Birth Defects Profile - Definitions

Indicator	Definition
Total birth defects	**The number of infants with one or more of the birth defects listed below.**
Cardiovascular Conditions	
Critical Congenital Heart Defects	Common truncus, dextro-Transposition of the Great Arteries (d-TGA), Tetralogy of Fallot, pulmonary valve atresia and stenosis, hypoplastic left heart syndrome, total anomalous pulmonary venous return, tricuspid valve atresia and stenosis, coarctation of aorta, double outlet right ventricle, Ebstein anomaly, interrupted aortic arch, and single ventricle
Aortic valve stenosis	Obstruction or narrowing of the aortic valve, which may impair blood flow from the left ventricle to the aorta.
Atrial septal defect	An opening in the wall (septum) that separates the left and right top chambers (atria) of the heart.
Atrioventricular septal defect (endocardial cushion defect)	A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum. Together, these defects producing a large opening (canal) in the central part of the heart.
Coarctation of aorta	Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body.
Common truncus (truncus arteriosus)	Failure of separation of the aorta and the pulmonary artery during development, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs.
Double outlet right ventricle	The pulmonary artery and the aorta both arise from the right ventricle.
Ebstein anomaly	Abnormal formation and downward displacement of the tricuspid valve into the right ventricle.
Hypoplastic left heart syndrome	A condition in which the structures on the left side of the heart and the aorta are extremely small.
Interrupted aortic arch	Complete loss of communication (interruption) between the ascending and descending aorta, usually associated with a malalignment-type ventricular septal defect (VSD).
Pulmonary valve atresia and stenosis	Congenital heart condition characterized by absence or constriction of the pulmonary valve.
Single ventricle	Instead of two separate ventricles, there is only one morphological ventricle, most commonly a double-inlet left ventricle.Disorder affecting one lower chamber of the heart. The chamber may be smaller, underdeveloped, or missing a valve.
Tetralogy of Fallot	The simultaneous presence of a ventricular septal defect (VSD), pulmonic and subpulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy.

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Total anomalous pulmonary venous return	A condition in which all 4 pulmonary veins connect anomalously into the systemic venous circulation to the right atrium or the body (systemic veins) instead of the left atrium.
Transposition of the great arteries (TGA)	Transposition of the aorta and the pulmonary artery such that the aorta arises from the right ventricle (instead of the left) and the pulmonary artery arises from the left ventricle (instead of the right).
Tricuspid valve atresia and stenosis	Obstruction or narrowing of the tricuspid valve, which may impair blood flow from the right atrium to the right ventricle.
Ventricular septal defect	An opening in the wall (septum) that separates the left and right ventricles of the heart.
Central Nervous System Conditions	
Anencephalus	Partial or complete absence of the brain and skull.
Encephalocele	Herniation of brain tissue and/or meninges through the skull. The hernia sac is usually covered by skin.
Holoprosencephaly	Structural brain anomaly in which the brain fails to cleave.
Spina bifida without anencephalus	Incomplete closure of the spine through which spinal cord tissue and/or the membranes covering the spine herniate.
Chromosomal Conditions	
Deletion 22q11.2	Chromosome abnormality resulting from genomic microdeletions within a critical region on the long arm of chromosome 22 (22q11.2)
Trisomy 13	The presence of three copies of all or a large part of chromosome 13.
Trisomy 18	The presence of three copies of all or a large part of chromosome 18.
Trisomy 21 (Down syndrome)	The presence of three copies of all or a large part of chromosome 21.
Turner syndrome	Presence of an absent or structurally abnormal second X chromosome in a phenotypic female.

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Ear and Eye Conditions	
Anophthalmia/microphthalmia	Total absence of eye tissue or absence of the globe / Reduce volume of the eye
Anotia/microtia	Congenital absence of one or both ears / malformation or small external ear
Aniridia	The complete absence of the iris of the eye or a defect of the iris.
Congenital cataract	Clouding of the lens of the eye that is present at birth.
Gastrointestinal Conditions	
Biliary atresia	Congenital absence of the lumen of the extrahepatic bile ducts.
Esophageal atresia/tracheoesophageal fistula	The esophagus ends in a blind pouch and fails to connect with the stomach / abnormal communication between the esophagus and the trachea.
Rectal and large intestinal atresia/stenosis	Complete or partial occlusion of the lumen of one or more segments of the large intestine and/or rectum.
Small intestinal atresia/stenosis	Complete or partial occlusion of the lumen of one or more segments of the small intestine.
Genitourinary Conditions	
Bladder exstrophy	A defect in the lower abdominal wall and anterior wall of the bladder through which the lining of the bladder is exposed to the outside.
Cloacal exstrophy	Congenital persistence of a common cloacal cavity into which gut, urethra, and reproductive tracts open with exstrophy of the cavity.
Congenital posterior urethral valves	Posterior urethral valves (PUV) are tissue folds of the posterior urethra and function as valves obstructing urine outflow.
Hypospadias	Displacement of the opening of the urethra
Renal agenesis/hypoplasia	Absence or incomplete development of the kidney

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Musculoskeletal Conditions	
Limb deficiencies (reduction defects)	Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), fingers (phalanges), thigh (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).
Clubfoot	An abnormality consisting of plantar flexion (downward pointing of the foot and toes), inversion (varus, or internal rotation), and metatarsus adductus (deviation of the forefoot toward the body) of the foot.
Craniosynostosis	Premature closure (fusion) of one or several cranial sutures (connective tissue membranes that separate the bones of the developing skull)
Diaphragmatic hernia	Incomplete formation of the diaphragm through which a portion of the abdominal contents herniate into the thoracic cavity.
Gastroschisis	A congenital opening or fissure in the anterior abdominal wall lateral to the umbilicus through which the small intestine, part of the large intestine, and occasionally the liver and spleen, may herniate.
Omphalocele	A defect in the anterior abdominal wall in which the umbilical ring is widened, allowing herniation of abdominal organs, including the small intestine, part of the large intestine, and occasionally the liver and spleen, into the umbilical cord. The herniating organs are covered by a nearly transparent membranous sac.
Orofacial Conditions	
Choanal atresia	Congenital obstruction of the opening of the nasal cavity into the nasopharynx on either side.
Cleft lip with cleft palate	A defect in the upper lip resulting from incomplete fusion of the parts of the lip, with an opening in the roof of the mouth.
Cleft lip alone	A defect in the upper lip resulting from incomplete fusion of the parts of the lip.
Cleft palate alone	An opening in the roof of the mouth resulting from incomplete fusion of the shelves of the palate.

Data Source: Florida Birth Defects Registry, July 2018