

Glossary of Birth Defects for Congenital Malformation Rates: Beginning 1992

(Courtesy of the Texas Birth Defects Monitoring Division)

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.

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.

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.

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.

Esophageal stenosis or atresia with/without tracheoesophageal fistula A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a tracheoesophageal fistula.

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

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

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.

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.

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

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).

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 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.

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.

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.

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*.