Chapter HFS 116

APPENDIX A

BIRTH DEFECTS AND SYNDROMES FOR WHICH REPORTING IS MANDATORY

Achondroplasia	Microphthalmia and Anophthalmia
Ambiguous Genitalia	Microtia/Anotia
Amniotic Bands	Multicystic or Dysplastic Kidney
Anencephaly	Noonan Syndrome
Angelman Syndrome	Obstructive Urinary Tract Defect [not posterior valves; not urethral stenosis/atresia]
Arthrogryposis Multiplex Congenita	Oculoauriculovertebral Association (including Goldenhar Association and Hemifacial Microsomia)
Atrial Septal Defect	Omphalocele
AV Canal/Endocardial Cushion Defect	Osteogenesis Imperfecta
Beckwith−Wiedemann Syndrome	Other Chromosomal Anomaly (not +13, +18, +21, XXY, Turner S., 22q−)
Biliary Atresia	Polycystic Kidney Disease, Autosomal Dominant Form
Bone Dysplasia/Dwarfism, Other (not Achondroplasia)	Polycystic Kidney Disease, Autosomal Recessive Form
Cardiac Arrhythmia (Congenital)	Polycystic Kidney Disease, Uncertain Form
Cataract (Congenital or Early)	Porencephaly
CHARGE Association	Posterior Urethral Valves
Choanal Atresia	Prader−Willi Syndrome
Cleft Lip with or without Cleft Palate	Pyloric Stenosis
Cleft Palate	Rectal/Colonic Atresia/Stenosis
Clubfoot (Congenital)	Reduction Deformity, Arm or Hand
Coarctation of the Aorta	Reduction Deformity, Leg or Foot
Coloboma	Renal Agenesis/Hypoplasia
Craniosynostosis	Robin Malformation Sequence (Pierre Robin Sequence)
Cystic Fibrosis	Scoliosis or Kyphosis/Hemivertebra (Infantile)
De Lange Syndrome (Cornelia De Lange Syndrome)	Small Bowel Atresia/Stenosis
Diaphragmatic Hernia	Smith−Lemli−Opitz Syndrome
Down Syndrome	Sotos Syndrome
Encephalocele	Spina Bifida
Epispadias	Spinal Muscular Atrophy (Infantile)
Exstrophy of the Bladder/Cloaca	Stickler Syndrome
Gastroschisis	Tetralogy of Fallot
Glaucoma (Congenital)	Total Anomalous Pulmonary Venous Return
Hemivertebra	Tracheo−Esophageal Fistula/Esophageal Atresia
Hemophilia	Transposition of the Great Vessels
Hereditary Spherocytosis	Trisomy 13
Hip Dislocation (Congenital)/Developmental Dysplasia of Hip (Congenital)	Trisomy 18
Hirschsprung Disease	Trisomy 21
Holoprosencephaly	Truncus Arteriosus
Hydranencephaly	Turner Syndrome
Hydrocephalus (Congenital or Early)	Urethral Stenosis/Atresia
Hypoplastic Left Heart	Valvular Heart Disease (Congenital)
Hypospadias	VATER Association
Hypothyroidism (Congenital)	Velocardiofacial Syndrome (22q Deletion Syndrome)
Klinefelter Syndrome	Ventricular Septal Defect
Marfan Syndrome	Von Willebrand Disease
Microcephaly (Congenital or Early)	Williams Syndrome

Note: Definitions can be found in the *Birth Defects Encyclopedia: The Comprehensive, Systematic, Illustrated Reference Source for the Diagnosis, Delineation, Etiology, Biodynamics, Occurrence, Prevention, and Treatment of Human Anomalies of Clinical Relevance,* Volumes I and II, Centers for Birth Defects Information Services, Inc. 1990.