Key facts

- An estimated 276,000 babies die within 4 weeks of birth every year, worldwide, from birth defects.
- Birth defects can result in long-term disability, which may have significant impacts on individuals, families, health-care systems and societies.
- The most common severe birth defects are heart defects, neural tube defects and Down syndrome.
- Although birth defects may be genetic, infectious, nutritional or environmental in origin, most often it is difficult to identify the exact causes.
- Some of them can be prevented. For example, vaccination, adequate intake of folic acid or iodine through fortification of staple foods or provision of supplements, and adequate antenatal care are keys for prevention.

WHO Resolution

Birth defects are important causes of childhood death, chronic illness and disability in many countries. In 2010, the World Health Assembly adopted a resolution on birth defects calling all Member States to promote primary prevention and improve the health of children with congenital anomalies by:

- Developing and strengthening registration and surveillance systems;
- Developing expertise and building capacity;
- Strengthening research and studies on etiology, diagnosis and prevention;
- Promoting international cooperation.

Birth Defects and Zika virus

On the 1st of February 2016 the World Health Organization (WHO) declared the strongly suspected causal link between Zika virus infection in pregnant women and microcephaly in new-borns a Public Health Emergency of International Concern. The link is not yet conclusive, and it will take time and effort to understand the nature and biological basis of the association. Potential adverse effects of exposure to Zika virus in utero may not be restricted to microcephaly alone. Surveillance approaches should also identify changes in rates of spontaneous abortion, stillbirths, low birth weight and small for gestational age (SGA). Postpartum prevalence of visual, hearing or other neurological and developmental abnormalities should also be looked for and monitored.

Definition

Birth defects are also known as congenital anomalies, congenital disorders or congenital malformations. Congenital anomalies can be defined as structural or functional anomalies (e.g. metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth or later in life.
Causes and risk factors
Although approximately 50% of all congenital anomalies cannot be linked to a specific cause, there are some known causes or risk factors. There are many potential causes of microcephaly, but they often remain unknown. The most common causes include:
- infections in the womb: toxoplasmosis (caused by a parasite found in undercooked meat), rubella, herpes, syphilis, cytomegalovirus and HIV;
- exposure to toxic chemicals: maternal exposure to heavy metals like arsenic and mercury, alcohol, radiation, and smoking;
- genetic abnormalities such as Down syndrome; and
- severe malnutrition during fetal life, amongst others

Detection
Health care before (preconception) and around the time of conception (peri-conception) includes basic reproductive health practices, as well as medical genetic screening and counselling. Screening can be conducted during the 3 periods listed next.
- Preconception screening can be useful to identify people at risk for specific disorders or at risk for passing a disorder onto their children. Screening includes obtaining family histories and carrier screening, and is particularly valuable in countries where consanguineous marriage is common.
- Peri-conception screening: maternal characteristics may increase risk, and screening results should be used to offer appropriate care, according to risk. This may include screening for young or advanced maternal age, as well as screening for use of alcohol, tobacco or other psychoactive drugs. Ultrasound can be used to screen for Down syndrome during the first trimester and for severe fetal anomalies during the second trimester. Additional tests and amniocentesis may help in the detection of neural tube defects and chromosomal abnormalities during the first and second trimesters.
- Neonatal screening includes clinical examination and screening for disorders of the blood, metabolism and hormone production. Screening for deafness and heart defects, as well as early detection of congenital anomalies, can facilitate life-saving treatments and prevent progression towards some physical, intellectual, visual or auditory disabilities. In some countries, babies are routinely screened for abnormalities of the thyroid or adrenal glands before discharge from the maternity unit.

Treatment and care
The treatment of birth defects depends on the nature and severity of the condition. Treatment options vary from case to case.
Many structural congenital anomalies can be corrected with paediatric surgery and early treatment can be administered to children with functional problems such as thalassemia (inherited recessive blood disorders), sickle cell disorders and congenital hypothyroidism (reduced function of the thyroid).
Prevention

Preventive public health measures delivered through health services decrease the frequency of certain congenital anomalies. Primary prevention of congenital anomalies includes:

- improving the diet of women throughout their reproductive years, ensuring an adequate dietary intake of vitamins and minerals, and particularly folic acid, through daily oral supplements or fortification of staple foods such as wheat or maize flours;
- ensuring mothers abstain from, or restrict, their intake of harmful substances, particularly alcohol;
- controlling preconception and gestational diabetes, through counselling, weight management, diet and administration of insulin when needed;
- avoiding environmental exposure to hazardous substances (e.g. heavy metals, pesticides) during pregnancy;
- ensuring that any exposure of pregnant women to medications or medical radiation (e.g. imaging rays) is justified, based on careful health risk–benefit analysis;
- improving vaccination coverage, especially against the rubella virus, for children and women. Rubella can be prevented through childhood vaccination. The rubella vaccine can also be given at least 1 month prior to pregnancy to women who have not been vaccinated and do not have a history of rubella in childhood;
- increasing and strengthening education of health staff and others involved in promoting prevention of congenital anomalies.

SEAR-NBBDD Surveillance network

The WHO SEARO New born and birth defect (NBBD) surveillance network is an initiative in partnership with US-CDC, operational since July, 2014 to ascertain the prevalence of birth defects and study new-born health and stillbirths in the South East Asia region.

The birth defects component of the online surveillance system is currently capturing externally visible birth defects, from selected participating hospitals in 8 SEAR countries. We are working to upgrade the system in response to the recent Zika virus Public Health Emergency to monitor occurrence of microcephaly and detect any unusual increase and clustering of cases in the region.

WHO Global response

In 2010, the World Health Assembly considered a report and adopted a resolution on birth defects. The report describes the basic components for creating a national programme for the surveillance, prevention and care of congenital anomalies before and after birth. It also recommends priorities for the international community to assist in establishing and strengthening these national programmes.

- World Health Assembly report on birth defects
- World Health Assembly resolution WHA63.17 on birth defects
**Miscellaneous Efforts**

The Global Strategy for Women’s and Children’s Health, launched in 2010 by the United Nations, in collaboration with leaders from governments and other organizations like WHO and UNICEF, has been crucial in implementing high-impact and cost-effective interventions to improve neonatal and child health.

- [Global Strategy for Women’s and Children’s Health](#)

The WHO Departments of Reproductive Health and Research and Nutrition for Health and Development, in collaboration with the International Clearinghouse for Birth Defects Surveillance and Research and CDC’s National Center on Birth Defects and Developmental Disabilities, convene annual training programmes on the surveillance and prevention of congenital anomalies and preterm births. The WHO Department of HIV and AIDS collaborates with these partners, to strengthen the surveillance of congenital anomalies for women receiving antiretroviral drugs during pregnancy, as an integral part of the monitoring and evaluation of national HIV programmes.

Gavi, the Vaccine Alliance, of which WHO is a partner, is assisting low- and middle-income countries in improving control and elimination of rubella and congenital rubella syndrome through immunization.

WHO develops normative tools, including guidelines and a global plan of action, to strengthen medical care and rehabilitation services to support the implementation of the United Nations Convention on the Rights of Persons with Disabilities. Similarly, WHO supports countries to integrate medical care and rehabilitation services into overall primary health care, supports the development of community-based rehabilitation programmes, and facilitates the strengthening of specialized rehabilitation centres and their links with community-based rehabilitation.

- [United Nations Convention on the Rights of Persons with Disabilities](#)

The WHO Department of Public Health and Environment focuses on a number of activities, and defines interventions, to address the environmental and social determinants of child development. These include children’s unique vulnerabilities to polluted indoor and outdoor air, contaminated water, lack of sanitation, toxicants, heavy metals, waste components and radiation; combined exposures with social, occupational and nutrition factors; and the settings in which children dwell (home, school). WHO is also working with the United States Centres for Disease Control and Prevention’s (CDC) National Centre on Birth Defects and Developmental Disabilities and other partners, to establish a global policy for folic acid fortification at the country level. WHO is also working with partners to provide the required technical expertise for the surveillance of neural tube defects, for monitoring fortification of staple foods with folic acid, and for improving laboratory capacity for assessing risks for folic acid-preventable congenital anomalies.

**For more information on birth defects**

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