

Medical Glossary



FACIAL



Cleft Lip AND Palate – (Children with cleft lip only are NOT typically available, may be unilateral or bilateral, first to fourth degree) Failure of the upper lip and roof of the mouth to close during fetal development. The cleft in the lip can be unilateral (one-sided) or bilateral (two-sided) and can range from a small notch to a complete separation extending into the nose. A cleft palate permits communication between nose and mouth and requires surgery to correct. Eating and speech development are the most common concerns.

Facial Malformation – Including hemifacial microsomia, a condition in which the lower half of one side of the face is underdeveloped and does not grow normally. The syndrome varies in severity, but always includes the maldevelopment of the ear and the mandible. This is the second most common facial birth defect after clefts.

HEART

Congenital Heart Disease – Any of several congenital coronary defects ranging from a small hole in a wall of the heart (**minor**) to multiple defects or transposition, etc. (**major**). **Ventricular septal defect** is the most common heart malformation, in which an opening between the lower chambers of the heart increases blood flow, under high pressure, to the lungs. Small defects can usually close on their own during the first year of life, while more severe defects require surgery. “Minor” conditions typically include VSD, ASD, PFO, PDA, etc., and “Major” conditions typically include TOF, multiple or structural pathologies.

BLOOD

Hepatitis B – An infection of the liver caused by a DNA virus, transmitted by contaminated blood or blood derivatives in transfusions, by sexual contact with an infected person, or by the use of contaminated needles and instruments. The disease has a long incubation and symptoms that may become severe or chronic, potentially causing serious damage to the liver.

Thalassemia – An inherited form of anemia caused by faulty synthesis of part of the hemoglobin molecule.

SKELETAL

Arthrogryposis/Joint Disorders – A congenital disorder characterized by multiple joint contractures and can include muscle weakness and fibrosis. It is a non-progressive disease, typically symmetrical and involving all four extremities with some variation seen.



Club Foot – A deformity in which one or both feet are twisted out of position. Surgically correctable.



Missing/malformed fingers/toes, hands/arms, feet/legs – Any congenital abnormality or partial/total absence of the digits, including webbing, fusing, etc., or of one or more limbs.



Scoliosis – Mild to severe curvature of the spine.

Short Stature/Dwarfism – Most commonly caused by achondroplasia, a genetic condition that results in disproportionately short arms and legs. Orthopedic complications are not uncommon.



Spina Bifida – A congenital defect in which the spinal column is imperfectly closed so that part of the meninges or spinal cord protrudes, often resulting in hydrocephalus (abnormal accumulation of fluid in the cerebral ventricles) and other neurological disorders.

VISION/HEARING



Ear Malformation (microtia/atresia) – Usually a malformation of the ear auricle, often accompanied by a closed external ear canal.

Hearing Loss – Ranging from partial to total hearing loss.

Nystagmus – Rapid rhythmic repetitive eye movements.

Strabismus – A vision condition in which one eye cannot focus with the other on an object because of imbalance of the eye muscles. Also known as crossed eyes.

Ptosis – Abnormal lowering or drooping of the upper eyelid caused by muscle weakness or paralysis.



Cataracts – An opacity that develops in the crystalline lens of the eye or in its envelope. Cataracts typically progress slowly to cause vision loss and are potentially blinding if untreated.

Glaucoma – An eye disorder in which the optic nerve suffers damage, permanently impacting vision in the affected eye(s) and progressing to complete blindness if untreated.

Vision Loss – Ranging from partial to total vision loss.

SKIN



Albinism AND low vision – Lack of pigment in the skin, hair, and irises, often associated with vision problems.



Birthmark/Nevus (moderate to significant/facial) - A congenital growth or mark on the skin, such as a mole or other birthmark.



Hemangioma/Lymphangioma - A tissue mass of blood vessels or overgrown lymph vessels, also known as a “strawberry mark.”

Scar/Burns – An injury- or surgery-related scar, possibly of unknown cause, or burns, varying in degree due to fire or scalding, possibly of unknown cause.



Vitiligo – A skin disease causing the loss of pigmentation in which patients develop white spots in the skin that vary in size and location.

NERVOUS SYSTEM/DEVELOPMENTAL

Cerebral anoxia/Brain damage or malformation – A reduced supply of oxygen to the brain or other brain injury, varying in severity depending on location/cause/etc.

Cerebral Palsy – A disorder usually caused by brain damage occurring at or before birth and marked by muscular impairment. Often accompanied by poor coordination, it sometimes involves speech and learning difficulties.

Down Syndrome – A genetic condition in which a person has 47 chromosomes instead of the usual 46. Symptoms vary from person to person and can range from mild to severe. About half of children with Down syndrome are born with heart problems, including atrial septal defect, ventricular septal defect, and endocardial cushion defects. The level of mental retardation varies from patient to patient, but is usually moderate.

Hydrocephalus – Also known as “water on the brain,” an abnormal accumulation of fluid in the cerebral ventricles causing enlargement of the skull and compression of the brain.

Development Delay – Above and beyond expected or typical institutionalization delays.

GENITAL

Ambiguous Genitalia – Unclear external genitalia or presence of both genders’ genitalia (“intersex”).

Hypospadias – The urethral opening is located on the shaft or base of the penis rather than the tip. Varies in severity. Surgical correction allows for a better cosmetic appearance and normal functioning of the penis.

DIGESTIVE

Anal atresia/Imperforated anus – Narrow or lack of an anal opening. Surgically correctable.

Esophageal/Stomach Abnormalities – Any of a range of conditions encompassing malformation of the digestive tract.

Kidney Conditions – Including single kidneys or hydronephrosis, distention (dilation) of the kidney with urine, caused by backward pressure on the kidney when the flow of urine is obstructed, etc.

OTHER

Epilepsy/Seizure disorder – Any of various neurological disorders characterized by sudden recurring attacks of motor, sensory, or psychic malfunction with or without loss of consciousness or convulsive seizures.

Paralysis – Of one or more limbs, possibly with an unknown cause.

Teratoma – An encapsulated tumor, usually benign, containing tissues that although normal in themselves, may be quite different from surrounding tissues. Normally relatively easy to resect from surrounding tissues.

The information contained on these pages was gathered from medical contacts, resources, and websites. We are not medical professionals, so CCAI urges you to do thorough medical research on the special needs you are considering.