Glossary of medical terms (*grouped by affected system or organ*)

Atrial septal defect (ASD) – disorder of the heart that is present at birth involving a hole in the wall (septum) separating the two upper chambers (atria)

Ventricular septal defect (VSD) – disorder of the heart that is present at birth involving a hole in the wall (septum) separating the two lower chambers (ventricles)

Patent ductus arteriosus (PDA) – failure of the ductus arteriosus, an arterial shunt in fetal life, to close before birth (patent refers to remaining open)

Polyvalvular disease – damage or defect to a heart valve (mitral, aortic, tricuspid or pulmonary); mitral and tricuspid valves control the flow of blood between the atria and the ventricles

Pulmonary stenosis – narrowing or obstruction to blood flow (stenosis) from the right ventricle to the pulmonary artery

Coarctation of aorta – narrowing of the aorta, the large blood vessel that delivers oxygen-rich blood to the body

Bicuspid aortic valve - aortic valve separates the lower left chamber (left ventricle) of the heart from the aorta. A bicuspid aortic valve has two flaps (cusps) instead of the usual three. This condition is often present with coarctation of aorta.

Mitral valve atresia – mitral valve connects the two chambers on the left side of the heart (atrium and ventricle). In this condition, blood is unable to flow between the two chambers.

Hypoplastic aorta (hypoplastic left heart syndrome) – left side of the heart is critically underdeveloped so unable to effectively pump blood to the body and causing the right side of the heart to pump blood to the lungs and body.

Pulmonary hypertension – changes in pulmonary arteries affecting blood flow through blood vessels leading to elevated blood pressure in the pulmonary arteries.

Cardiac myopathy (cardiomyopathy) - disease of heart muscle, which makes it difficult for the heart to deliver blood to the body. Condition can lead to heart failure.

Apnea – temporary cessation of breathing; often occurs during sleep. There are two types of apnea: central and obstructive. The former is when brain doesn't send proper signals to the muscles that control breathing. The latter relates to the muscles of the throat relaxing.

Glossary of medical terms continued

Laryngomalacia – softening of the tissues of the larynx (voice box) above the vocal cords. This is the most common cause of stridor (noisy breathing) in infants. Some infants struggle with breathing and eating when diagnosed with this condition.

Tracheobronchomalacia – condition characterized by flaccidity (low tone, floppiness) of the tracheal support cartilage, which can lead to tracheal collapse

Esophageal atresia – condition in which upper esophagus (tube that leads from the throat to the stomach) ends and does not connect with the lower esophagus and stomach. Most infants with this condition also have tracheoesophageal fistula (see below).

Tracheoesophageal fistula - abnormal connection (fistula) between the esophagus and the trachea (tube that leads from the throat to the windpipe and lungs)

Cyanosis – bluish tint to the skin due to less than optimal oxygenation of red blood cells

Occipital bone – bone that forms the back and base of the skull, and through which the spinal cord passes; malformation can impact brain development

Palprebral fissures – opening between eyelids; may be smaller than expected, up slanted or down slanted

Hypoplastic orbital ridges – underdevelopment of brow ridge (forms the separation between the forehead portion and the roof of the eye sockets)

Strabismus – condition in which eyes do not properly align with each other (crossed eyes)

Amblyopia - vision in one eye is reduced because the eye and the brain are not working together properly because the brain is favoring the other (lazy eye)

Enophthalmos – posterior displacement of the eye into the orbit; exophthalmos is the term for the anterior displacement of the eye)

Microphthalmia (microphthalmos) – disorder of the eye in which one (unilateral microphthalmia) or both (bilateral microphthalmia) eyes are abnormally small and have one or more anatomic malformation(s)

Anophthalmia - absence of one or both eyes; both the globe (eye) and the ocular tissue are missing from the orbit (socket)

Glossary of medical terms continued

Philtrum – vertical groove in the middle of the upper lip; concern if it is flattened or smooth

Bulbous nose – rounded, pear shaped nose
Hypertelorism - increased distance between two organs or bodily parts, usually referring to an increased distance between the orbits (eyes)

Dandy Walker malformation – a brain malformation of the cerebellum affecting movement and coordination

Corpus callosum – bundle of nerve fibers joining the two hemispheres of the brain; may be partial or complete absence of corpus callosum affecting communication between hemispheres

Ventriculomegaly – brain condition that occurs when the lateral ventricles become dilated, fluid filled structures (lateral ventricles) in the brain are too large

Hydrocephaly – accumulation of cerebrospinal fluid (CSF) within the brain, typically causes increased pressure inside in the brain. May need placement of a shunt to drain the excess fluid.

Microcephaly – neurological condition in which an infant's head is significantly smaller than the heads of other children of the same age; often due to abnormal brain development

Holoprosencephaly – disorder caused by the failure of the prosencephalon (embryonic forebrain) to sufficiently divide into the double lobes of the cerebral hemispheres resulting in a single-lobed brain structure and severe skull and facial defects

Choroid plexus cysts – cysts that occur within choroid plexus of the brain (middle part of the fetal brain with spaces called ventricles with a spongy layer of cells and blood); often resolve before birth.

Omphalocele – birth defect of the abdominal wall in which intestines, liver, or other organs are outside of the belly through the umbilical cord (belly button). The organ(s) is (are) covered in a thin, nearly transparent sac. Surgery is required to return the organ(s) inside the body.

Aspiration – entry of secretions, liquids etc. into the larynx (voice box) and lower respiratory tract (from the trachea to the lungs); can be difficult to expel and lead to aspiration pneumonia and/or restrictions on oral intake

Dysphagia – difficulty in swallowing liquids or food items caused by a malformation or other defect of the throat or esophagus

Glossary of medical terms continued

Reflux – stomach acid or bile flows from the stomach into the esophagus and irritates the stomach lining causing discomfort or pain. May require medication and/or restrictions on intake. Referred to as gastroesophageal reflux disease if occurs more than twice a week.

Micrognathia – condition in which the jaw is undersized, associated with a variety of craniofacial conditions and may interfere with breathing and/or feeding. Surgery can correct this condition.

Cystic hygroma – cyst or group of cysts most commonly found in the neck ("webbed" neck) and armpits; believed to be caused by an error in the development of lymph sacs and lymph vessels as the baby develops during pregnancy. Treatment can be non-surgical or surgical removal.

Hydrops – severe swelling (edema) in an unborn baby or a newborn baby; considered life-threatening

Meckel's diverticulum – abnormal pouch or bulge in the intestine present from birth; treatment is needed if there is bleeding via a laparoscopic procedure or abdominal surgery

Diaphragmatic hernia – birth defect in which there is an abnormal opening in the diaphragm (muscle between the chest and abdomen that assists with breathing). The opening allows part of the organs from the abdominal area to move into the chest cavity near the lungs. Treatment usually requires surgery.

Anorectal atresia (imperforate anus) – absence of an opening at the bottom end of the intestinal anus; opening is either not present or it is in the wrong place. Requires immediate treatment after birth.

Radial aplasia – congenital defect which affects the formation of the radius bone in the arm. Radius is the lateral bone which connects to the wrist via the carpal bones. This condition manifests with either a short or absent radius bone in one or both arm(s) (unilateral, bilateral). Radial aplasia also results in the thumb being either partly formed or completely absent from one or both hands.

Polydactyly – additional digits on hand(s) or foot/feet

Syndactyly – two or more digits are fused together

Hydronephrosis – swelling of kidney due to build-up of urine; urine cannot drain out from the kidney to the bladder from a blockage or obstruction. Can affect one or both kidneys.

Glossary of medical terms continued

Renal cystic dysplasia – malformation of the kidney such that cysts replace kidney tissue and affect its functioning. Can affect one or both kidneys.

Polycystic kidney disease – numerous/clusters of fluid filled cysts grow in the kidneys. If too many cysts grow or become too large, the kidneys can become damaged.

Heptoblastoma – most common form of liver cancer in children aged birth to three

Rocker bottom feet (congenital vertical talus) – an anomaly of the foot characterized by a prominent calcaneus (heel bone) and a convex rounded bottom of the foot. Can affect one or both feet.

Prominent calcaneus – large heel bone (foundation of rear part of foot)

Hypoplastic nails – underdevelopment of nails on one or both hands or feet

Kyphosis – forward rounding of the back, may require bracing or surgery

Scoliosis – sideways curvature of the spine, may require bracing or surgery

Hemihyperplasia – one part or one side of the body grows more than the other (overgrowth) causing asymmetry

Cryptorchidism – undescended testicle(s); may require surgery

Definitions adapted from http://www.mayoclinic.org, https://www.medline.com and http://www.webmd.com