Reportable Birth Defects Included in Case Definition

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| **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 |

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| 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** | | |