina Bifida and Hydrocephalus: Tel: 011-646-9156; (andrea@icon.co.za)

Tay Sachs: Tel: 011-440-1613; 082-953-6303

Thalassaemia: Tel: 031-401-3442

Turner Syndrome: Tel: 011-453-0024; (turnerza@yahoo.com) 

Protocol for the Identification of Discrimination Against People Living with HIV

One of the objectives of National AIDS Programmes (NAPs) is to reduce the personal and social impact of HIV infection, including discrimination against those living with or suspected of having HIV/AIDS. This Protocol seeks to further that objective by providing a tool for measuring arbitrary discrimination in a range of key areas in everyday life.

The Protocol is aimed at national AIDS Programmes, but it is not just for them. It may also be used by others - interested organisations, groups or persons, including those living with or affected by HIV/AIDS - in order to detect arbitrary discrimination. Indeed, a broader range of users will help in making the Protocol a more effective human rights tool. The Protocol is not intended to be used only for the detection of discrimination, but also to encourage the adoption and enforcement of measures against such discrimination, as well as the dissemination of good practice measures.

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The following are some examples:

Caribbean Food and Nutrition Institute. 1986. Nutrition handbook for community workers in the tropics. Jamaica : MacMillan Press.

Lesley, D. 1995. *What makes women sick: gender and the political economy of health.* London : MacMillan Press.

Person, A., Borbasi, S. & Gott, M. 1997. Doctoral education in nursing for practitioner knowledge and for academic knowledge: the university of Adelaide, Australia. *Image: Journal of Nursing Scholarship*, 3(4):365-368, Dec.

Werner, D. 1992. *Where there is no doctor: a village health care handbook for Africa.* Palo Alto, Ca 94302 : MacMillan Press.

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