

World Database for Pediatric and Congenital Heart Surgery

Appendix D: Non Cardiac Congenital Anatomic Abnormality Terms and Definitions

Major Abnormality of Craniofacial (Head)

Any abnormality of the head including: **Choanal Atresia**, a congenital anomaly in which a bony or membranous occlusion blocks the passageway between the nose and pharynx; **Cleft lip**, a congenital anomaly consisting of one or more clefts in the upper lip that results from failure of the maxillary and median nasal process to close during embryonic development; **Cleft Palate**, a congenital fissure in the roof of the mouth, resulting from incomplete fusion of the palate during embryonic development. It may involve only the uvula or extend through the entire palate

Major Abnormalities of the Abdominal Wall.

Any abnormality of the abdominal wall including: **Congenital Diaphragmatic Hernia (CDH)**, A developmental defect of the diaphragm that allows abdominal viscera to herniate into the chest; **Gastroschisis**, a congenital defect characterized by a defect in the abdominal wall through which intestines protrude; **Omphalocele**, a defect in the medial anterior abdominal wall through which intraabdominal contents are extruded, usually at the base of the umbilical cord

Major Abnormality of the Biliary System

Biliary atresia is characterized by absence or discontinuity of the extrahepatic biliary system, resulting in obstruction to bile flow.

Major Abnormality of the Brain

Any abnormality of the brain including: **Hydrocephalus**, excessive CSF accumulation in the brain creating potentially harmful pressures; **Macrocephaly**, defined as a head circumference which is greater than 2 standard deviations larger than the average for age and sex; **Microcephaly**, defined as smaller than normal circumference of the head because the cerebral cortex has not developed properly or has stopped growing.

Major Abnormality of the Diaphragm

A developmental defect of the diaphragm that allows abdominal viscera to herniate into the chest.

Major Abnormality of the Gastrointestinal System

Any abnormality of the gastrointestinal system including: **Duodenal atresia and stenosis**, an absence or narrowing of the duodenum; **Jujenal atresia or stenosis**, an absence or narrowing of the middle section of the small bowel; **Ileal atresia or stenosis**, and absence or narrowing of a portion of the ileum; **Intestinal malrotation**, an abnormal placement and fixation of intestine; **Hirschsprung's disease**, A disorder of the enteric nervous system characterized by an absence of ganglion cells in the distal colon resulting in functional obstruction; **Colonic Atresia or stenosis**, an absence or narrowing of the large bowel extending to the rectum; **Rectal atresia or stenosis**, absence or narrowing of a portion of the rectum; **Imperforate anus**, a specific type atresia or the anal canal with or without a fistulous opening to an ectopic location on the perineum, within the urinary system, or into the vaginal vestibule

Q: If a patient has a condition that was corrected, for example, "imperforate anus", should this still be recorded?

A: Yes.

Major Abnormality of the Kidney, Ureter, or Bladder

A major abnormality of the kidney(s), ureter(s) or bladder

Major Abnormality of the Larynx-Trachea- or Bronchus

Any abnormality including: **Laryngomalacia**, abnormal laxity of the laryngeal support cartilage resulting in excessive inward collapse and collapse of the lumen with inspiration during spontaneous ventilation. Characterized by inspiratory stridor; **Congenital Tracheal Stenosis**, a primary tracheal narrowing at any level between the larynx and carina with significantly smaller than expected luminal diameter (not secondary to trauma or prolonged intubation). Frequently related to complete cartilagenous tracheal rings; **Trachesophageal Fistula (TEF)**, the presence of any type of patent communication below the larynx connecting the tracheo-bronchial tree to the esophagus. May be associated with other anomalies, including VATER, VACTERL and tracheal clefts. Typically, congenital, but may occur due to trauma or pressure necrosis; **Bronchomalacia**, A deficiency in the cartilaginous wall of the bronchus that may lead to atelectasis or obstructive emphysema.

Major Abnormality of the Lung

Any abnormality of the lung including; **Congenital Lobar Emphysema (CLE)**, a developmental anomaly of the lower respiratory tract characterized by isolated hyperinflation of a lobe in the absence of extrinsic bronchial obstruction; **Cystic Congenital Adenomatous Malformation(CAM)**, a spectrum of cystic and solid lesions of the lung that result from abnormal embryogenesis and typically present with symptoms of respiratory distress in newborns and infants; **Cystic Fibrosis**, is an autosomal recessive genetic disorder affecting most critically the lungs, and also the pancreas, liver, and intestine. It is characterized by abnormal transport of chloride and sodium across an epithelium, leading to thick, viscous secretions; **Pulmonary Lymphangiectasia**, a rare developmental disorder involving the lung characterized by pulmonary subpleural, interlobar, perivascular and peribronchial lymphatic dilatation. PL presents at birth with severe respiratory distress, tachypnea and cyanosis, with a very high mortality rate at or within a few hours.

Major Abnormality of the Spine and Spinal cord

Any abnormality of the spine or spinal cord including: **Myelomeningocele**, a defect in which a portion of the spinal cord protrudes through a gap in the vertebral column, frequently accompanied by hydrocephalus and mental retardation; **Spina Bifida**, characterized by defective closure of the vertebral canal with herniation of the spinal cord and or meninges; **Spinal Scoliosis**, a lateral curve in the spine, usually combined with rotation of the vertebra

Q: I have a patient who was adopted at age of 2 years old, and this information is unknown. What should I enter?

A: If not known to be present, then nothing would be recorded.