

Congenital disorder

Congenital disorder is any medical condition that is present at birth. However, a congenital disorder can be recognized before birth (parentally), at birth, years later, or never. The term congenital does not imply or exclude a genetic cause. Congenital disorders can be a result of genetic abnormalities, the intrauterine environment, a mixture of both, errors of morphogenesis, or unknown factors.

Congenital conditions can be referred to as diseases, defects, disorders, anomalies, or simply genetic differences or uncomfortable. The usage overlaps, but also involves a value judgment as to the harmfulness of the condition. In particular, people may disagree as to whether a specific physical anomaly should be considered a birth defect or a normal variation. See human variability and disease for more on the occasional difficulties of drawing these distinctions.

A congenital disorder can have trivial or grave consequences. The most severe, such as anencephaly, are incompatible with life. Others, such as congenital tumors, vary from causing stillbirth to requiring fetal intervention or special delivery procedures such as the EXIT procedure, to needing surgery in the neonatal period. The most common congenital tumor is teratoma. Congenital physical anomalies (birth defects) are a leading cause of death in early infancy

Congenital disorders" is a broad category that includes a variety of conditions. Congenital disorders include minor physical anomalies (e.g., a birthmark), severe malformations of single systems (e.g., congenital heart disease or dysmelia), and combinations of abnormalities affecting several parts of the body. Congenital defects of metabolism are also considered congenital disorders.

Types of congenital disorder

A **congenital physical** anomaly is an abnormality of the structure of a body part. An anomaly may or may not be perceived as a problem condition. Many, if not most, people have one or more minor physical anomalies if examined carefully. Examples of minor anomalies can include curvature of the 5th finger (clinodactyly), tiny indentations of the skin near the ears (preauricular pits), shortness of the 4th metacarpal or metatarsal bones, or dimples over the lower spine 7 Some minor anomalies may be clues to more significant internal abnormalities.

A **congenital malformation** is a congenital physical anomaly that is deleterious, i.e. a structural defect perceived as a problem. A typical combination of malformations affecting more than one body part is referred to as a malformation syndrome.

Birth defect is a widely-used term for a congenital malformation, i.e. a congenital, physical anomaly which is recognizable at birth, and which is significant enough to be considered a problem.

Genetic disorder or diseases are all congenital, though they may not be expressed or recognized until later in life. Genetic diseases may be divided into single-gene defects, multiple-gene disorders, or chromosomal defects. Single-gene defects may arise from abnormalities of both copies of an autosomal gene (a recessive disorder) or of only one of the two copies (a dominant disorder). Some conditions result from deletions or abnormalities of a few genes located contiguously on a chromosome. Chromosomal disorders involve the loss or duplication of larger portions of a chromosome (or an entire chromosome) containing hundreds of genes. Large chromosomal abnormalities always produce effects on many different body parts and organ systems.

A **congenital metabolic** disease is also referred to as an inborn error of metabolism. Most of these are single gene defects, usually heritable. Many affect the structure of body parts but some simply affect the function.

Congenital physical anomalies (birth defects)

Types

A limb malformation is called a dysmelia. These include all forms of malformations of limbs, such as amelia, ectrodactyly, phocomelia, polymelia, polydactyly, syndactyly, polysyndactyly, oligodactyly, brachydactyly, achondroplasia, congenital aplasia or hypoplasia, amniotic band syndrome, and cleidocranial dysostosis.

Congenital malformations of the heart include patent ductus arteriosus, atrial septal defect, ventricular septal defect, and tetralogy of fallot. Helen Taussig has been a major force in research on congenital malformations of the heart.

Congenital malformations of the nervous system include neural tube defects such as spina bifida, meningocele, meningomyelocele, encephalocele and anencephaly. Other congenital malformations of

the nervous system include the Arnold-Chiari malformation, the Dandy-Walker malformation, hydrocephalus, microencephaly, megencephaly, lissencephaly, polymicrogyria, holoprosencephaly, and agenesis of the corpus callosum.

Occurrence rate

About 3% of newborns have a "major physical anomaly", meaning a physical anomaly that has cosmetic or functional significance.

Congenital malformations involving the brain are the largest group at 10 per 1000 live births, compared to heart at 8 per 1000, kidneys at 4 per 1000, and limbs at 1 per 1000. All other physical anomalies have a combined incidence of 6 per 1000 live births.

Congenital malformations of the heart have the highest risk of death in infancy, accounting for 28% of infant deaths due to birth defects, while chromosomal abnormalities and respiratory abnormalities each account for 15%, and brain malformations about 12%.

Causes

The cause of 40-60% of congenital physical anomalies (birth defects) in humans is unknown. These are referred to as sporadic birth defects, a term that implies an unknown cause, random occurrence, and a low recurrence risk for future children. For 20-25% of anomalies there seems to be a "multifactorial" cause, meaning a complex interaction of multiple minor genetic abnormalities with environmental risk factors. Another 10-13% of anomalies have a purely environmental cause (e.g. infections, illness, or drug abuse in the mother). Only 12-25% of anomalies have a purely genetic cause. Of these, the majority are chromosomal abnormalities.

Genetic causes of congenital anomalies include inheritance of abnormal genes from the parents, as well as new mutations in one of the germ cells that gave rise to the fetus.

Environmental causes of congenital anomalies are referred to as teratogenic. These are generally problems with the mother. Teratogens can include dietary deficiencies, toxins, or infections. For example, dietary deficiency of maternal folic acid is associated with spina bifida. Ingestion of harmful substances by the mother (e.g. alcohol, mercury, or prescription drugs such as phenytoin) can cause recognizable combinations of birth defects.

Several infections which a mother can contract during pregnancy can also be teratogenic. These are referred to as the TORCH infections.

Teratogens

The greatest risk of a malformation due to environmental exposure to a teratogen (terato = monster, gen = producing) is between the third and eighth week of gestation. Before this time, any damage to the embryo is likely to result in fatality, and the baby will not be born. After eight weeks, the fetus and its organs are more developed and less sensitive to teratogenic incidents.

The type of birth defect is also related to the time of exposure to a teratogen. For instance, the heart is susceptible from three to eight weeks, but the ear is susceptible from a slightly later time to about twelve weeks.

Many common skin care ingredients can be absorbed through the skin in small amounts, where they can enter the bloodstream and pass into baby's circulation.

Infection in the mother early in the third week may damage the heart. An infection in the eleventh week is less likely to damage the heart, but the baby may be born deaf.

PNAC Resource Centre

Noman Farooq

Senior program Associate

PAKISTAN NATIONAL **AIDS** CONSORTIUM

House No 441, Street 57, Sector I 8/3 Islamabad

Email: noman@pnac.net.pk

www.pnac.net.pk

www.aidspnac.org