

Birth defects

Report by the Secretariat

1. The 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. An earlier version of this report was considered by the Executive Board at its 126th session,¹ following which the Board adopted resolution EB126.R6.

DEFINITION

2. The *International statistical classification of diseases and related health problems, tenth revision* (ICD-10), 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.² The eleventh revision of the classification provides an opportunity for a review of the current entry.

3. 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 affect individuals, families, health-care systems and societies.

BIRTH DEFECTS AND GLOBAL NEWBORN AND CHILD MORTALITY

4. 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 2004.³ They are most prominent as a cause of death in settings where overall mortality rates are lower, for example in the European Region, where as many as 25% of neonatal deaths are due to congenital anomalies.

¹ See document EB126/2010/REC/2, summary record of the seventh meeting.

² *Management of birth defects and haemoglobin disorders: report of a joint WHO–March of Dimes meeting, Geneva, Switzerland, 17–19 May 2006*. Geneva, World Health Organization, 2006.

³ *The global burden of disease: 2004 update*. Geneva, World Health Organization, 2008.

5. There are currently no sound estimates of the number of children born with a serious congenital disorder attributable to genetic or environmental causes. The most common serious congenital disorders are congenital heart defects, neural tube defects and Down syndrome. Haemoglobinopathies (including thalassaemia and sickle-cell disease) and glucose-6-phosphate dehydrogenase deficiency, which are not covered by the ICD-10 definition of congenital anomalies, account for 6% of all congenital disorders.

6. Considerable uncertainties remain as to the incidence of and mortality attributable to congenital disorders, especially in countries that lack adequate registration of deaths. However, existing figures indicate that work on reducing the incidence of 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

7. 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 infectious diseases 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 medicines and recreational drugs including alcohol and tobacco, certain environmental chemicals, and high doses of radiation are other factors that cause birth defects.

PREVENTION

8. 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 infections, legislation controlling 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).

9. 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 increase the likelihood of a normal pregnancy and the delivery of a healthy infant. It enables the timely deployment of primary prevention interventions which aim to prevent teratogen-induced birth defects (including those caused by congenital syphilis and rubella), defects caused by iodine deficiency disorder, neural tube defects (and possibly other malformations), and maternal-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.

10. 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 are currently available, such as taking maternal blood for 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 undertaken 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

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

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

13. 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 in Rhesus incompatibility, and therapy for 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).

14. Surgery is an important but largely unheralded component of the services required to treat children with birth defects. More than 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, club foot, congenital cataracts, and gastrointestinal and urogenital abnormalities.

15. 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.

16. With appropriate training, primary health-care practitioners can offer basic care for children with birth defects. They are able to recognize birth defects, diagnose common problems and identify associated disabilities, which in turn enables them to offer basic treatment and counselling, 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 health-care level.

IMPLICATIONS FOR SERVICES

17. 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 capacities, these 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.

18. 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 health-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, and 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.

19. 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

20. 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.

21. Basic components of 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 to the prevention and care of birth defects into primary health care, with an 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

22. The establishment or strengthening of national programmes for the control of birth defects needs technical guidance, and in this regard there are several 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 ICD-10 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 health ministries of an organized assessment of requirements and costs, and support them 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 an 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 in establishing surveillance systems may be obtained by collaborating with existing birth defect surveillance systems, including the International Clearinghouse for Birth Defects Surveillance and Research, which includes the Latin American Collaborative Study of Congenital Malformations, the WHO-supported International Database of Craniofacial Anomalies, and the European Registration of Congenital Anomalies.

ACTION BY THE HEALTH ASSEMBLY

23. The Health Assembly is invited to adopt the resolution recommended by the Executive Board in resolution EB126.R6.

ANNEX

INTERVENTIONS TO PREVENT OR TREAT BIRTH DEFECTS

Preconception care	Pregnancy care	Newborn infant and child care
<p>Family planning</p> <ul style="list-style-type: none"> • Introducing women to the concept of reproductive choice • Reducing the 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 <p>Preconception screening and counselling</p> <ul style="list-style-type: none"> • Using family history taken in primary health care facilities to identify individuals at risk of having affected children • Undertaking carrier screening for common recessive disorders (e.g. thalassaemia and sickle cell disorders) <p>Optimizing women's diet before and throughout pregnancy</p> <ul style="list-style-type: none"> • Promoting the use of salt fortified with iodine to prevent iodine deficiency disorder • Promoting the use of a staple food fortified with folic acid and of supplementary multivitamins with folic acid to prevent neural tube defects and other malformations • Promoting avoidance of alcohol, tobacco and cocaine • Promoting an adequate general diet (e.g. with sufficient protein, calories, iron) • Ensure adequate general diet (protein, calories, iron) <p>Preventing and treating teratogen-induced infections before and throughout pregnancy</p> <p>Such infections include:</p> <ul style="list-style-type: none"> • syphilis • rubella (67 countries do not have national rubella immunization programmes) <p>Optimizing preconception maternal health and treatment for</p> <ul style="list-style-type: none"> • Insulin-dependent diabetics • Women on treatment for epilepsy • Women on treatment with warfarin 	<p>Antenatal screening for:</p> <ul style="list-style-type: none"> • 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 (maternal serum screening) • Major malformations (ultrasound fetal anomaly scanning (18+ weeks gestation)) • Carriers of common recessive disorders (e.g. thalassaemia and sickle cell disorders) <p>Prenatal diagnosis</p> <ul style="list-style-type: none"> • Ultrasound • Amniocentesis • Chorionic villus biopsy <p>Fetal treatment for</p> <ul style="list-style-type: none"> • Syphilis • Fetal anaemia with intrauterine transfusion 	<p>Newborn infant examination</p> <ul style="list-style-type: none"> • Trained examiner clinically examining all newborn infants for birth defects <p>Newborn infant screening</p> <ul style="list-style-type: none"> • Congenital hypothyroidism • Phenylketonuria • Cystic fibrosis • Others, as dictated by each country's needs and circumstances <p>Medical treatment</p> <p>Examples:</p> <ul style="list-style-type: none"> • Neonatal jaundice in glucose-6-phosphate dehydrogenase deficiency and Rhesus incompatibility • Treatment and care for children with blood disorders such as sickle-cell disorder, thalassaemia, etc. • Treatment of some inborn errors of metabolism • Care of children with cystic fibrosis <p>Surgery</p> <p>Examples include the correction of:</p> <ul style="list-style-type: none"> • Simple congenital heart defects • Cleft lip and palate • Club foot • Congenital cataracts <p>Rehabilitation and palliative care</p> <p>As appropriate</p>

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