World Health Organization

EXECUTIVE BOARD **EB126/10**
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Birth defects

Report by the Secretariat

1. In May 2009 the Executive Board at its 125th session considered an agenda item on birth defects.[1] The Board noted the report on the subject[2] but postponed further discussion to the present session of a draft resolution submitted by China, India and the Republic of Korea. This report is a revised version of the earlier report and reflects comments made by members of the Board. Document EB126/10 Add.1 contains the draft resolution initially considered by the Board and additionally reflects comments and proposals thereon made by members of the Board.

2. This report aims to inform the discussion on birth defects, including definition, epidemiology, burden of disease and interventions for prevention and care, as well as indications of how these interventions might be integrated into existing health services.

DEFINITION

3. The *International statistical classification of diseases and related health problems Tenth Revision* (ICD10), includes birth defects in Chapter XVII "Congenital malformations, deformations and chromosomal abnormalities". Birth defects like inborn errors of metabolism and blood disorders of prenatal origin appear in other chapters. Birth defects can be defined as structural or functional abnormalities, including metabolic disorders, which are present from birth. The term congenital disorder is considered to have the same definition; the two terms are used interchangeably.[3] The eleventh revision of the *International statistical classification of diseases and related health problems* provides an opportunity for a review of the current entry.

4. Irrespective of definition, birth defects can cause spontaneous abortions and stillbirths and are a significant but underrecognized cause of mortality and disability among infants and children under five years of age. They can be life-threatening, result in long-term disability, and negatively impact individuals, families, health-care systems and societies.

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[1] See document EB125/2009/REC/1, summary record of the second meeting, section 1.

[2] Document EB125/7.

[3] *Management of birth defects and haemoglobin disorders: report of a joint WHO–March of Dimes meeting,* Geneva, Switzerland. May, 2006.

BIRTH DEFECTS AND GLOBAL NEWBORN AND CHILD MORTALITY

5. Congenital disorders are a common condition. WHO estimates that some 260 000 deaths worldwide (about 7% of all neonatal deaths) were caused by congenital anomalies in 2004.[1] They are most prominent as a cause of death where overall mortality rates are lower, for example in the WHO European Region, where as many as 25% of neonatal deaths are due to congenital anomalies.

6. Currently, sound estimates do not exist of the number of children who were born with a serious congenital disorder due to genetic or environmental causes. The most common serious congenital disorders are congenital heart defects, neural tube defects and Down syndrome. Haemoglobinophathies (including thalassaemia and sickle cell disease) and glucose-6-phosphate dehydrogenase deficiency, which are not covered by the ICD10 definition of congenital anomalies account for 6% of all congenital disorders.

7. Considerable uncertainties remain as to the incidence and mortality due to congenital disorders, especially in countries which lack adequate registration of deaths. However, these figures indicate that addressing the incidence and mortality associated with congenital anomalies needs to be linked to efforts to achieve the Millennium Development Goal 4 target, of a two thirds reduction in the mortality rate of children under five years of age, between 1990 and 2015.

COMMON CAUSES OF BIRTH DEFECTS

8. Birth defects are a diverse group of disorders of prenatal origin which can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and micronutrient deficiencies. Maternal infections such as syphilis and rubella are a significant cause of birth defects in low- and middle-income countries. Maternal illnesses like diabetes mellitus, conditions such as iodine and folic acid deficiency, and exposure to medicinal and recreational drugs including alcohol and tobacco, certain environmental chemicals, and high doses of radiation are other factors that cause birth defects.

PREVENTION

9. The wide range of causes of birth defects means that a portfolio of prevention approaches is needed. Most birth defects of environmental origin can be prevented by public health approaches, including prevention of sexually-transmitted diseases, legislation controlling sound management of toxic chemicals (e.g. certain agricultural chemicals), vaccination against rubella, and fortification of basic foods with micronutrients (iodine and folic acid). Prevention may be considered in terms of life stage (see Annex).

10. Preconception care aims to ensure the optimal physical and mental well-being of women, and their partners, at the onset of and during early pregnancy, to promote a normal pregnancy and the delivery of a healthy infant. It enables the timely deployment of primary prevention of teratogen-induced birth defects (including those caused by congenital syphilis and rubella), of defects caused by iodine deficiency disorder, of neural tube defects (and possibly other malformations), and of maternal-

[1] *The global burden of disease: 2004 update*. Geneva, World Health Organization, 2008.

age-related chromosomal disorders (e.g. Down syndrome). The timely identification of a family risk of inherited disease, and carrier screening with genetic counselling, enable couples to limit family size where there is a known risk.

11. Prevention during pregnancy requires risk identification and management. Some of the interventions and services related to this can raise ethical, legal and social issues and may have cost implications. Such services include prenatal screening and diagnosis for birth defects, selective termination of pregnancy, and the availability of counselling services. Minimally invasive screening methods currently available involve the measurement of several metabolites in the maternal serum. Abnormal levels of biochemical markers are also associated with fetal structural defects such as Down syndrome, neural tube defects and open ventral wall defects. The detection rate of congenital disorders in the first trimester through biochemical screening is improved when it is done in tandem with ultrasound screening involving nuchal translucency and other ultrasonographical assessments. Ultrasonography in the second trimester is useful to detect major structural defects.

DETECTION, TREATMENT AND CARE

12. Screening of newborn infants for congenital disorders facilitates early detection, treatment and care. Neonatal screening programmes (physical examination of all neonates, screening for congenital hypothyroidism, phenylketonuria, sickle cell disease and glucose-6-phosphate dehydrogenase deficiency) as well as training of primary care providers will support the diagnosis and appropriate referral for treatment of infants with congenital disorders. The physical examination of all newborn infants by trained primary care practitioners is practicable in most health systems and will allow the identification and referral of many birth defects, including cardiovascular defects that are associated with a high risk of early mortality.

13. Treatment of birth defects depends on the level of health care available. It comprises medical therapy, surgery, rehabilitation and palliative care when appropriate.

14. Effective life-saving medical treatment is available for several birth defects, including some common functional single gene defects. Examples include treatment of neonatal jaundice in glucose-6-phosphate dehydrogenase deficiency and with Rhesus incompatibility, congenital hypothyroidism, sickle cell disorders, thalassaemia, haemophilia, cystic fibrosis, and other inborn errors of metabolism. Other treatment options include in utero therapy and postnatal surgical corrections. These are now under research and evaluation in a few selected centres for a number of conditions (e.g. congenital diaphragmatic hernia, congenital heart lesions, myelomeningocele, twin-to-twin transfusion syndrome).

15. Surgery is an important but largely unheralded component of the services required to treat children with birth defects. Over 60% of children with a birth defect have a congenital malformation of a single organ, system or limb. Many birth defects are amenable to cost-effective surgery that can be life saving and improve long-term prognosis. Examples are surgery for simple congenital heart defects, cleft lip and palate, clubfoot, congenital cataracts, and gastrointestinal and urogenital abnormalities.

16. Appropriate treatment is also needed for impairments manifesting themselves after the neonatal period. This includes the early detection and treatment of physical, mental, intellectual or sensory impairments. Access to health and rehabilitation services is important to support the participation and inclusion of affected children.

17. With appropriate training, primary health care practitioners can offer basic care for children with birth defects; recognizing birth defects, diagnosing common problems and identifying associated disabilities. This facilitates basic treatment and counselling at the primary care level, taking into account family and community circumstances and available medical services. Referral to specialist advice is considered when diagnosis is not possible at the primary care level.

IMPLICATIONS FOR SERVICES

18. Services and interventions for the prevention and care of birth defects should be part of existing health-care services, in particular those concerned with maternal and child health. They should combine the best possible patient care with a preventive strategy encompassing education, pre-conception care, population screening, genetic counselling, and the availability of diagnostic services. That strategy must deliver services for the prevention and care of birth defects as part of a continuum of interventions for maternal and child health. Depending on countries' health-care capacity, the services should go beyond primary health care to include obstetric, paediatric, surgical, laboratory, radiological and, if available, clinical genetic services in secondary and tertiary health care.

19. Effective delivery of services for the prevention and care of birth defects depends on the availability of a range of specialist clinical and diagnostic services, and a primary care system that is able to use them. A nucleus of expertise in medical genetics, paediatric surgery, imaging, and fetal medicine is required, with the potential to expand to meet needs. Conventional laboratory services (haematological, microbiological, biochemical) need to be supplemented with cytogenetic and DNA-based diagnostic services. Introduction may need to be a gradual process. Over time, the new technologies will support more efficient and cost-effective service delivery.

20. The diversity of priority conditions, social structures, cultural conventions and health-care capabilities, means that countries need to be able to consider a range of possible services, assessing costs and relative effectiveness, in order to make a selection and decide the sequence of implementation. However, no organized guidance is yet available on this. The WHO Secretariat has an important potential role to play in identifying successful models, and providing coherent information on community genetics that is accessible to public health policy-makers.

POTENTIAL ACTIONS

21. There are several country-level actions that can support the development of services for the prevention and care of birth defects. Prevention requires basic public health approaches to be integrated into health systems including maternal and child health services. Many of the services and interventions proposed are already within the reach of low- and middle-income countries while others can be added as needs and resources determine.

22. Basic components for a national programme for the prevention and care of birth defects include:

(a) Commitment of policy-makers and provision of adequate managerial support.

(b) A core network of appropriate specialist clinical and laboratory services that can be expanded in response to demand.

(c) Integration of approaches for prevention and care of birth defects into primary health care, with emphasis on maternal and child health.

(d) Education and training for health-care providers, particularly those in primary health care.

(e) Organization of health education programmes for the general population and recognized high-risk groups.

(f) Establishment of effective mechanisms to foster development of patient–parent support organizations, and collaboration with them in caring for people with birth defects and their families.

(g) Definition of the ethical, legal, religious and cultural issues relevant to formulating services appropriate for the local population.

(h) Initiation and monitoring of population-screening programmes such as screening of newborn infants, premarital/pre-pregnancy screening, and screening during pregnancy.

(i) Establishment of appropriate surveillance systems for birth defects.[1]

23. There is a need for technical guidance to establish or to strengthen national programmes for the control of birth defects. The following are priority actions for the international community:

(a) Resolve currently divergent opinions on the health burden of both environmental and constitutional birth defects, using the revision of ICD10 to draw on expert review of available data and to consider broadening the groups of conditions beyond those currently included in the classification of congenital anomalies.

(b) Promote legislation and public health activities to minimize exposure of the population, and particularly of pregnant women, to potentially teratogenic infections, chemicals and other environmental risk factors.

(c) Define effective community services, and support the integration of the prevention and care of birth defects into maternal and child health programmes. Support the provision to ministries of health of an organized assessment of requirements and costs and support in choosing priorities.

(d) Identify successful models that can be applied in low- and middle-income countries.

(e) Facilitate and support international networking on birth defect prevention and care programmes, with emphasis on developing common approaches, and optimizing instruments for information, education, cost analysis and surveillance, among others. Promote informatics approaches in view of their potential to support cost–effectiveness.

[1] Support with this may be obtained by collaboration with existing birth defect surveillance systems including the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) which includes ECLAMC (Latin American Collaboration Study of Congenital Malformation), the WHO-supported Craniofacial Anomalies database, and EUROCAT (European Registration of Congenital Anomalies).

ACTION BY THE EXECUTIVE BOARD

24. The Board is invited to note the report, provide further guidance and consider the draft resolution contained in document EB126/10 Add.1, including whether to recommend such a resolution for adoption by the Sixty-third World Health Assembly.

ANNEX

INTERVENTIONS TO PREVENT OR TREAT BIRTH DEFECTS

Preconception care

Family planning

- Introducing women to the concept of reproductive choice
- Reducing total number of children born with a birth defect
- Reducing the proportion of mothers of advanced maternal age, which reduces the birth prevalence of autosomal trisomies, particularly Down syndrome
- Allowing women with affected children the option of not having further children

Preconception screening and counselling

- Using family history taken in primary health care to identify individuals at risk of having affected children
- Carrier screening for common recessive disorders (thalassaemia and sickle cell disorder)

Optimize women's diet before and throughout pregnancy

- Promote use of salt fortified with iodine to prevent iodine deficiency disorder
- Promote use of a staple food fortified with folic acid and use of supplementary multivitamins with folic acid to prevent neural tube defects and other malformations
- Avoid alcohol, tobacco and cocaine
- Ensure adequate general diet (protein, calories, iron)

Prevent and treat teratogen-induced infections before and throughout pregnancy

- Syphilis
- Rubella (67 countries do not have national rubella immunization programmes)

Optimize pre-conception maternal health and treatment for

- insulin-dependent diabetics
- women on treatment for epilepsy
- women on treatment with warfarin

Pregnancy care

Antenatal screening for

- Rhesus status
- Syphilis
- Individuals at risk of having children with birth defects using a family history
- Down syndrome: advanced maternal age; maternal serum screening; early ultrasound scanning
- Neural tube defects with maternal serum screening
- Major malformations with ultrasound fetal anomaly scanning (18+ weeks gestation)
- Carriers of common recessive disorders (thalassaemia and sickle cell disorder)

Prenatal diagnosis

- Ultrasound
- Amniocentesis
- Chorionic villus biopsy

Fetal treatment

- For syphilis
- Intrauterine transfusion for fetal anaemia Rhesus negativity

Newborn infant and child care

Newborn infant examination

- Trained examiner clinically examining all newborn infants for birth defects

Newborn infant screening

- Congenital hypothyroidism
- Phenylketonuria
- Cystic fibrosis
- Others, as dictated by each country's needs and circumstances

Medical treatment

Examples:

- Neonatal jaundice in glucose-6-phosphate deficiency and Rhesus incompatibility
- Treatment and care for children with blood disorders like sickle cell disorder, thalassaemia, etc.
- Treatment of some inborn errors of metabolism
- Care of children with cystic fibrosis

Surgery

Examples: correction of

- simple congenital heart defects
- cleft lip and palate
- clubfoot
- congenital cataracts

Rehabilitation and palliative care

As appropriate

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