

## **Birth Defects Included in Maine's Reportable List**

### **Central Nervous System**

- ◇ **Anencephalus** - Partial or complete absence of the brain and skull.
- ◇ **Spina Bifida without Anencephalus** - Incomplete closure of the vertebral spine (usually posteriorly) through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate.
- ◇ **Hydrocephalus without Spina Bifida** – An increase in the amount of cerebrospinal fluid (CSF) within the brain resulting in enlargement of the cerebral ventricles and increased intracranial pressure.
- ◇ **Encephalocele** - Herniation of brain tissue and/or meninges through a defect in the skull. The hernia sac is usually covered by skin.
- ◇ **Microcephalus** – A cranial vault that is smaller than normal for age.

### **Eye**

- ◇ **Anophthalmia** – Total absence of eye tissue or apparent absence of the globe in an otherwise normal orbit.
- ◇ **Microphthalmia** – Reduced volume of the eye.
- ◇ **Congenital cataract** – An opacity of the lens of the eye that has its origin prenatally
- ◇ **Aniridia** – Hypoplasia of the iris of both eyes.

### **Ear**

- ◇ **Anotia** – Total absence of the external ear and canal
- ◇ **Microtia** – Malformation or hypoplasia of the external ear (auricle, pinna)

### **Cardiovascular**

- ◇ **Common Truncus (Truncus Arteriosus or TA)** - Failure of separation of the aorta and the pulmonary artery, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs.
- ◇ **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).

- ◇ **Tetralogy of Fallot** - The simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy.
- ◇ **Ventricular septal defect** – An opening in the septum that separates the left and right ventricles of the heart.
- ◇ **Atrial septal defect** – An opening in the septum that separates the left and right atria of the heart.
- ◇ **Endocardial cushion defect** – A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum, producing a large opening (canal) in the central part of the heart.
- ◇ **Pulmonary Valve Atresia** - Lack of patency, or failure of formation altogether, of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the pulmonary artery.
- ◇ **Pulmonary Valve Stenosis** – Obstruction or narrowing of the pulmonary valve, which may impair blood flow from the right ventricle to the pulmonary artery.
- ◇ **Tricuspid Valve Atresia** - Lack of patency, or failure of formation altogether, of the tricuspid valve, resulting in obstruction of blood flow from the right atrium to the right ventricle.
- ◇ **Tricuspid Valve Stenosis** – Obstruction or narrowing of the tricuspid valve, which may impair blood flow from the right atrium to the right ventricle.
- ◇ **Ebstein’s anomaly** – Downward displacement of the tricuspid valve into the right ventricle.
- ◇ **Aortic valve stenosis** – Obstruction or narrowing of the aortic valve, which may impair blood flow from the left ventricle to the aorta.
- ◇ **Hypoplastic Left Heart Syndrome (HLHS)** - A condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves, and hypoplasia and coarctation of the aorta.
- ◇ **Patent ductus arteriosus** – Abnormally persistent blood flow through the ductus arteriosus beyond the first few days of life.

- ◇ **Coarctation of the Aorta** - Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body.

### **Orofacial**

- ◇ **Cleft Palate without Cleft Lip** - An opening in the roof of the mouth resulting from incomplete fusion of the shelves of the palate. The opening may involve the hard palate only, the soft palate only, or both.
- ◇ **Cleft Lip with and without Cleft Palate** - A defect in the upper lip resulting from incomplete fusion of the parts of the lip.
- ◇ **Choanal atresia** – Congenital obstruction of the opening of the nasal cavity into the nasopharynx on either side.

### **Gastrointestinal**

- ◇ **Esophageal atresia** – A condition in which the esophagus ends in a blind pouch and fails to connect with the stomach.
- ◇ **Tracheoesophageal fistula** – An 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.
- ◇ **Pyloric stenosis** – hypertrophy (thickening) of the muscles of the pylorus connecting the stomach to the duodenum, resulting in complete or partial obstruction of the passage of food and gastric contents.
- ◇ **Hirschsprung's disease (congenital megacolon)** – Absence of the parasympathetic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum, which may result in congenital megacolon. Megacolon – enlargement of the diameter of part or all of the colon.
- ◇ **Biliary atresia** – Congenital absence of the lumen of the extrahepatic bile ducts.

### **Genitourinary**

- ◇ **Renal agenesis** – Complete absence of the kidney.
- ◇ **Renal hypoplasia**- Incomplete development of the kidney.
- ◇ **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.

- ◇ **Obstructive genitourinary defect** – Partial or complete obstruction of the flow of urine at any level of the genitourinary tract from the kidney to the urethra.
- ◇ **Hypospadias** - Displacement of the opening of the urethra (urethral meatus) ventrally and proximally (underneath and closer to the body) in relation to the tip of the glans of the penis.
- ◇ **Epispadias** – Displacement of the opening of the urethra (urethral meatus) dorsally and proximally (on the top and closer to the body) in relation to the tip of the glans of the penis.

### **Musculoskeletal**

- ◇ **Reduction Deformity, Upper Limbs** - Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), or fingers (phalanges).
- ◇ **Reduction Deformity, Lower Limbs** - Complete or partial absence of the upper leg (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).
- ◇ **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.
- ◇ **Congenital hip dislocation** – Location of the head of the femur (bone of the upper leg) outside its normal location in the cup-shaped cavity formed by the hip bones (acetabulum).
- ◇ **Diaphragmatic hernia** – Incomplete formation of the diaphragm through which a portion of the abdominal contents herniated into the thoracic cavity.

### **Chromosomal**

- ◇ **Trisomy 13** – The presence of three copies of all or a large part of chromosome 13.

- ◇ **Down Syndrome (Trisomy 21)** - The presence of three copies of all or a large part of chromosome 21.
- ◇ **Trisomy 18** – The presence of three copies of all or a large part of chromosome 18

**Other**

- ◇ **Fetal alcohol syndrome** – A spectrum of abnormalities resulting from exposure to alcohol in utero. While the specific abnormalities vary among individuals, the hallmarks include growth deficiency, microcephaly, facial dysmorphisms, and neurodevelopmental abnormalities.
- ◇ **Amniotic bands** – Strands of tissue that float in the amniotic fluid as a consequence of tears or ruptures in the amniotic membrane which surrounds the fetus during development.