MANAGEMENT OF BIRTH DEFECTS AND HAEMOGLOBIN DISORDERS

REPORT OF A JOINT
WHO-MARCH OF DIMES MEETING

Geneva, Switzerland, 17-19 May 2006

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EXECUTIVE SUMMARY

On 17-19 May 2006, the World Health Organization (WHO) and the March of Dimes Birth Defects Foundation held a joint meeting in Geneva entitled *The Management of Birth Defects and Haemoglobin Disorders*. The meeting was convened at the request of the WHO's Human Genetics Programme (HGN) following publication of the March of Dimes Global Report on Birth Defects in January 2006 (Christianson et al, 2006).

Meeting participants included 18 experts from developing and industrialized countries and nine staff from WHO Headquarters representing the following programmes: HGN, Newborn and Child Health, Classification and Terminology, Nutrition, Reproductive Health and Research, Disability and Rehabilitation and Making Pregnancy Safer.

The meeting had five goals. These were to (1) ratify the data on the global toll of birth defects presented in the 2006 March of Dimes Global Report on Birth Defects; (2) agree upon a definition of terms; (3) develop a five-year collaborative plan for strengthening care and prevention of birth defects in low- and middle-income countries, bearing in mind the different spectrum of genetic disorders that may occur; (4) develop a five-year plan for the WHO for strengthening care and prevention of haemoglobin disorders (sickle cell disease (SCD) and thalassaemias) in low- and middle-income countries; and (5) determine how potential stakeholders (WHO Regional Offices; international and national governmental agencies; foundations and other nongovernmental organizations; parent/patient and other lay support groups; the private sector; and donor organizations) could contribute to these efforts.

The following summarizes the meeting consensus for each of the five goals listed above.

- Participants endorsed the estimates in the March of Dimes Global Report, and they encouraged efforts to strengthen and expand the report's database. In addition, participants supported the report's recommendations for implementation and strengthening of programmes, including medical genetic services in low- and middle-income countries.

- Participants concluded that the term "birth defect" is synonymous with the term "congenital disorder" as defined and used by the HGN and agreed that both could be used interchangeably. The term "congenital anomalies" used currently in the 10th International Classification of Diseases Participants should be avoided. Participants encouraged the WHO's Classification and Terminology Unit in its development of the next version of the International Classification of Diseases (ICD 11) to actively engage those organizations and individuals necessary to develop an internationally acceptable classification of birth defects. The point of departure could be this committee's acceptance of the definition of the terms birth defect and congenital disorder as synonymous.

- Participants agreed that up to 70 percent of birth defects could be prevented, ameliorated or treated effectively, but that to do so on a global basis would require strengthening programmes, including medical genetic services, in low- and middle-income countries. Such services should build on a continuum involving pre-conception care, maternal health, management of labour, and newborn and child health care for infants and children with acute and chronic disorders. In addition, medical genetic services should have a strong base in primary health care and be integrated with secondary and tertiary health care services. Participants identified three priorities for action to assist in the initiation and development of services for the care and prevention of birth defects: (1) support continued research for the collection and refinement of birth defects data to assist the development of medical genetic services; (2) provide practical advice and support for countries

wishing to develop pre-conception and medical genetic services; and (3) promote human resource capacity development and technology transfer. Services for the care and prevention of birth defects must also link to and build on programmes in nutrition, immunization, infectious disease control and disability and rehabilitation, among others.

- Participants agreed that efforts must be made to improve the situation in developing countries where no services exist for the control of haemoglobin disorders. Activities could be conducted either through continued North/South partnerships or the development of South/South networks based on the Asian Thalassaemia Network model. Given the WHO's current focus on haemoglobin disorders, participants strongly encouraged the WHO to dedicate itself through its various organs to the accomplishment of this endeavour. By activating and organising these networks, the following priorities should be addressed: (1) support of continued research for the collection and refinement of data relevant for the control of haemoglobin disorders; (2) provision of practical advice and support for countries wishing to develop medical services for care and prevention of haemoglobin disorders; and (3) development of human resource capacity and technology transfer through training and education of clinicians, scientists, nurses, and counsellors, and the evolution of parent associations.

- The realization of Goals 3 and 4 in this report will require the combined efforts and political will of the WHO, its hierarchy, the HGN and, importantly, the WHO programmes associated with newborn and child health; reproductive health and research; nutrition; disability and rehabilitation; classification and terminology; and making pregnancy safer, among others. Progress will require that the WHO partner with other organizations dedicated to the care and prevention of birth defects. Also vital to the process is the experience and involvement of national patient/parent support organizations and existing and future North/South and South/South partnerships and networks. The roles and responsibilities of this consortium should include advocacy, technology transfer and capacity building, research, promotion of ethics, and finance.

1. BACKGROUND

The WHO and March of Dimes, in the late 1950s and early 1960s, recognized that health transition in industrialized nations would lead to the necessity of developing medical genetic services (Rose, 2003; Stevenson et al, 1966; WHO, 1964). Subsequently, based on the developing science and technology, individual- and family-directed medical genetic practice rapidly evolved, primarily in academic and tertiary medical centres (Penchaszadeh, 1992; Christianson and Modell 2004).

1.1 Role of the WHO

In 1985, a group of WHO experts anticipated that the health transition in middle- and low-income nations would require the need for medical genetic services in the foreseeable future (WHO, 1985). Based on experiences gained in low- and middle-income countries from the implementation of medical services for the haemoglobin disorders, the expert advisory committee recognized such services would be fundamentally different from those in industrialized nations, because of differences both in culture and in the relative incidences of specific genetic diseases. The model of academic and tertiary care services in industrialized countries would need to be translated into a more holistic community-based strategy, incorporating public health approaches applied through primary health care and closely linked to secondary and tertiary services (WHO, 1989, 1996a; Alwan and Modell, 1997).

The HGN subsequently produced a series of reports supporting this approach providing guidelines for birth defect surveillance (WHO, 1993); the care and prevention of common genetic disorders, including the haemoglobin disorders (WHO, 1994, 1996b,c); and for ethics (WHO, 1998a). Additional reports expanded on the need to initiate and develop services for care and prevention of birth defects in low- and middle-income countries (WHO, 1998b, 1999, 2000; WHO/ICBD/EUROCAT, 2003).

In 2002, the WHO's Advisory Committee on Health Research published a report, entitled *Genomics and World Health*, which stated that it "should be considered in the primacy of fundamental overarching strategies to improve health" including "development of health systems, improved education and classical public health approaches to disease control and prevention and health promotion" (WHO, 2002a). In May 2004, the WHO's World Health Assembly (WHA) endorsed *Genomics and World Health*, adopting it as a relevant WHO resolution (WHO, 2004b).

This year, the Executive Board/117 adopted a resolution on sickle cell anaemia (WHO, 2006a) and made recommendations to the forthcoming WHA to support national and international activities on the control of SCA worldwide. Subsequently, similar discussions on SCD was held at the WHA/59 of this year and on thalassaemia and other haemoglobinopathies at the EB/118 and similar resolutions have been adopted (WHO, 2006b,c).

1.2 Role of the March of Dimes

The March of Dimes Foundation, a non-governmental organization was created in 1938 at the behest of President Franklin D. Roosevelt to combat polio. In 1958, recognizing the implications of health transition, the March of Dimes Foundation redirected its mission, which is currently to improve the health of babies by preventing birth defects, premature birth, and infant mortality. In 1998, the March of Dimes Birth Defects Foundation extended its mission internationally through the creation of its Global Programmes (Rose, 2003). Since 2003, the March of Dimes in its NGO capacity has been in official relations with the WHO.

In 2000, Global Programmes began to document the global toll of birth defects. This effort culminated in the publication in January 2006 of the *March of Dimes Global Report on Birth Defects: the Hidden Toll of Dying and Disabled Children* (Christianson et al, 2006). The March of Dimes report, endorsed by its National Board of Trustees and the American Academy of Pediatrics, features a database of modelled estimates of birth prevalence of common birth defects of genetic or partly genetic origin for 191 countries. The report recognized the principles and practices proposed in previous WHO documents, and it offered phased recommendations for

developing medical genetic services for the care and prevention of birth defects in middle- and low-income nations. The report's recommendations were designed to be implemented within existing systems of primary health care and to link to and strengthen current programmes in women, maternal, newborn and child health; nutrition; disability and rehabilitation; and disease surveillance, among others.

Publication of the March of Dimes Global Report (Christianson et al, 2006) and recent consideration of the WHO Executive Board on the need to control genetic diseases (WHO, 2005a) and on the importance of care and prevention of sickle cell disease (WHO, 2006a) prompted a joint WHO-March of Dimes *Meeting on the Management of Birth Defects and Haemoglobin Disorders*.