

LIST OF MAINE REPORTABLE BIRTH DEFECTS

Birth Defect	ICD-10-CM Codes
Central Nervous System	
Anencephalus	Q00.0-Q00.1
Encephalocele	Q01-Q01.9
Microcephalus	Q02
Holoprosencephaly	Q04.2
Spina Bifida without anencephalus	Q05.0-05.9 Q07.01, 07.03 w/o Q00.0-00.1
Eye	
Anophthalmia/microphthalmia	Q11.0-Q11.2
Congenital cataract	Q12.0
Ear	
Anotia/microtia	Q16.0, Q17.2
Cardiovascular	
Common truncus (truncus arteriosus or TA)	Q20.0
Double outlet right ventricle (DORV)	Q20.1
Single Ventricle	Q20.4
Transposition of great arteries	Q20-Q20.9
Tetralogy of Fallot	Q21.3
Ventricular septal defect	Q21.0
Atrial septal defect	Q21.1
Atrioventricular septal defect (Endocardial cushion defect)	Q21.2
Pulmonary valve atresia and stenosis	Q22.0, 22.1
Tricuspid valve atresia and stenosis	Q22.4
Ebstein's anomaly	Q22.5
Aortic valve stenosis	Q23.0
Hypoplastic left heart syndrome	Q23.4
Coarctation of aorta	Q25.1
Interrupted aortic arch (IAA)	Q25.2, Q25.4 post 2016 25.21

Total anomalous pulmonary venous connection (TAPVC)	Q26.2
Orofacial	
Choanal atresia	Q30.0
Cleft palate without cleft lip	Q35.1 - Q35.9
Cleft lip with and without cleft palate	Q36.0 - 36.9, Q37.0 - Q37.9
Gastrointestinal	
Esophageal atresia/tracheoesophageal fistula	Q39.0 - 39.4
Small intestinal atresia/stenosis	Q41.0 – Q41.9
Rectal and large intestinal atresia/stenosis	Q42.0 - Q42.9
Biliary atresia	Q44.2 – Q44.3
Genitourinary	
Hypospadias	Q54.0 - Q54.9 (excluding Q54.4)
Renal agenesis/hypoplasia	Q60 – Q60.6
Bladder exstrophy	Q64.10 – Q64.19
Cloacal exstrophy	Q64.12
Congenital Posterior Urethral Valves	Q64.2
Musculoskeletal	
Clubfoot	Q66.0, Q66.89
Reduction deformity, upper limbs	Q71.0-Q71.9, 73.0 – Q73.8
Reduction deformity, lower limbs	Q72.0- Q72.9
Craniosynostosis	Q75.0
Gastroschisis	Q79.3
Omphalocele	Q79.2
Diaphragmatic hernia	Q79.0, Q79.1
Chromosomal	
Down syndrome (Trisomy 21)	Q90.0 – Q90.9
Trisomy 18	Q91.0 – Q91.3
Trisomy 13	Q91.4 – Q91.7
Deletion 22q11	Q93.81
Turner syndrome	Q96.0 – Q96.9