

Reportable Birth Defects Included in Case Definition

Birth Defect	ICD-10-CM Codes
Central Nervous System	
Anencephalus	Q00.0-Q00.1
Spina Bifida without anencephalus	Q05.0-Q05.9 Q07.01, Q07.03 w/o Q00.0-Q00.1
Encephalocele	Q01-Q01.9
Microcephalus	Q02
Holoprosencephaly	Q04.2
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
Interrupted aortic arch (IAA)	Q25.2, Q25.4 post 2016 25.21
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

Total anomalous pulmonary venous connection (TAPVC)	Q26.2
Single Ventricle	Q20.4
Orofacial	
Cleft palate without cleft lip	Q35.1 - Q35.9
Cleft lip with and without cleft palate	Q36.0 - 36.9, Q37.0 - Q37.9
Choanal atresia	Q30.0
Gastrointestinal	
Esophageal atresia/tracheoesophageal fistula	Q39.0 - 39.4
Rectal and large intestinal atresia/stenosis	Q42.0 - Q42.9
Biliary atresia	Q44.2 – Q44.3
Small intestinal atresia/stenosis	Q41.0 – Q41.9
Genitourinary	
Renal agenesis/hypoplasia	Q60 – Q60.6
Bladder exstrophy	Q64.10 – Q64.19
Hypospadias and Epispadias	Q51.0 - Q54.9 (excluding Q54.4)
Cloacal exstrophy	Q64.12
Congenital Posterior Urethral Valves	Q64.2
Musculoskeletal	
Reduction deformity, upper limbs	Q71.0-Q71.9, 73.0 – Q73.8
Reduction deformity, lower limbs	Q72.0- Q72.9
Gastroschisis	Q79.3
Omphalocele	Q79.2
Diaphragmatic hernia	Q79.0, Q79.1
Clubfoot	Q66.0, Q66.89
Craniosynostosis	Q75.0
Chromosomal	
Trisomy 13	Q91.4 – Q91.7
Down syndrome (Trisomy 21)	Q90.0 – Q90.9
Trisomy 18	Q91.0 – Q91.3
Deletion 22q11	Q93.81
Turner syndrome	Q96.0 – Q96.9
Other	