

## Glossary of Birth Defects and Related Terms

**Achondroplasia:** A genetic dysplasia of cartilage and long bones caused by mutations in the gene FGFR3. It results in disproportionate short stature with short limbs and relatively more normal trunk size. Persons affected with achondroplasia can have abnormalities of the foramen magnum potentially causing damage to the upper spinal cord. Spinal canal stenosis is a problem starting in late adolescence. There may also be lumbar lordosis, limited elbow extension and early arthritis. People with achondroplasia typically have normal intelligence.

**Agenesis** Absence of part(s) of the body.

**Agenesis, aplasia, or hypoplasia of the lung** The absence or incomplete development of a lung or lung tissue.

**Anencephaly** Congenital absence of the skull, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Anencephaly is not compatible with life.

**Aniridia** The complete absence of the iris of the eye or a defect of the iris. Can be congenital or traumatically induced.

**Anomalies of the tricuspid valve** Includes tricuspid valve atresia or stenosis, as well as enlargement, dilation, or aneurysm of the tricuspid valve. See also tricuspid valve atresia or stenosis.

**Anophthalmia** A developmental defect characterized by complete absence of the eyes, or by the presence of vestigial eyes.

**Anotia** A congenital absence of one or both ears.

**Aorta** The large arterial trunk that carries blood from the heart to be distributed by branch arteries through the body

**Aortic valve stenosis** A cardiac anomaly characterized by a narrowing or stricture of the aortic valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can be repaired surgically in some cases.

**Atresia** Imperforation; absence or closure of a normal opening.

**Atrial septal defect** A congenital cardiac malformation in which there are one or several openings in the atrial septum (muscular and fibrous wall between the right and left atria) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or may require surgical treatment. Also called *ostium secundum defect*.

**Atrium** One of the two upper chambers of the heart (plural atria). The right atrium receives unoxygenated blood from the body. The left atrium receives oxygenated blood from the lungs.

**Biliary atresia** A congenital absence or underdevelopment of one or more of the ducts in the biliary tract. Correctable surgically.

### Birth prevalence

$$\frac{\text{\# of cases with birth defect}}{\text{A in an area and time period}} \times 10,000$$

# of live births in that area  
and time period

**Bladder exstrophy** Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The upper urinary tract is generally normal. Often associated with anorectal and genital malformations, and epispadias. Affected persons are at a markedly increased risk of bladder carcinoma (squamous cell). This condition is usually corrected surgically after birth.

**Cataract** An opacity (clouding) of the lens of the eye.

**Choanal atresia or stenosis** A congenital anomaly in which a bony or membranous formation blocks the passageway between the nose and the pharynx. This defect is usually repaired surgically after birth. Bilateral choanal atresia is a surgical emergency.

**Cleft lip** The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip. Infants with this condition can have difficulty feeding, and may use assistive devices for feeding. This condition is corrected when the infant can tolerate surgery.

**Cleft palate** The congenital failure of the palate to fuse properly, forming a grooved depression or fissure in the roof of the mouth. This defect varies in degree of severity. The fissure can extend into the hard and soft palate and into the nasal cavities. Infants with this condition have difficulty feeding, and may use assistive devices for feeding. Surgical correction is begun as soon as possible. Children with cleft palates are at high risk for hearing problems due to ear infections.

**Clubfoot:** See Talipes Equinovarus.

**Cluster** An apparently unusual concentration of a health condition in a particular area and time period.

**Coarctation of the aorta** Localized narrowing of the aorta. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Surgical correction is recommended even for mild defects.

**Common truncus arteriosus** A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta. This is corrected surgically.

**Confidence interval (95%)** The interval that contains the true prevalence (which we can only estimate) 95% of the time. See Methods for more explanation.

**Congenital** Existing at or dating from birth.

**Congenital hip dislocation** A congenital defect in which the head of the femur does not articulate with the acetabulum of the pelvis because of an abnormal shallowness of the acetabulum. Treatment in early infancy consists of bracing of the joint to cause a deepening of the acetabulum.

**Craniosynostosis** A premature ossification (closing) of the cranial sutures before birth or soon after birth. This condition is occasionally associated with other skeletal defects. If no surgical correction is made, the growth of the skull is inhibited, and the head is deformed. The eyes and the brain are often damaged.

**Diaphragmatic hernia** A failure of the diaphragm to form completely, leaving a hole. Abdominal organs can protrude through the hole into the chest cavity and interfere with development of the heart and lungs. Usually life-threatening and requires emergent surgery.

**Down syndrome (Trisomy 21)** The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. The extra copy can be free-lying, or can be attached to some other chromosome, most frequently number 14. Down syndrome can occur in mosaic, so that there is a population of normal cells and a population of trisomy 21 cells. Down syndrome is characterized by moderate to severe mental retardation, sloping forehead, small ear canals, flat bridged nose, and short fingers and toes. One third of infants have congenital heart disease, and one third have duodenal atresia. (Both can be present in the same infant.) Affected people can survive to middle or old age. There is an increased incidence of Alzheimer disease in adults with Down syndrome.

**Dysgenesis** Impaired or faulty development of part(s) of the body.

**Ebstein anomaly** A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle causing abnormal patterns of cardiac circulation.

**Edwards syndrome (Trisomy 18)** The chromosomal abnormality characterized by an extra copy of chromosome 18. The extra chromosome can be free lying or attached to another chromosome. Trisomy 18 can occur in mosaic. Edwards syndrome is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation, and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare.

**Embryogenesis** The development and growth of an embryo, especially the period from the second through the eighth week after conception.

**Encephalocele** The protrusion of the brain substance through a defect in the skull.

**Endocardial cushion defect** A variety of septal defects (malformations of the walls separating the two atria and two ventricles of the heart) resulting from imperfect fusion of the endocardial cushions in the embryonic heart.

**Epispadias** A congenital defect in which the urinary meatus (urinary outlet) opens above (dorsal to) the normal position. The urinary sphincters are defective, so incontinence does occur. Surgical correction is aimed at correcting incontinence and permitting sexual functioning. The corresponding defect in females is rare. *See also Hypospadias.*

**Esophageal stenosis or atresia** A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a tracheoesophageal fistula.

**Fetal alcohol syndrome** A constellation of physical abnormalities (including characteristic abnormal facial features and growth retardation), and problems of behavior and cognition in children born to mothers who drank alcohol during pregnancy.

**Fistula** An abnormal passage from an internal organ to the body surface or between two internal organs or structures.

**Folate** B vitamin necessary for red blood cell production; folate deficiency can lead to anemia and, during embryogenesis, can affect the normal development of the fetus' neural tube; found in liver, green leafy vegetables, beans, beets, broccoli, cauliflower, citrus fruits, and sweet potatoes. *See folic acid.*

**Folic acid** One of the B vitamins especially important for a woman to take before conception to help prevent neural tube defects in a fetus; essential for DNA synthesis and therefore the growth and division of cells; obtained from fortified foods or from a multivitamin containing at least 4mg; also found in natural sources including liver, beans, and leafy green vegetables. While folate and folic acid are both forms of water-soluble B vitamins, folic acid refers to the synthetic vitamin used in supplements, whereas folate is the form found in foods.

**Gastroschisis** A congenital opening of the abdominal wall with protrusion of the intestines. This condition is surgically treated. Contrast with Omphalocele, below.

**Hernia** A protrusion of an organ or part through connective tissue or through a wall of the cavity in which it is normally enclosed.

**Hirschsprung disease** The congenital absence of autonomic ganglia (nerves controlling involuntary and reflexive movement) in the muscles of the colon. This results in immobility of the intestines and may cause obstruction or stretching of the intestines. This condition is repaired surgically in early childhood by the removal of the affected portion of the intestine.

**Holoprosencephaly** Failure of the brain to develop into two equal halves, so there is structural abnormality of the brain. There may be associated midline facial defects including cyclopia (fusion of the eye orbits into a single cavity containing one eye) in severe cases. About half the cases are probably due to a single gene defect (the HPE gene). Frequently occurs with Trisomy 13.

**Hydrocephaly** The abnormal accumulation of fluid within the spaces of the brain.

**Hyperplasia** Overgrowth characterize by an increase in the number of cells of a tissue.

**Hypoplasia** A condition of arrested development in which an organ or part remains below the normal size or in an immature state.

**Hypoplastic left heart syndrome** Atresia, or marked hypoplasia, of the aortic opening or valve, with hypoplasia of the ascending aorta and defective development of the left ventricle (with mitral valve atresia). This condition can be surgically repaired in a series of three procedures over a period of one year. Transplantation is also a treatment. This condition is usually fatal in the first month of life if not treated.

**Hypospadias** A congenital defect in which the urinary meatus (urinary outlet) is on the underside of the penis or on the perineum (area between the genitals and the anus). The urinary sphincters are not defective so incontinence does not occur. The condition may be surgically corrected if needed for cosmetic, urologic, or reproductive reasons. The corresponding defect in women is rare. *See also epispadias*

**Limb defects** *See* Reduction defects.

**Meninges** Membranes that cover the brain and spinal cord.

**Microcephaly** The congenital smallness of the head, with corresponding smallness of the brain.

**Microphthalmia** The congenital abnormal smallness of one or both eyes. Can occur in the presence of other ocular defects.

**Microtia** A small or maldeveloped external ear and atretic or stenotic external auditory canal.

**Mosaic** In genetics, this refers to an individual organism that has two or more kinds of genetically different cell types. The degree of abnormality depends on the type of tissue containing affected cells. Individuals may vary from near normal to full manifestation of the genetic syndrome. Can occur in any chromosome abnormality syndrome.

**Neural tube defect** A defect resulting from failure of the neural tube to close in the first month of pregnancy. The major conditions include anencephaly, spina bifida, and encephalocele.

**Obstructive genitourinary defect** Stenosis or atresia of the urinary tract at any level. Severity of the defect depends largely upon the level of the obstruction. Urine accumulates behind the obstruction and damages the organs.

**Omphalocele** The protrusion of an organ into the umbilicus. The defect is usually closed surgically soon after birth. Contrast with Gastroschisis.

**Ostium secundum defect** *See atrial septal defect.*

**Patau syndrome (Trisomy 13)** The chromosomal abnormality caused by an extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Patau syndrome can occur in mosaic so that there is a population of normal cells and a population of trisomy 13 cells. Patau syndrome is characterized by impaired midline facial development, cleft lip and palate, polydactyly, and mental retardation. Most infants do not survive beyond 6 months of life.

**Patent ductus arteriosus** A blood vessel between the pulmonary artery and the aorta. This is normal in fetal life, but can cause problems after birth, particularly in premature infants. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. The vast majority close spontaneously and cause no problems. Medical or surgical correction may be done. This is only an abnormality if it causes significant medical problems.

**Poisson regression** a type of statistical analysis based on the Poisson distribution used to compare rates of rare occurrences such as birth defects between different population groups, different areas, or different times.

**Prevalence** With respect to the prevalence of birth defects, *see "Birth prevalence"*.

**Pulmonary artery anomaly** Abnormality in the formation of the pulmonary artery such as stenosis or atresia. *See also common truncus.*

**Pulmonary valve atresia or stenosis** A congenital heart condition characterized by absence or constriction of the pulmonary valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Mild forms are relatively well tolerated and require no intervention. More severe forms are surgically corrected.

**Pyloric stenosis** A narrowing of the pyloric sphincter at the outlet of the stomach. This causes a blockage of food from the stomach into the small intestine. Usually treated surgically.

**Reduction defects of the lower limbs** The congenital absence of a portion of the lower limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing tibia and great toe).

**Reduction defects of the upper limbs** The congenital absence of a portion of the upper limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing radius and thumb).

**Renal agenesis or dysgenesis** The failure, or deviation, of embryonic development of the kidney.

**Spina bifida** A neural tube defect resulting from failure of the spinal neural tube to close. The spinal cord and/or meninges may or may not protrude. This usually results in damage to the spinal cord with paralysis of the involved limbs. Includes myelomeningocele (involving both spinal cord and meninges) and meningocele (involving just the meninges).

**Stenosis** A narrowing or constriction of the diameter of a bodily passage or orifice.

**Stenosis or atresia of large intestine, rectum and anus** The absence, closure or constriction of the large intestine, rectum or anus. Can be surgically corrected or bypassed.

**Stenosis or atresia of the small intestine** A narrowing or incomplete formation of the small intestine obstructing movement of food through the digestive tract.

**Talipes equinovarus (Clubfoot):** A development disorder of the foot and ankle that affects one (unilateral) or both (bilateral) feet. The foot is in an incorrect anatomical position, and is inclined inward, axially rotated outward, and points downward. Clubfoot is a complex disorder that is caused by genetic and environmental influences.

**Tetralogy of Fallot** A congenital cardiac anomaly consisting of four defects: ventricular septal defect, pulmonary valve stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle. The condition is corrected surgically.

**Tracheoesophageal fistula** An abnormal passage between the esophagus and trachea. Leads to pneumonia. Corrected surgically. It is frequently associated with esophageal atresia.

**Translocation** The rearrangement of genetic material within the same chromosome or the transfer of a segment of one chromosome to another one. People with balanced translocations do not always manifest genetic syndromes, but may be carriers of genetic syndromes and can have children with unbalanced translocations. Can occur with any chromosomal anomaly syndrome.

**Transposition of the great vessels** A congenital malformation in which the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (opposite of normal), so that the venous return from the peripheral circulation is recirculated without being oxygenated in the lungs. Immediate surgical correction is needed. When this is not associated with other cardiac defects, and not corrected, it is fatal.

**Tricuspid valve atresia or stenosis** A congenital cardiac condition characterized by the absence or constriction of the tricuspid valve. The opening between the right atrium and right ventricle is absent or restricted, and normal circulation is not possible. This condition is often associated with other cardiac defects. This condition is surgically corrected depending on the severity.

**Trisomy** A chromosomal abnormality characterized by one more than the normal number of chromosomes. Normally, cells contain two of each chromosome. In trisomy, cells contain three copies of a specific chromosome.

**Trisomy 13 (Patau syndrome)** The chromosomal abnormality caused by an extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Trisomy 13 can occur in mosaic so that there is a population of normal cells and a population of trisomy 13 cells. Trisomy 13 is characterized by impaired midline facial development, cleft lip and palate, polydactyly, and mental retardation. Most infants do not survive beyond 6 months of life.

**Trisomy 18 (Edwards Syndrome)** The chromosomal abnormality characterized by an extra copy of chromosome 18. The extra chromosome can be free lying or attached to another chromosome. Trisomy 18 can occur in mosaic so that there is a population of normal cells and a population of trisomy 18 cells. Trisomy 18 is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation, and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare.

**Trisomy 21 (Down Syndrome)** The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. The extra copy can be free-lying, or can be attached to some other chromosome, most frequently number 14. Trisomy 21 can occur in mosaic, so that there is a population of normal cells and a population of trisomy 21 cells. Trisomy 21 is characterized by moderate to severe mental retardation, sloping forehead, small ear canals, flat bridged nose, and short fingers and toes. One third of infants have congenital heart disease, and one third have duodenal atresia. (Both can be present in the same infant.) Affected people can survive to middle or old age. There is an increased incidence of Alzheimer disease in adults with Trisomy 21.

**Truncus arteriosus** See *Common truncus*.

**Ventricle** One of the two lower chambers of the heart (plural ventricles). The right ventricle sends blood to the lungs, and the left ventricle passes oxygen-rich blood to the rest of the body.

**Ventricular septal defect (VSD)** A congenital cardiac malformation in which there are one or several openings in the ventricular septum (muscular and fibrous wall between the right and left ventricle or right and left lower chambers of the heart) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or require surgical treatment.