Appendix 2A: NBDPN Birth Defect Case Definition Criteria Descriptions

Contents

Preface to Appendix 2A

Case Definitions

This document introduces two considerations that should be used for counting birth defects in surveillance data: the type of surveillance performed for case investigation and verification, and the level of certainty about the diagnosis.

- Case Investigation and Verification Types
  - **Tier 1 Surveillance:** The process of case investigation, verification, and classification is based on review and/or abstraction of medical record information, such as clinical notes, laboratory testing, or imaging data.
  - **Tier 2 Surveillance:** The case is reviewed and classified based only on coded or written diagnosis data. Case investigation, verification, and classification do not involve review of medical record information.
- Levels of Certainty
  - ***Confirmed***: Case data meet criteria that give the diagnosis a high level of certainty.
  - ***Accepted***: Case data meet criteria that give the diagnosis an acceptable level of certainty.

Confirmed conveys more certainty about the diagnosis than *Accepted.* A birth defects surveillance program can select which process is most appropriate for each defect, with the understanding that the level of surveillance and types of data available may vary within a single program. These tiers are designed to aid in standardization when aggregating data across jurisdictions and ensure that data collected and reviewed using different methods can be consolidated appropriately.

Generally, the Tier 2 surveillance definitions only allow for an *Accepted* classification. However, in some cases a condition may be classified as *Confirmed* based on code alone, diagnosis alone, the diagnosis being made independently by multiple providers, or the diagnosis code being consistently entered across multiple encounters (without abstraction/review of additional supporting medical information). Those exceptions are called out within the condition definition, and citations are included within the document to justify achieving a *Confirmed* level of certainty using only coded diagnosis data, based on evidence of high positive predictive value (PPV) utilizing these methods. Future updates to case definitions can consider a broader approach to implementation of *Confirmed* definitions for Tier 2 surveillance.

Case Ascertainment (Identification) versus Case Classification

The condition definitions below include codes relevant to both case ascertainment and case classification. Definitions of these terms are provided below:

- ***Case Ascertainment***: Identifying and gathering information on potential cases of a disease within a population
- ***Case Classification***: Evaluating identified potential cases and assigning an official status based on standardized criteria

Generally, the presence of case classification codes is sufficient to count a case when using the Tier 2 definitions (as either *Accepted* or *Confirmed*, depending on the specific condition). Case ascertainment codes are broader and are not sufficient on their own to count a case in surveillance

data. The presence of these codes may be used to flag a record for further review, but more information is needed to meet a confirmed or accepted case definition.

Usage Notes

- When considering diagnoses for case classification, do not include diagnoses that are listed as one of several equally-weighted differential diagnoses in the clinical notes included in the medical record. For example, if the clinician notes that they are considering an atrial septal defect along with several other potential diagnoses, that should not be counted for surveillance without any further confirmatory findings.
- Postnatal findings always supersede prenatal findings.
- Findings from appropriate diagnostic imaging, laboratory testing, surgery, or autopsy (when available) will supersede previous diagnoses based solely on physical examination findings.
- Many of the birth defect conditions may co-occur as part of a particular syndrome. In these cases, each of the concurrent major malformations should be coded separately (see **Recommendation 5.3.3b**) unless otherwise specified in the definition, since their presence may vary among affected individuals.
- Generally, the presence of a syndrome diagnosis can be used for case ascertainment but further follow-up and confirmation of the specific defect(s) are required for case classification.
- Electronic case reporting (eCR) utilizes the presence of ICD-10-CM, SNOMED-CT, and LOINC codes to trigger an electronic initial case report (eICR) to be sent to public health from a healthcare organization for case ascertainment. The eICR document can contain information about procedures, clinical notes, and imaging results that may or may not be sufficient for verifying if a potential case meets the Tier 1 surveillance case definitions. When utilizing eICR data, the birth defects surveillance program should consider the data quality found within their eICRs and how eICRs fit into their broader surveillance strategy to determine which case definition applies to their surveillance method for the condition.

Standard Code Systems

- The following standard code systems are utilized in the NBDPN case definitions:
    - Diagnoses: ICD-10-CM and SNOMED CT
    - Laboratory Testing: LOINC
- ICD-9 codes were included in previous versions of the Guidelines but are now deprecated. Please refer to earlier versions of this document (e.g., NBDPN Guidelines Appendix 3.1: Birth Defects Descriptions for NBDPN Core, Recommended, and Extended Conditions, published March 2021) if ICD-9 codes are needed for historical analysis.
- CDC/BPA codes are not typically used for reporting from healthcare providers to public health agencies (PHA) but are sometimes used for reporting from PHA to CDC. Therefore, they are included for informational purposes but are not a part of the case definitions.
    - Some programs use the 6th digit of a CDC/BPA code to denote information related to the defect laterality or type of diagnosis. To allow for this functionality where appropriate, some CDC/BPA codes in this document are listed with only 5 digits.

VSAC and OIDs

This document references value sets stored on the National Library of Medicine (NLM) tool, the Value Set Authority Center (VSAC). The value set object identifiers (OIDs) relevant for each condition are directly linked in the document, and the VSAC homepage can be accessed at this link: https://vsac.nlm.nih.gov/welcome

A UMLS account is required to view value sets on VSAC. Instructions on how to request a UMLS account are found at this link: https://www.nlm.nih.gov/vsac/support/usingvsac/requestumlslicense.html

The value sets referenced are from the Reportable Conditions Knowledge Management System (RCKMS), and are created and maintained by the RCKMS Content Team. At the time of publication, not all birth defect conditions in this document are available in RCKMS. Those conditions will not have associated value set OIDs, but have relevant codes listed individually.

NBDPN Defect Tiers

The Core and Enhanced defect tiers have been assigned to help programs prioritize high-impact conditions for national surveillance standardization. These designations are not intended to limit programs from conducting surveillance on conditions of interest within their jurisdiction. However, it is recommended that programs prioritize meeting quality standards for Core conditions before expanding to surveillance activities to Enhanced conditions (see **Recommendation 2.2.1a**). Core conditions are intended to be those that allow for comparable surveillance results even though case identification, investigation, and verification procedures vary across different birth defects surveillance programs and jurisdictions.

New NBDPN Defect Tiers have been assigned as Core based on the following factors:

- >90% of cases diagnosed before 1 year of age
- >90% of cases are live births
- Ease of surveillance, including consideration for whether the condition has a spectrum of severity that may impact the consistency and completeness of diagnosis and reporting, or the need for specialized testing.

Tier	Category	Conditions
Core	Cardiovascular	Aortic valve stenosis Atrioventricular septal defect (Endocardial cushion defect) Coarctation of the aorta Common truncus (truncus arteriosus or TA) Double outlet right ventricle (DORV) Ebstein anomaly Hypoplastic left heart syndrome (HLHS) Interrupted aortic arch (IAA) Single Ventricle Tetralogy of Fallot (TOF) Total anomalous pulmonary venous connection (TAPVC) Transposition of the great arteries (TGA)
	Orofacial	Cleft lip alone (without cleft palate) Cleft lip with cleft palate
	Gastrointestinal	Biliary atresia Esophageal atresia Rectal and large intestinal atresia/stenosis Small intestinal atresia/stenosis
	Musculoskeletal	Craniosynostosis Diaphragmatic hernia Gastroschisis
	Chromosomal	Trisomy 21 (Down syndrome)
Enhanced	Central Nervous system	Anencephaly Encephalocele Holoprosencephaly Spina bifida
	Eye	Anophthalmia / microphthalmia Congenital cataract
	Ear	Anotia/microtia
	Cardiovascular	Atrial septal defect Pulmonary valve atresia Pulmonary valve stenosis Tricuspid valve atresia and stenosis Ventricular septal defect
	Orofacial	Choanal atresia Cleft palate alone (without cleft lip)
	Genitourinary	Bladder exstrophy Cloacal exstrophy Congenital Posterior Urethral Valves Hypospadias Renal agenesis/hypoplasia
	Musculoskeletal	Clubfoot Limb deficiencies (reduction defects) Omphalocele
	Chromosomal	Deletion 22 q11 Trisomy 13 Trisomy 18 Turner syndrome

Changes Compared to Prior Editions

- **Anencephaly:**
  Acrania is no longer included under Anencephaly. Though the terms are sometimes used interchangeably, they are distinct conditions. Acrania is a rare malformation where the skull is partially or completely absent, but there is a normal or near-normal brain, typically covered only by a thin layer of membrane. Acrania is now included in Case Ascertainment for anencephaly, as the cases of acrania require further review to be potentially classified as anencephaly.
- **Spina bifida:**
  Removed "without anencephaly" because recommendations for coding these defects separately are now included.
- **Pulmonary valve atresia and stenosis:**
  Pulmonary valve atresia and pulmonary valve stenosis have been separated into two separate conditions to enable easier tracking and surveillance across the spectrum of severity of these conditions. This is enabled by the presence of separate ICD-10-CM codes for pulmonary valve atresia and pulmonary valve stenosis.
- **Transposition of the great arteries (TGA):**
  Corrected transposition/levo-transposition/L-transposition/discordant atrioventricular connection of the great vessels is now excluded from this condition. Corrected transposition is clinically distinct in that the pulmonary and systemic circulations are "normal" and are still in series. Typically, corrected transposition is acyanotic, and the clinical issues arise from associated defects (VSD, heart block, tricuspid valve insufficiency) and right ventricular failure over time.
- **Cleft Palate Alone:**
  Cleft uvula was included in previous versions of this condition but is now excluded due to its mild presentation and high prevalence. Cleft uvula typically does not require treatment and cases should not be included for surveillance, but may be monitored for potential other craniofacial defects.
- **Esophageal Atresia/Tracheoesophageal Fistula:**
  Tracheoesophageal Fistula alone has been removed from Esophageal Atresia/ Tracheoesophageal Fistula, so that the listed defect is now just Esophageal Atresia. Tracheoesophageal Fistula is rarely identified in the window monitored by birth defect programs and sometimes even diagnosed as late as adulthood. In some instances, TE fistula without esophageal atresia may not be diagnosed until weeks, months, or even a year or more after birth if the communication between the esophagus and trachea is small and symptoms are intermittent. This change was enabled by the presence of separate ICD-10-CM codes for these conditions.
- **NBDPN Birth Defect Case Definition Codes:** The appendix now includes an NBDPN Birth Defect Case Definition Code for each defect. These codes are to be used when reporting data to NBDPN to improve consistency in code assignments for the sake of data aggregation across birth defects surveillance programs. These codes are summarized on the following page, and also included with each birth defect description throughout Appendix 2A.

NBDPN Birth Defect Case Definition Codes

Birth Defect	NBDPN Case Definition Code
Anencephaly	1
Anophthalmia / Microphthalmia	6
Anotia / Microtia	9
Aortic valve stenosis	20
Atrial septal defect	15
Atrioventricular septal defect (Endocardial cushion defect)	16
Biliary atresia	33
Bladder exstrophy	35
Choanal atresia	28
Clef lip with cleft palate	69
Cleft lip alone (without cleft palate)	68
Cleft palate alone	26
Cloacal exstrophy	66
Clubfoot	64
Coarctation of the aorta	23
Common truncus (Truncus arteriosus or TA)	10
Congenital cataract	7
Congenital posterior urethral valves	65
Craniosynostosis	63
Deletion 22 q11	61
Diaphragmatic hernia	43
Double outlet right ventricle (DORV)	72
Ebstein anomaly	19
Encephalocele	4
Esophageal atresia	29
Gastroschisis	40
Holoprosencephaly	73
Hypoplastic left heart syndrome (HLHS)	21
Hypospadias	50
Interrupted aortic arch (IAA)	71
Limb deficiencies (reduction defect)	62
Omphalocele	41
Pulmonary valve atresia	75
Pulmonary valve stenosis	17
Rectal and large intestinal atresia / stenosis	30
Renal agenesis / hypoplasia	34
Single ventricle	70
Small intestinal atresia / stenosis	67
Spina bifida	2
Tetralogy of Fallot	12
Total anomalous pulmonary venous connection (TAPVC)	24
Transposition of the great arteries	11 – Transposition (TGA) 74 – Dextro-transposition (d-TGA)
Tricuspid valve atresia and stenosis	18
Trisomy 13	44
Trisomy 18	46
Trisomy 21 (Down syndrome)	45
Turner syndrome	60
Ventricular septal defect	14

Format for Birth Defect Descriptions

Defect Name *Defect Category*	
Description	Description of the defect, +/- illustration of the defect
NBDPN Data Management	
NBDPN Defect Tier	Defect Tier (Core or Enhanced)
NBDPN Birth Defect Case Definition Code	New codes for NBDPN reporting
CDC/BPA Codes	Applicable CDC/BPA codes for the defect
NBDPN Case Definition	
Tier 1 Surveillance	How to define the condition when performing Tier 1 surveillance (based on review and/or abstraction of medical record information)
Tier 2 Surveillance	How to define the condition when performing Tier 2 surveillance (based only on coded or written diagnosis data, without review and/or abstraction of medical record information)
Diagnoses included for case classification of defect	
Inclusions	Names, conditions, and diagnoses that should be included in the code for the defect.
ICD-10-CM Codes	Applicable ICD-10-CM codes for the defect
SNOMED CT Codes	Applicable SNOMED CT codes for the defect
Additional Information	
Exclusions	Names, conditions, and diagnoses that should not be included in the code for the defect.
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for the defect.
Additional Information	Other useful information.

Central Nervous System

Anencephaly

Anencephaly *Central Nervous System*	
Description	Congenital absence of a major portion of the brain, skull, and scalp. Cranial neural tissue is exposed, appearing as hemorrhagic fibrotic tissue, and is not covered by bone or skin. Cerebral hemispheres are not recognizable.  Terms such as holoanencephaly and meroanencephaly are used to distinguish the complete or near-complete absence of the brain (holoanencephaly) from partial absence (meroanencephaly). Anencephaly is considered a neural tube defect, a group of congenital anomalies resulting from the failure of the neural tube to close.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	1
CDC/BPA Codes	**740.00** Absence of brain **740.02** Anencephaly **740.03** Hemianencephaly **740.08** Other anomalies similar to anencephaly **740.10** Craniorachischisis
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal physical exam, imaging, surgery or autopsy documents anencephaly. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of anencephaly [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of anencephaly without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A

	Accepted: The following criterion must be met: • Presence of a coded or written diagnosis of anencephaly assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Anencephaly" section below.
Diagnoses Included for Case Classification of Anencephaly	
Inclusions	• Absent brain, with or without skull bones present • Anencephaly • Craniorachischisis – Anencephaly continuous with an open posterior spinal defect with no meninges covering the neural tissue • Exencephaly - Absence of cranial vault and overlying scalp with a large amount of protruding, disorganized brain tissue covered by a membrane
ICD-10-CM Codes	**Q00.0** Anencephaly **Q00.1** Craniorachischisis **O35.02X0–O35.02X9** Maternal care for (suspected) central nervous system malformation or damage in fetus, anencephaly **Anencephaly (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2111 **Fetal Anencephaly (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2113
SNOMED CT Codes	**Anencephaly (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2110 **Fetal Anencephaly (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2112
Additional Information	
Exclusions	• Encephalocele • Hydranencephaly • Iniencephaly • Rachischisis – When used alone, this term refers only to the spinal defect and should be coded as spina bifida without anencephaly
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential anencephaly: **ICD-10-CM:** **Q04.9** Congenital malformation of brain, unspecified **SNOMED-CT:** **253098009** Neural tube defect (disorder) **1208338004** Dysraphism, cleft lip and palate, limb reduction defect syndrome (disorder) **763527007** Distal monosomy 13q syndrome (disorder) **609416008** Fetal anencephaly suspected (situation) **359824007** Incomplete anencephaly (disorder) **95464003** Congenital absence of cranial vault (disorder)

Additional Information	Maternal serum alpha-fetoprotein (MSAFP) and/or amniotic fluid alpha-fetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) are typically elevated with anencephaly. However, these screening tests alone are not sufficient to diagnose the condition. In cases where both anencephaly and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephaly and spina bifida should be coded. Previously acrania was part of the Inclusions for this condition. Though the terms are sometimes used interchangeably, they are distinct conditions. Acrania is a rare malformation where the skull is partially or completely absent, but there is a normal or near-normal brain, typically covered only by a thin layer of membrane. This condition is now included in Case Ascertainment, as it requires further review to be classified as anencephaly.

Encephalocele

Encephalocele *Central Nervous System*	
Description	Herniation of brain tissue and/or meninges through a congenital defect in the skull. The hernia sac is usually covered by skin. Encephaloceles are classified by the location of the skull defect: occipital (back of the head), parietal (top-back of the head), frontal, or basal (including nasofrontal, frontoethmoidal, nasopharyngeal, and nasal-orbital).  In the United States, occipital encephaloceles are the most common form. Frontal and basal forms are more common in populations of Southeast Asian ancestry.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	4
CDC/BPA Codes	**742.00** Encephalocele, unspecified **742.01** Occipital encephalocele **742.02** Cervical encephalocele **742.03** Frontal encephalocele **742.04** Nasofrontal encephalocele **742.05** Parietal encephalocele **742.06** Orbital encephalocele **742.07** Nasopharyngeal encephalocele **742.09** Encephalocele, other
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal imaging (cranial ultrasound, CT, or MRI), surgery, or autopsy demonstrates herniation of brain tissue and/or meninges through a congenital skull defect. • An encephalocele is diagnosed based on postnatal physical examination and no subsequent imaging, surgery, or autopsy refutes the diagnosis. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of encephalocele [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of encephalocele without confirmatory exam, imaging, surgery, or autopsy information.

Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of encephalocele assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Encephalocele" section below.
Diagnoses Included for Case Classification of Encephalocele	
Inclusions	• Cephalocele • Cranial meningocele (herniation of meninges only, without brain tissue) • Encephalocystocele • Encephalocystomeningocele • Encephalomyelocele (herniation through a combined defect in the skull and upper cervical spine) • Hydrencephalocele / Hydroencephalocele • Meningoencephalocele • Ventriculocele • Arnold-Chiari malformation, type III
ICD-10-CM Codes	**Q01.0** Frontal encephalocele **Q01.1** Nasofrontal encephalocele **Q01.2** Occipital encephalocele **Q01.8** Encephalocele of other sites **Q01.9** Encephalocele, unspecified **O35.04X0 - O35.04X9** Maternal care for (suspected) central nervous system malformation or damage in fetus, encephalocele **Encephalocele (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2495 **Fetal Encephalocele (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2494
SNOMED CT Codes	**Encephalocele (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2493
Additional Information	
Exclusions	• Herniation of brain tissue through an acquired skull defect (trauma, surgery, or other postnatal cause) • Iniencephaly, open type — though an encephalocele component may be present, cases of open iniencephaly should not be counted under encephalocele • Cephalohematoma, caput succedaneum, or other scalp swellings without an underlying skull defect • Cranial dermal sinus without herniation of brain or meninges
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential encephalocele: **ICD-10-CM:** **Q61.9** Cystic kidney disease, unspecified **Q04.9** Congenital malformation of brain, unspecified

	SNOMED-CT: **29076005** Meckel-Gruber syndrome (disorder) **773307006** Zechi-Ceide syndrome (disorder) **719021005** DK phocomelia syndrome (disorder) – also called von Voss-Cherstvoy syndrome
Additional Information	Encephalocele is classified among the neural tube defects. Maternal serum alpha-fetoprotein (MSAFP), amniotic fluid alpha-fetoprotein (AFAFP), and amniotic fluid acetylcholinesterase (AChE) may be elevated when the lesion is not skin covered but are insufficient to establish a diagnosis. Most encephaloceles are skin-covered and do not produce elevated AFP. Many encephaloceles are recognizable on physical examination of the neonate (visible skin-covered mass at a cranial site). Definitive diagnosis, particularly for small, midline, or internal (basal, sphenoid, ethmoid, orbital, or pharyngeal) lesions, requires direct visualization of brain/meningeal herniation. Certain syndromes in which encephalocele is often but not always present include: Meckel-Gruber syndrome, Zechi-Ceide syndrome, DK phocomelia syndrome – also called von Voss-Cherstvoy syndrome. Per ICD-10-CM, Q01 excludes Meckel-Gruber syndrome. Cases with Meckel-Gruber will typically be coded under Q61.9 rather than Q01. Case ascertainment can include Q61.9 but to be confirmed the record should be reviewed for documented encephalocele.

Holoprosencephaly

Holoprosencephaly *Central Nervous System*	
Description	Congenital brain anomaly characterized by incomplete separation of the brain hemispheres, resulting in variable degrees of fusion of the forebrain. The spectrum of holoprosencephaly (HPE) includes alobar HPE, in which the failure of separation results in a single centrally located cerebral ventricle, semilobar HPE, in which there is partial forebrain cleavage, and lobar HPE, in which there is almost complete forebrain cleavage. A rare variant is the middle interhemispheric variant (MIHV), characterized by an abnormal midline union of the posterior frontal and parietal lobes. The more severe forms are often associated with facial anomalies, including a single eye (cyclopia), a tubular nose (cebocephaly), or a median cleft lip. Facial findings may be subtle and consist only of closely spaced eyes (hypotelorism), or of a single upper central incisor.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	73
CDC/BPA Codes	**742.26** Holoprosencephaly
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (CT or MRI scan), surgery, or autopsy demonstrates holoprosencephaly. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of holoprosencephaly [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of holoprosencephaly without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of holoprosencephaly assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Holoprosencephaly" section below.

Diagnoses Included for Case Classification of Holoprosencephaly	
Inclusions	• Alobar holoprosencephaly • Semilobar holoprosencephaly • Lobar holoprosencephaly • Middle interhemispheric variant (MIHV) • Syntelencephaly • Cyclopia • Cebocephaly • Ethmocephaly • Aprosencephaly • Atelencephaly
ICD-10-CM Codes	**Q04.2** Holoprosencephaly **O35.05X0–O35.05X9** Maternal care for (suspected) central nervous system malformation or damage in fetus, holoprosencephaly
SNOMED CT Codes	**30915001** Holoprosencephaly sequence (disorder) **55709000** Ethmocephalus (disorder) **204052006** Cebocephaly (disorder) **253136007** Lobar holoprosencephaly (disorder) **253137003** Alobar holoprosencephaly (disorder) **253138008** Semi-lobar holoprosencephaly (disorder) **890346002** Holoprosencephaly co-occurrent with congenital nasal pyriform aperture stenosis (disorder) **866053004** Middle interhemispheric variant of holoprosencephaly (disorder) **205798005** Cyclopia (disorder) **734001** Opocephalus (disorder) **37281006** Cyclops hypognathus (disorder) **95241002** Rhinocephaly (disorder) **1237366005** Aprosencephaly cerebellar dysgenesis (disorder)
Additional Information	
Exclusions	• Hydranencephaly • Porencephaly • Arhinencephaly without holoprosencephaly
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential holoprosencephaly: **ICD-10-CM:** **Q04.9** Congenital malformation of brain, unspecified **SNOMED-CT:** **30278004** Kundrat's syndrome (disorder) **715434005** Holoprosencephaly craniosynostosis syndrome (disorder) **716091000** Holoprosencephaly and postaxial polydactyly syndrome (disorder)

	716169009 Holoprosencephaly sequence with hypokinesia and congenital joint contracture syndrome (disorder) **716233007** Steinfeld syndrome (disorder) **722283003** Agnathia, holoprosencephaly, situs inversus syndrome (disorder) **766032007** Holoprosencephaly, ectrodactyly, cleft lip, cleft palate syndrome (disorder) **771146007** Holoprosencephaly with caudal dysgenesis syndrome (disorder) **1222660008** Pancreatic agenesis, holoprosencephaly syndrome (disorder) **1162839003** XK aprosencephaly syndrome (disorder) **763529005** Distal monosomy 7q36 syndrome (disorder) **763527007** Distal monosomy 13q syndrome (disorder)
Additional Information	As discussed holoprosencephaly, especially the alobar type, is commonly associated with facial anomalies, ranging from hypotelorism and single central upper incisor to median cleft lip (premaxillary agenesis) to cyclopia, a rare abnormality characterized by a single central eye in the low frontal area and a missing nose or a proboscis (a tubular shaped nose) located above the eye. When co-occurring, these defects should be coded separately. An important consideration in birth defect surveillance is that chromosomal abnormalities are identified in roughly one-quarter to one-half of individuals with holoprosencephaly (HPE). Trisomy 13 is consistently the predominant chromosomal finding, accounting for up to approximately 75% of HPE associated with chromosomal abnormalities, while triploidy and other aneuploidies such as trisomy 18 and 22 contribute a smaller but significant fraction.

Spina bifida

Spina bifida *Central Nervous System*	
Description	Incomplete closure of the spine (usually posteriorly) through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate. Spina bifida is considered a neural tube defect—one in a group of congenital anomalies resulting from the failure of the neural tube to close. 
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	2
CDC/BPA Codes	**741.00** Spina bifida aperta (open) with hydrocephalus - Any site **741.01** Spina bifida cystica (closed) with hydrocephalus and Arnold-Chiari malformation - Any site **741.02** Spina bifida cystica (closed) with stenosed aqueduct of Sylvius - Any site **741.03** Spina bifida cystica (closed) with unspecified hydrocephalus - Cervical **741.04** Spina bifida cystica (closed) with unspecified hydrocephalus - Thoracic **741.05** Spina bifida cystica (closed) with unspecified hydrocephalus - Lumbar **741.06** Spina bifida cystica (closed) with unspecified hydrocephalus - Sacral **741.07** Spina bifida with hydrocephalus of late onset - Any site **741.08** Other spina bifida with hydrocephalus - Specified site **741.085** Spina bifida cystica (closed) with unspecified hydrocephalus - Cervicothoracic **741.086** Spina bifida cystica (closed) with unspecified hydrocephalus - Thoracolumbar **741.087** Spina bifida cystica (closed) with unspecified hydrocephalus - Lumbosacral **741.09** Spina bifida, unspecified type, with hydrocephalus **741.90** Spina bifida aperta (open) without hydrocephalus - Any site **741.91** Spina bifida cystica (closed) without hydrocephalus - Cervical **741.92** Spina bifida cystica (closed) without hydrocephalus - Thoracic **741.93** Spina bifida cystica (closed) without hydrocephalus - Lumbar **741.94** Spina bifida cystica (closed) without hydrocephalus - Sacral **741.98** Other spina bifida without hydrocephalus - Specified site **741.985** Lipomyelomeningocele **741.99** Spina bifida without hydrocephalus - Unspecified site

NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal physical exam, imaging (CT or MRI scan), surgery or autopsy documents spina bifida. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of spina bifida [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of spina bifida without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** One of the following criteria must be met[1]: • Presence of more than one distinct coded or written diagnosis of spina bifida within an encounter, as described in the "Diagnoses Included for Case Classification of Spina Bifida" section below. • Presence of a coded or written diagnosis of spina bifida in more than one healthcare encounter, as described in the "Diagnoses Included for Case Classification of Spina Bifida" section below. **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of spina bifida assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Spina Bifida" section below, without the presence of confirmatory information.
Diagnoses Included for Case Classification of Spina Bifida	
Inclusions	• Lipomeningocele • Lipomyelomeningocele • Meningocele – Herniation of meninges only • Meningomyelocele, Myelomeningocele – Herniation of meninges and spinal cord tissue • Myelocystocele • Myeloschisis • Open spina bifida • Rachischisis – Open spina bifida without meninges covering the spinal cord tissue • Spina bifida aperta • Spina bifida cystica
ICD-10-CM Codes	**Q05.0** Cervical spina bifida with hydrocephalus **Q05.1** Thoracic spina bifida with hydrocephalus **Q05.2** Lumbar spina bifida with hydrocephalus **Q05.3** Sacral spina bifida with hydrocephalus

	Q05.4 Unspecified spina bifida with hydrocephalus **Q05.5** Cervical spina bifida without hydrocephalus **Q05.6** Thoracic spina bifida without hydrocephalus **Q05.7** Lumbar spina bifida without hydrocephalus **Q05.8** Sacral spina bifida without hydrocephalus **Q05.9** Spina bifida, unspecified **Q07.01** Arnold-Chiari syndrome with spina bifida **Q07.03** Arnold-Chiari syndrome with spina bifida and hydrocephalus **O35.08X0–O35.08X9** Maternal care for (suspected) central nervous system malformation or damage in fetus, spina bifida **Spina Bifida (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2123 **Fetal Spina Bifida (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2676
SNOMED CT Codes	**Spina Bifida (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2122 **Fetal Spina Bifida (SNOMED) OID**: 2.16.840.1.113762.1.4.1146.2121
Additional Information	
Exclusions	• Diastematomyelia • Diplomyelia • Hydromyelia • Spina bifida with coexisting anencephaly – Code only as anencephaly • Spina bifida occulta • Syringomyelia • Tethered spinal cord
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential spina bifida: **ICD-10-CM:** **Q07.02** Arnold-Chiari syndrome with hydrocephalus **Q03.0** Malformations of aqueduct of Sylvius **Q03.1** Atresia of foramina of Magendie and Luschka **Q03.8** Other congenital hydrocephalus **Q03.9** Congenital hydrocephalus, unspecified **Q07.9** Congenital malformation of nervous system, unspecified **SNOMED-CT:** **253098009** Neural tube defect (disorder) **237152002** Fetal spina bifida suspected (situation)
Additional Information	Open lesions (spina bifida aperta) have no skin covering, Neural tissue is exposed or covered only by meninges. They typically leak cerebrospinal fluid. Closed lesions (spina bifida cystica) are covered by skin, which may be normal or exhibit cutaneous markers (e.g., dimple, hairy patch, hemangioma, or subcutaneous lipoma).

	Coding Notes, open vs. closed lesions: • ICD-10 codes do not differentiate between open and closed lesions. • The CDC/BPA coding system uses "aperta" to designate open defects and "cystica" to designate closed defects. The broader clinical literature treats spina bifida aperta and cystica as synonyms. the CDC/BPA convention (cystica = closed) is an outlier relative to standard clinical usage. The specific type of defect (meningocele vs. myelomeningocele) may only be reliably distinguished by imaging (CT or MRI), at surgery, or at autopsy. Chiari II (Arnold-Chiari) malformation and hydrocephalus frequently, though not always, accompany myelomeningocele. When either is present in association with spina bifida, there is no need to code them separately. Note that Chiari I malformation is a distinct entity and should not be subsumed under a spina bifida code. Maternal serum alpha-fetoprotein (MSAFP), amniotic fluid alpha-fetoprotein (AFAFP), and amniotic fluid acetylcholinesterase (AChE) may be elevated in open spina bifida. AChE is more specific to open neural tube defects than AFP alone. However, these screening tests are not sufficient to diagnose the condition. In cases where both anencephaly and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephaly and spina bifida should be coded. If the coding system includes unique codes for different levels of spina bifida (cervical, thoracic, lumbar, sacral) and a defect involves more than one level (e.g., cervicothoracic, thoracolumbar, lumbosacral), the highest level should be coded. The highest level of involvement determines the degree of associated neurologic impairment. When the level is not documented, code as unspecified level.

[1] Salemi JL, Rutkowski RE, Tanner JP, Matas JL, Kirby RS. Identifying algorithms to improve the accuracy of unverified diagnosis codes for birth defects. Public Health Rep. 2018;133(3):303-310. doi:10.1177/0033354918763168.

Eye

Anophthalmia / Microphthalmia

Anophthalmia / Microphthalmia *Eye*	
Description	**Anophthalmia** – Total absence of eye tissue or apparent absence of the globe in an otherwise normal orbit. **Microphthalmia** – Reduced volume of the eye. The corneal diameter is usually less than 10 millimeters, or the anteroposterior globe diameter is less than 20 millimeters.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	6
CDC/BPA Codes	**743.000** Anophthalmos, laterality unknown **743.001** Anophthalmos, left eye **743.002** Anophthalmos, right eye **743.003** Anophthalmos, unilateral NOS **743.004** Anophthalmos, both eyes **743.100** Microphthalmos, laterality unknown **743.101** Microphthalmos, left eye **743.102** Microphthalmos, right eye **743.103** Microphthalmos, unilateral NOS **743.104** Microphthalmos, both eyes
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal physical exam by an ophthalmologist, imaging, surgery, or autopsy documents a complete absence or reduced volume (corneal diameter < 10 millimeters or anteroposterior globe diameter < 20 millimeters) of the eye. • Measurement of the anteroposterior diameter of the globe by ultrasound, CT or MRI scan, or at autopsy shows reduced volume (corneal diameter < 10 millimeters, or anteroposterior globe diameter < 20 millimeters) of the eye. **Accepted:** The following criterion must be met: • Postnatal diagnosis of anophthalmia or microphthalmia without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A

	Accepted: The following criterion must be met: • Presence of a coded or written diagnosis of anophthalmia or microphthalmia assigned postnatally, as described in the "Diagnoses Included for Case Classification of Anophthalmia / Microphthalmia" section below.
Diagnoses Included for Case Classification of Anophthalmia / Microphthalmia	
Inclusions	• Anophthalmia • Microphthalmia • Nanophthalmia – Microphthalmia with normal internal eye (intraocular) structures. This is a distinct genetic condition.
ICD-10-CM Codes	**Q11.0** Cystic eyeball **Q11.1** Other anophthalmos **Q11.2** Microphthalmos **Anophthalmia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2483 **Microphthalmia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2485
SNOMED CT Codes	**Anophthalmia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2482 **Microphthalmia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2484
Additional Information	
Exclusions	• Small eyes or small palpebral fissures for which the diagnosis of microphthalmia or anophthalmia has not been made • Microcornea with otherwise normal eye size • Prenatal finding of anophthalmia / microphthalmia without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential anophthalmia / microphthalmia: **ICD-10-CM:** **Q15.9** Congenital malformation of eye, unspecified **SNOMED-CT:** **720496006** Anophthalmia plus syndrome (disorder) **1217628008** Congenital absence of eye with orbital implant (disorder) **1303582008** Congenital aphakia, iris hypoplasia, microphthalmia, microcornea syndrome (disorder) **438504004** Lenz microphthalmia syndrome (disorder) **715533002** Microcephaly, microphthalmia, ectrodactyly of lower limbs and prognathism syndrome (disorder) **1003369001** Microphthalmos due to Delleman syndrome (disorder) **1003370000** Microphthalmos due to Fryns syndrome (disorder) **1003372008** Microphthalmos due to branchio-oculo-facial syndrome (disorder) **1230344000** Microphthalmia, microtia, fetal akinesia syndrome (disorder)

	1231626009 Syndromic nanophthalmos due to Kenny-Caffey syndrome (disorder) **1332382002** Coloboma, osteopetrosis, microphthalmia, macrocephaly, albinism, deafness syndrome (disorder) **717222003** Microphthalmia with ankyloblepharon and intellectual disability syndrome (disorder) **718761007** Syndromic microphthalmia due to orthodenticle homeobox 2 mutation (disorder) **720010009** Microphthalmia with brain atrophy syndrome (disorder) **721879006** Microphthalmia with linear skin defect syndrome (disorder) **723503006** Retinal degeneration, nanophthalmos, glaucoma syndrome (disorder) **771148008** X-linked colobomatous microphthalmia, microcephaly, intellectual disability, short stature syndrome (disorder) **773282001** Macrosomia, microphthalmia, cleft palate syndrome (disorder) **773628009** Frontonasal dysplasia, severe microphthalmia, severe facial clefting syndrome (disorder) **776204008** Colobomatous microphthalmia, obesity, hypogenitalism, intellectual disability syndrome (disorder) **778021002** Microphthalmia, retinitis pigmentosa, foveoschisis, optic disc drusen syndrome (disorder) **840492002** Microphthalmos co-occurrent with congenital ocular coloboma of bilateral eyes (disorder) **18821006** Dysplasia of eye (disorder)
Additional Information	Microphthalmia may occur in association with colobomas (gaps) in the uvea, iris, choroid and/or optic nerve (colobomatous microphthalmia). Anophthalmia and microphthalmia can be accompanied by malformations of the brain and face, and frequently are components of genetic syndromes.

Congenital cataract

Congenital cataract *Eye*	
Description	An opacity of the lens of the eye that has its origin prenatally.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	7
CDC/BPA Codes	**743.320** Cataract - NOS, laterality unknown **743.321** Cataract - NOS, left eye **743.322** Cataract - NOS, right eye **743.323** Cataract - NOS, unilateral NOS **743.324** Cataract - NOS, both eyes **743.325** Cataract - Anterior polar **743.326** Cataract - Other specified
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal physical exam by an ophthalmologist, surgery or autopsy documents a congenital cataract. **Accepted:** The following criterion must be met: • Postnatal diagnosis of congenital cataract without confirmatory exam, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of congenital cataract assigned postnatally, as described in the "Diagnoses Included for Case Classification of Congenital Cataract" section below.
Diagnoses Included for Case Classification of Congenital Cataract	
Inclusions	• Anterior polar cataract • Cataract, type not specified • Infantile cataract • Lamellar cataract • Nuclear cataract • Posterior lentiglobus/lenticonus cataract

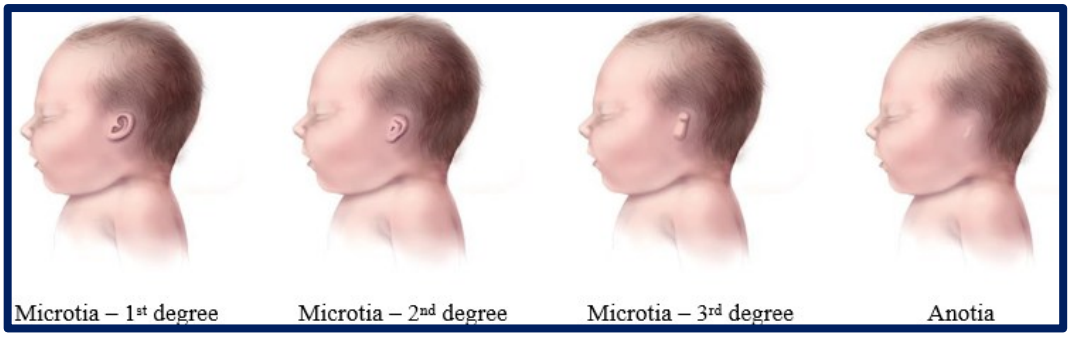
	• Posterior cortical cataract • Sectoral cataract • Zonular cataract
ICD-10-CM Codes	**Q12.0** Congenital cataract
SNOMED CT Codes	**9410001** Congenital cataract (disorder) **5361003** Embryonal nuclear cataract (disorder) **21590003** Congenital zonular cataract (disorder) **28550007** Congenital capsular cataract (disorder) **29590001** Congenital total cataract (disorder) **66499004** Congenital cortical cataract (disorder) **76562003** Congenital subcapsular cataract (disorder) **204127006** Cortical and zonular cataract (disorder) **204128001** Congenital lamellar cataract (disorder) **204138006** Congenital blue dot cataract (disorder) **204139003** Congenital membranous cataract (disorder) **253223002** Congenital polar cataract (disorder) **253224008** Congenital anterior polar cataract (disorder) **253225009** Congenital posterior polar cataract (disorder) **253226005** Congenital sutural cataract (disorder) **370483001** Mittendorf's dot (disorder) **890350009** Coralliform cataract (disorder) **1003424006** Hutterite type cataract (disorder) **1003884001** Pulverulent cataract (disorder) **1279837000** Congenital cataract microcornea with corneal opacity (disorder) **335831000119107** Congenital posterior subcapsular polar cataract of right eye (disorder) **335841000119103** Congenital nuclear cataract of right eye (disorder) **335851000119101** Congenital combined form cataract of right eye (disorder) **335861000119104** Congenital cataract of right eye (disorder) **335871000119105** Congenital anterior subcapsular polar cataract of right eye (disorder) **341441000119102** Congenital posterior subcapsular polar cataract of left eye (disorder) **341451000119100** Congenital nuclear cataract of left eye (disorder) **341461000119103** Congenital combined form cataract of left eye (disorder) **341471000119109** Congenital cataract of left eye (disorder) **341481000119107** Congenital anterior subcapsular polar cataract of left eye (disorder) **342821000119103** Congenital posterior subcapsular polar cataract (disorder) **342831000119100** Congenital combined form cataract (disorder) **342911000119104** Congenital anterior subcapsular polar cataract (disorder) **346691000119104** Congenital posterior subcapsular polar cataract of bilateral eyes (disorder) **346701000119104** Congenital nuclear cataract of bilateral eyes (disorder)

	346711000119101 Congenital combined form cataract of bilateral eyes (disorder) **346721000119108** Congenital cataract of bilateral eyes (disorder) **346731000119106** Congenital anterior subcapsular polar cataract of bilateral eyes (disorder) **348321000119108** Congenital zonular cataract of right eye (disorder) **348771000119108** Congenital zonular cataract of left eye (disorder) **349281000119109** Congenital zonular cataract of bilateral eyes (disorder) **679911000119104** Congenital cortical cataract of left eye (disorder) **679931000119109** Congenital cortical cataract of right eye (disorder) **681041000119106** Congenital cortical cataract of bilateral eyes (disorder) **15665801000119104** Congenital capsular cataract of bilateral eyes (disorder) **15665841000119102** Congenital capsular cataract of right eye (disorder) **15665881000119108** Congenital capsular cataract of left eye (disorder)
Additional Information	
Exclusions	• Any of the above types of cataract that has its origin after birth • Corneal opacities • Prenatal finding of congenital cataract without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential congenital cataract: **ICD-10-CM:** **Q12.9** Congenital lens malformation, unspecified **SNOMED-CT:** **15552004** Osteogenesis imperfecta, recessive perinatal lethal, with microcephaly AND cataracts (disorder) **79385002** Lowe syndrome (disorder) **80734006** Marinesco-Sjögren syndrome (disorder) **445257004** Nance-Horan syndrome (disorder) **715988005** Cataract with aberrant oral frenula and growth delay syndrome (disorder) **715989002** Congenital cataract with intellectual disability and anal atresia and urinary defect syndrome (disorder) **716099003** Absence deformity of leg and congenital cataract syndrome (disorder) **716170005** Deafness with cataract and skeletal anomaly syndrome (disorder) **717812000** Congenital cataract, hypertrophic cardiomyopathy, mitochondrial myopathy syndrome (disorder) **718766002** Spondyloepiphyseal dysplasia, craniosynostosis, cleft palate, cataract and intellectual disability syndrome (disorder) **718851007** Cataract glaucoma syndrome (disorder) **719102004** Congenital cataract with ataxia and deafness syndrome (disorder)

	721015008 Hydrocephalus with endocardial fibroelastosis and cataract syndrome (disorder) **721076000** Siegler Brewer Carey syndrome (disorder) **721233005** Hypergonadotropic hypogonadism with cataract syndrome (disorder) **722378009** Congenital cataract with deafness and hypogonadism syndrome (disorder) **722379001** Congenital cataract with hypertrichosis and intellectual disability syndrome (disorder) **722380003** Congenital cataract with intellectual disability and hypogonadotropic hypogonadism syndrome (disorder) **722381004** Congenital cataract, nephropathy, encephalopathy syndrome (disorder) **722382006** Cataract and microcornea syndrome (disorder) **723612001** Spinal muscular atrophy, Dandy-Walker malformation, cataract syndrome (disorder) **72670400** Cataract, congenital heart disease, neural tube defect syndrome (disorder) **726709001** Intellectual disability, cataract, calcified pinna, myopathy syndrome (disorder) **732952003** Congenital cataract ichthyosis syndrome (disorder) **772224009** Warburg micro syndrome (disorder) **77339800** Congenital cataract, progressive muscular hypotonia, hearing loss, developmental delay syndrome (disorder) **773627004** Porencephaly, microcephaly, bilateral congenital cataract syndrome (disorder) **773648002** Congenital cataract, hearing loss, severe developmental delay syndrome (disorder) **1172683008** Microcephaly, congenital cataract, psoriasiform dermatitis syndrome (disorder) **1255268002** Oculocerebrodental syndrome (disorder) **1260140008** Congenital cataract, severe neonatal hepatopathy, global developmental delay syndrome (disorder)
Additional Information	Cataracts may be congenital, acquired, or inherited. They may involve all or only part of the lens of either or both eyes. They may be an isolated finding in an otherwise normal eye or may be part of a more general eye malformation. They may be seen with metabolic disorders, such as galactosemia; genetic syndromes, such as chondrodysplasia punctata; chromosomal abnormalities, such as Trisomy 21; intrauterine infection, such as congenital rubella; or trauma. In some instances, the severity of the cataract progresses over time. The need for surgical treatment depends on the degree of visual impairment. When congenital cataract occurs with microphthalmia in the same infant, both conditions should be coded.

Ear

Anotia / Microtia

Anotia / Microtia *Ear*	
Description	Anotia – Total absence of the external ear and canal. Microtia – Small and abnormally shaped external ear (auricle, pinna). 
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	9
CDC/BPA Codes	**744.010** External ear - Absence, laterality unknown **744.011** External ear - Absence, left ear **744.012** External ear - Absence, right ear **744.013** External ear - Absence, unilateral NOS **744.014** External ear - Absence, both ears **744.210** Microtia, laterality unknown **744.211** Microtia, left ear **744.212** Microtia, right ear **744.213** Microtia, unilateral NOS **744.214** Microtia, both ears
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal physical exam, imaging, surgery, or autopsy documents complete absence, malformation, or hypoplasia of the external ear (auricle, pinna). **Accepted:** The following criterion must be met: • Postnatal diagnosis of anotia or microtia without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A

	Accepted: The following criterion must be met: • Presence of a coded or written diagnosis of anotia or microtia assigned postnatally, as described in the "Diagnoses Included for Case Classification of Anotia / Microtia" section below.
Diagnoses Included for Case Classification of Anotia / Microtia	
Inclusions	• Anotia • Microtia
ICD-10-CM Codes	**Q16.0** Congenital absence of (ear) auricle **Q17.2** Microtia **Anotia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2477 **Microtia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2478
SNOMED CT Codes	**Anotia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2480 **Microtia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2479
Additional Information	
Exclusions	• Small ears that retain most of the overall structure of the normal auricle, including lop or cup ear defects. In these, the auditory meatus is usually patent and defects of the ossicular chain of the middle ear are infrequent. • Type I Microtia • Isolated absence, atresia, stenosis or malformation of the ear canal without anotia or microtia • Prenatal finding of anotia or microtia without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential anotia or microtia: **ICD-10-CM:** **Q17.9** Congenital malformation of ear, unspecified **SNOMED-CT:** **702360007** Congenital deafness with labyrinthine aplasia, microtia and microdontia (disorder) **717909004** Bilateral microtia with deafness and cleft palate syndrome (disorder) **721836009** Hypertelorism with microtia and facial clefting syndrome (disorder) **722006004** Isotretinoin embryopathy-like syndrome (disorder) **724139004** Microtia, eye coloboma, imperforation of nasolacrimal duct syndrome (disorder) **732248005** Coxoauricular syndrome (disorder) **1230344000** Microphthalmia, microtia, fetal akinesia syndrome (disorder) **205418005** Goldenhar syndrome (disorder) **82203000** Treacher Collins syndrome (disorder) **254025006** Hemifacial microsomia (disorder) **726722009** Hemifacial microsomia with radial defect syndrome (disorder)

Additional Information	The spectrum of severity of microtia may range from a measurably small external ear with minimal structural abnormality to major structural alteration of the external ear with an absent or blind-ending canal. Following is the classification system of Meurman (modified from Marks): • Type I – Generally small ears that retain most of the overall structure of the normal auricle. These should not be coded as microtia for surveillance purposes • Type II – A moderately severe anomaly with a longitudinal mass of cartilage with some resemblance to a pinna. The rudimentary auricle may be hook-shaped, have an S-shape, or the appearance of a question mark. • Type III – The ear is a rudiment of soft tissue, and the auricle has no resemblance to a normal pinna. • Type IV – Complete absence of all external ear structures (anotia). Abnormalities that may be associated with anotia/microtia include anomalies of the middle and/or inner ear, the mandible and face, and hearing loss. Anotia/microtia is a component of many genetic and teratogenic syndromes but should be coded separately.

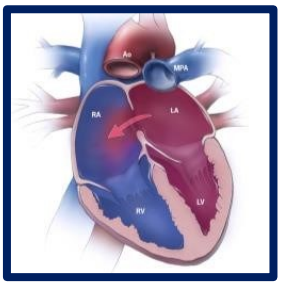
Cardiovascular

Aortic valve stenosis

Aortic valve stenosis *Cardiovascular*	
Description	Obstruction or narrowing of the aortic valve orifice, which obstructs blood flow from the left ventricle to the aorta.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	20
CDC/BPA Codes	**746.30** Aortic valve - Stenosis, congenital
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates valve morphology consistent with aortic valve stenosis. **Accepted:** The following criterion must be met: • Postnatal diagnosis of aortic valve stenosis without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of aortic valve stenosis assigned postnatally, as described in the "Diagnoses Included for Case Classification of Aortic Valve Stenosis" section below.
Diagnoses Included for Case Classification of Aortic Valve Stenosis	
Inclusions	• Stenosis of the aortic valve
ICD-10-CM Codes	**Q23.0** Congenital stenosis of aortic valve **Aortic valve stenosis (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2476
SNOMED CT Codes	**Aortic valve stenosis (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2475

Additional Information	
Exclusions	• Stenosis of the aorta without mention of the aortic valve • Supra-valvular or sub-valvular aortic stenosis
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential aortic valve stenosis: **ICD-10-CM:** **Q23.88** Other congenital malformations of aortic and mitral valves **Q23.9** Congenital malformation of aortic and mitral valves, unspecified **O35.BXX0 - O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **448743001** Abnormality of aortic valve (disorder) **194987006** Aortic valve stenosis with insufficiency (disorder) **427515002** Critical stenosis of aortic valve (disorder) **60573004** Aortic valve stenosis (disorder) **703236000** Non-rheumatic aortic valve stenosis with regurgitation (disorder) **836480008** Mild stenosis of aortic valve (disorder) **836481007** Moderate stenosis of aortic valve (disorder) **836482000** Severe stenosis of aortic valve (disorder) **472847005** Stenosis of fetal aortic valve (disorder)
Additional Information	While this condition may be identified by prenatal ultrasound, it should only be included as an accepted case without postnatal confirmation. The absence of aortic valve stenosis on prenatal ultrasound does not exclude postnatal diagnosis, as this condition may not be apparent prenatally or may progress after birth.

Atrial septal defect

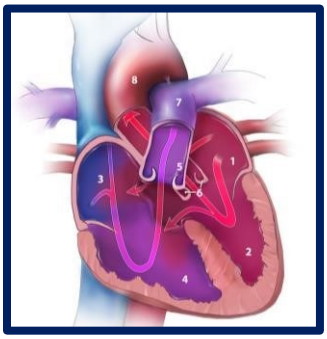
Atrial septal defect *Cardiovascular*	
Description	A persistent opening in the wall (septum) separating the left and right upper chambers (atria) of the heart that allows blood to flow between the atria after birth. Different anatomic types of atrial septal defects (ASD) are recognized. 
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	15
CDC/BPA Codes	**745.51** Atrial septal defect (ASD) - secundum **745.57** PFO vs. ASD **745.58** Atrial septal defect (ASD) - Other specified **745.59** Atrial septal defect (ASD) - NOS
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates cardiac wall opening consistent with atrial septal defect. **Accepted:** The following criterion must be met: • Postnatal diagnosis of atrial septal defect without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of atrial septal defect assigned postnatally, as described in the "Diagnoses Included for Case Classification of Atrial Septal Defect" section below.
Diagnoses Included for Case Classification of Atrial Septal Defect	
Inclusions	• Atrial septal defect (ASD), type not specified (NOS) • ASD other specified (OS) – which includes sinus venosus type

	• ASD secundum type (ASD 2 or ASD II) • ASD vs. PFO – In the first days of life, it may not be possible to distinguish whether the opening in the atrial septum is a true ASD or a patent foramen ovale that has not yet closed (see below). ASD vs. PFO should be included only if the exact nature of the condition was never resolved.
ICD-10-CM Codes	**Q21.10** Atrial septal defect unspecified **Q21.11** Secundum atrial septal defect **Q21.13** Coronary sinus atrial septal defect **Q21.14** Superior sinus venosus atrial septal defect **Q21.15** Inferior sinus venosus atrial septal defect **Q21.16** Sinus venosus atrial septal defect, unspecified **Q21.19** Other specified atrial septal defect **Atrial septal defect (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2487
SNOMED CT Codes	**Atrial septal defect (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2486
Additional Information	
Exclusions	• Atrioventricular septal defects (AVSD) • ASD primum type (1° ASD) – This is included under atrioventricular septal defects (see below) • Patent foramen ovale (PFO) – A PFO is normal in utero to allow blood to flow properly during fetal circulation. Functional closure typically occurs shortly after birth due to pressure changes, but anatomic closure may take weeks or months, and sometimes patency persists through adulthood • Prenatal finding of atrial septal defect without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential atrial septal defect: **ICD-10-CM:** **Q21.8** Other congenital malformations of cardiac septa **Q21.9** Congenital malformation of cardiac septum, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **204319006** Lutembacher's syndrome (disorder) **447665008** Interatrial communication through coronary sinus orifice (disorder) **447780005** Restrictive interatrial communication with obligatory shunt (disorder) **447933008** Nonfenestrated interatrial communication within oval fossa (disorder) **448575000** Fenestrated interatrial communication within oval fossa (disorder)

	721978002 Lymphedema, atrial septal defect, facial changes syndrome (disorder) **725145002** Atrial septal defect, atrioventricular conduction defect syndrome (disorder) **253273004** Cardiac septal defects (disorder) **17394001** Ebstein's anomaly with atrial septal defect (disorder)
Additional Information	ASDs are classified by anatomic location. Each type has a distinct embryologic origin, associated anomalies, and clinical significance: *Secundum ASD (ostium secundum defect):* The most common type, accounting for approximately 75–80% of all ASDs. The defect is located in the central atrial septum within the region of the fossa ovalis (the oval fossa). It is the only type that represents a true defect of the atrial septum proper. Secundum ASDs vary considerably in size; small defects may close spontaneously during infancy or early childhood, while larger ones typically do not. *Sinus venosus ASD:* Accounts for approximately 5–10% of ASDs. The defect is located outside the true atrial septum, at the junction of the right atrium with either the superior vena cava (superior type, more common) or the inferior vena cava (inferior type, less common). Sinus venosus defects are almost always associated with partial anomalous pulmonary venous return (PAPVR), in which one or more right-sided pulmonary veins drain anomalously into the right atrium or vena cava rather than the left atrium. *Coronary sinus ASD (unroofed coronary sinus):* A rare defect, accounting for less than 1% of ASDs. It results from a deficiency in the wall between the coronary sinus and the left atrium, allowing blood to shunt from left atrium to right atrium through the coronary sinus orifice. It is often associated with a persistent left superior vena cava draining to the coronary sinus. **Note the following conditions, excluded from this category:** *Patent foramen ovale (PFO):* The foramen ovale is a normal interatrial opening during fetal life that allows oxygenated blood from the placenta to bypass the fetal lungs. Functional closure normally occurs within hours of birth due to the reversal of interatrial pressure following the first breath. Anatomic fusion occurs later in infancy; in approximately 25% of adults, complete fusion never occurs, resulting in a persistent PFO. A PFO is a normal anatomic variant, not a congenital heart defect, and is excluded from surveillance case counts. In the first days of life it may not always be possible to distinguish a small secundum ASD from a PFO that has not yet closed; cases that remain unresolved should be classified as ASD vs. PFO and included (see Inclusions). **Primum ASD (ostium primum defect):** Accounts for approximately 15–20% of ASDs. The defect is located in the inferior atrial septum immediately adjacent to the atrioventricular valves and does not close spontaneously. It is part of the spectrum of atrioventricular septal defects (AVSD) and is etiologically distinct from secundum ASD. A cleft of the anterior (septal) leaflet of the mitral valve is typically present. Because of its relationship to AVSD, primum ASD is excluded from the ASD surveillance

	category and is classified under atrioventricular septal defects (see that section). A prenatal diagnosis of ASD requires postnatal confirmation before a case can be classified as confirmed. Prenatal identification of an opening in the atrial septum cannot reliably distinguish a true ASD from a physiologically normal foramen ovale, which occupies the same anatomic region and is open in every fetus. Apparent defects identified prenatally — particularly suspected secundum ASDs — may not be confirmed postnatally. Conversely, a normal prenatal ultrasound does not exclude postnatal ASD diagnosis. Complete visualization of the atrial septum is not reliably achievable prenatally, and postnatal echocardiography remains the diagnostic standard.

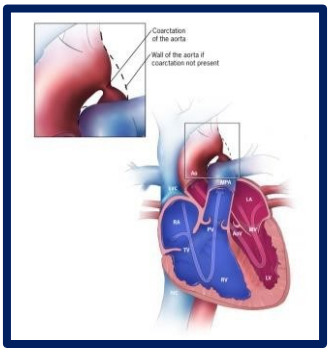
Atrioventricular septal defect (Endocardial cushion defect)

Atrioventricular septal defect (Endocardial cushion defect) *Cardiovascular*	
Description	 A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum. In extreme cases, virtually the entire atrial and ventricular septa may be missing. The tricuspid and mitral valves, which are the atrioventricular valves controlling blood flow from the atria to the ventricles, may also be abnormal. These valves may fail to form properly from the endocardial cushions during cardiac development and separate into two separate valves, forming instead of a single common atrioventricular valve. Consequently, communication between atria and ventricles can be a single large opening (canal) in the central part of the heart. A more limited form, the primum atrial septal defect, involves primarily the atrial septum with less extensive anomalies of the atrioventricular valves (so-called cleft mitral valve).
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	16
CDC/BPA Codes	**745.60** Ostium primum defects **745.61** Single common atrium **745.62** Common atrioventricular (AV) canal with ventricular septal defect (VSD) **745.63** Common atrioventricular (AV) canal, no mention of ventricular septal defect (VSD) **745.68** Endocardial cushion defect - Other specified **745.69** Endocardial cushion defect – NOS **745.487** Ventricular septal defect (VSD) – Inlet
NBDPN Case Definition	
Tier 1 Surveillance	**<u>Confirmed:</u>** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates cardiac wall opening consistent with atrioventricular septal defect described below. **<u>Accepted:</u>** One of the following criteria must be met: • A prenatal diagnosis of atrioventricular septal defect from a fetal echocardiogram [in a pregnancy that results in stillbirth,

	spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of atrioventricular septal defect without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of atrioventricular septal defect assigned postnatally, as described in the "Diagnoses Included for Case Classification of Atrioventricular Septal Defect (Endocardial Cushion Defect)" section below.
Diagnoses Included for Case Classification of Atrioventricular Septal Defect (Endocardial Cushion Defect)	
Inclusions	• Atrioventricular septal defect (AVSD) • Common or complete atrioventricular (AV) canal • Endocardial cushion defect • Primum type atrial septal defect (1° ASD) – A defect only in the lower portion of the atrial septum. While this does not involve a defect in the upper portion of the ventricular septum, it is etiologically related to the more complete form of AVSD. A cleft mitral valve is often present with a primum type ASD (see partial AVC). • Common atrium – Near absence of the atrial septum. • Partial AV canal (partial endocardial cushion defect) – Refers to a primum ASD with cleft mitral valve. • Inflow-type, subtricuspid, or canal-type ventricular septal defect ('inlet VSD' or 'VSD of the AVC type')– A defect in the upper (inflow) portion of the ventricular septum. While this does not also involve a defect in the lower portion of the atrial septum, it is etiologically related to the more complete form.
ICD-10-CM Codes	**Q21.20** Atrioventricular septal defect unspecified as to partial or complete **Q21.21** Partial atrioventricular septal defect **Q21.22** Transitional atrioventricular septal defect **Q21.23** Complete atrioventricular septal defect **Atrioventricular septal defect (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2333
SNOMED CT Codes	**Atrioventricular septal defect (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2332

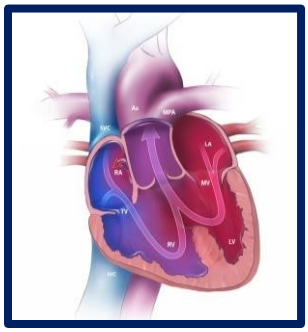
Additional Information	
Exclusions	• Secundum ASDs that coexist with a VSD. In this instance, both the ASD and the VSD should be coded.
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential atrioventricular septal defect: **ICD-10-CM:** **Q21.8** Other congenital malformations of cardiac septa **Q21.9** Congenital malformation of cardiac septum, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **59494005** Congenital septal defect of heart (disorder) **737156006** Congenital anomaly of atrioventricular septum (disorder) **763066009** Atrioventricular septal defect, blepharophimosis, radial and anal defect syndrome (disorder)
Additional Information	Atrioventricular septal defects are frequently seen in Down syndrome. Approximately half of all children with Down syndrome have a CHD, and about 20% have an atrioventricular septal defect. Conversely, approximately 60% of children with an atrioventricular septal defect have Down syndrome.

Coarctation of the aorta

Coarctation of the aorta *Cardiovascular*	
Description	Narrowing of the aorta, most commonly at the aortic isthmus in the region of the ductus arteriosus, which obstructs blood flow from the heart to the rest of the body. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	23
CDC/BPA Codes	**747.10** Coarctation of aorta - Preductal (proximal to ductus arteriosus) **747.11** Coarctation of aorta - Postductal (distal to ductus arteriosus) **747.12** Coarctation of aorta - Ductal (in the area of the ductus arteriosus) **747.19** Coarctation of aorta – Unspecified
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with coarctation of the aorta. **Accepted:** The following criterion must be met: • Postnatal diagnosis of coarctation of the aorta without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of coarctation of the aorta assigned postnatally, as described in the "Diagnoses Included for Case Classification of Coarctation of the Aorta" section below.

Diagnoses Included for Case Classification of Coarctation of the Aorta	
Inclusions	• Coarctation of the aorta, type not specified • Preductal, juxtaductal, and postductal coarctations – These terms refer to the exact placement of the segment of coarctation relative to the insertion of the ductus arteriosus
ICD-10-CM Codes	**Q25.1** Coarctation of aorta **Coarctation of the Aorta (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2314
SNOMED CT Codes	**Coarctation of the Aorta (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2313
Additional Information	
Exclusions	• Complete loss of luminal continuity at the aortic isthmus in the region of the ductus arteriosus (this is classified as interrupted aortic arch Type A rather than coarctation) • Prenatal finding of coarctation of aorta without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential coarctation of aorta: **ICD-10-CM:** **Q25.8** Other congenital malformations of other great arteries **Q25.9** Congenital malformation of great arteries, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **41371000119100** Shone complex (disorder) **450304006** Coarctation of suprarenal abdominal aorta (disorder) **450305007** Coarctation of infrarenal abdominal aorta (disorder) **449271003** Residual coarctation of aorta (disorder) **59877000** Congenital anomaly of aorta (disorder)
Additional Information	Coarctation of the aorta is associated with Turner syndrome (45,X and variant karyotypes). Female infants diagnosed with coarctation who have not had chromosomal evaluation should be considered for karyotype testing. While coarctation of the aorta may be suspected by prenatal ultrasound, false positives do occur. It should not be included as a confirmed case without postnatal confirmation. Conversely, coarctation of the aorta is often missed on prenatal ultrasound (high false-negative rate).

Common truncus (Truncus arteriosus or TA)

Common truncus (Truncus arteriosus or TA) *Cardiovascular*	
Description	Failure of separation of the aorta and the pulmonary artery during development, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	10
CDC/BPA Codes	**745.00** Persistent truncus arteriosus
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with common truncus. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of common truncus from a fetal echocardiogram [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of common truncus without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of common truncus assigned postnatally, as described in the "Diagnoses Included for Case Classification of Common Truncus (Truncus Arteriosus or TA)" section below.

Diagnoses Included for Case Classification of Common Truncus (Truncus Arteriosus or TA)	
Inclusions	• Common truncus • Truncus arteriosus (TA) • Persistent truncus arteriosus
ICD-10-CM Codes	**Q20.0** Common arterial trunk **Truncus Arteriosus (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2331
SNOMED CT Codes	**Truncus Arteriosus (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2330
Additional Information	
Exclusions	• Aorto-pulmonary window- a hole (the "window") between a separate aorta and pulmonary artery. This is distinct from truncus arteriosus, in which the common arterial trunk fails to divide into separate aortic and pulmonary vessels during development
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential common truncus: **ICD-10-CM:** **Q20.8** Other congenital malformations of cardiac chambers and connections **Q20.9** Congenital malformation of cardiac chambers and connections, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **234122004** Persistence of primitive artery (disorder) **253302006** Single outlet ventriculoarterial connection (disorder) **1230344000** Microphthalmia, microtia, fetal akinesia syndrome (disorder) **762253006** Common arterial trunk with pulmonary dominance co-occurrent with interrupted aortic arch (disorder)
Additional Information	A ventricular septal defect is nearly always present in association with truncus defects and should be coded separately. Truncus arteriosus is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Approximately 25–35% of infants with truncus arteriosus have a deletion of chromosome 22q11.2. This deletion is reliably diagnosed by fluorescent in situ hybridization (FISH) or microarray technology but can be missed by routine karyotype analysis. Edwards' Type IV (pulmonary atresia with MAPCAs) is no longer classified as truncus arteriosus; cases with this code are excluded or require individual chart review.

Double outlet right ventricle (DORV)

Double outlet right ventricle (DORV) *Cardiovascular*	
Description	Both the pulmonary artery and the aorta arise from the right ventricle, usually accompanied by a ventricular septal defect (VSD). DORV subtypes are usually distinguished by the great artery anatomic relationship: DORV with normally related great arteries and DORV with "transposed" or malposed or side-by-side great arteries. Note that in TGA (transposition of the great arteries), the aorta arises from the right ventricle and the pulmonary artery from the left ventricle. DORV differs because both great arteries arise from the right ventricle.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	72
CDC/BPA Codes	**745.13** Double outlet right ventricle (DORV) - normally related great vessels **745.14** Double outlet right ventricle (DORV) - transposed great vessels **745.15** Double outlet right ventricle (DORV) - relationship of great vessels not specified
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with double outlet right ventricle. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of double outlet right ventricle from a fetal echocardiogram [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of double outlet right ventricle without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of double outlet right ventricle assigned postnatally, as described in the "Diagnoses

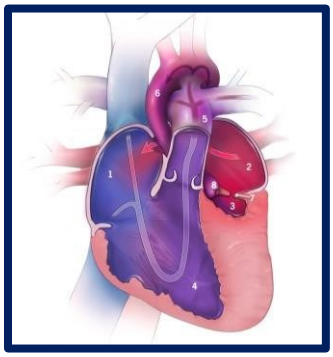
	Included for Case Classification of Double Outlet Right Ventricle" section below.
Diagnoses Included for Case Classification of Double Outlet Right Ventricle	
Inclusions	• Double outlet right ventricle (DORV) with normally related great vessels • DORV with transposed great vessels • DORV with unknown relationship of great vessels • Taussig-Bing syndrome (anomaly) • If a case has separate codes for DORV and TGA, include the case in the DORV category only and not in the TGA category.
ICD-10-CM Codes	**Q20.1** Double outlet right ventricle **Double Outlet Right Ventricle (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2323
SNOMED CT Codes	**Double Outlet Right Ventricle (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2322
Additional Information	
Exclusions	N/A
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential double outlet right ventricle: **ICD-10-CM:** **Q20.8** Other congenital malformations of cardiac chambers and connections **Q20.9** Congenital malformation of cardiac chambers and connections, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **871668002** Congenital right ventricular anomaly (disorder) **871599009** Double outlet right ventricle with subaortic or doubly committed ventricular septal defect and pulmonary stenosis Fallot type (disorder) **871601006** Double outlet right ventricle with subaortic ventricular septal defect and pulmonary stenosis Fallot type (disorder)
Additional Information	In previous surveillance guidelines, all DORV was included in the TGA category. DORV now has its own separate surveillance category; all subtypes should be coded as DORV. When DORV co-occurs with ventricular septal defect (VSD), the VSD and DORV should be coded separately.

Ebstein anomaly

Ebstein anomaly *Cardiovascular*	
Description	Abnormal formation and downward displacement of the tricuspid valve into the right ventricle. The tricuspid valve is typically hypoplastic and regurgitant, causing an enlarged right atrium and a small right ventricle. There may be associated functional pulmonary stenosis due to severely impaired right ventricular output from the small functional right ventricle.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	19
CDC/BPA Codes	**746.20** Ebstein's anomaly
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with Ebstein anomaly. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of Ebstein anomaly from a fetal echocardiogram [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of Ebstein anomaly without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of Ebstein anomaly assigned postnatally, as described in the "Diagnoses Included for Case Classification of Ebstein Anomaly" section below.

Diagnoses Included for Case Classification of Ebstein Anomaly	
Inclusions	• Ebstein's anomaly • Ebstein malformation
ICD-10-CM Codes	**Q22.5** Ebstein's anomaly **Ebstein Anomaly (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2316
SNOMED CT Codes	**Ebstein Anomaly (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2315
Additional Information	
Exclusions	N/A
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Ebstein anomaly: **ICD-10-CM:** **Q22.8** Other congenital malformations of tricuspid valve **Q22.9** Congenital malformation of tricuspid valve, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **871668002** Congenital right ventricular anomaly (disorder) **447830005** Congenital abnormality of tricuspid leaflet (disorder)
Additional Information	Although Ebstein's anomaly has been associated with lithium exposure during gestation, the magnitude of this association is likely small.

Hypoplastic left heart syndrome (HLHS)

Hypoplastic left heart syndrome (HLHS) *Cardiovascular*	
Description	A condition in which the structures on the left side of the heart and the aorta are extremely small, insufficient to support systemic circulation. In most cases, the great arteries are normally related. Typically, in this condition the severe hypoplasia of the left ventricle is associated with atresia or severe hypoplasia of both the mitral and aortic valves, and often with hypoplasia of the aortic arch and coarctation of the aorta. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	21
CDC/BPA Codes	**746.70** Hypoplastic left heart syndrome (HLHS)
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with hypoplastic left heart syndrome. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of hypoplastic left heart syndrome from a fetal echocardiogram [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of hypoplastic left heart syndrome without confirmatory imaging, surgery, or autopsy information.

Tier 2 Surveillance	**Confirmed:** One of the following criteria must be met[1]: • Presence of more than one distinct coded or written diagnosis of hypoplastic left heart syndrome within an encounter, as described in the "Diagnoses Included for Case Classification of Hypoplastic Left Heart Syndrome" section below. • Presence of a coded or written diagnosis of hypoplastic left heart syndrome in more than one healthcare encounter, as described in the "Diagnoses Included for Case Classification of Hypoplastic Left Heart Syndrome" section below. **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of hypoplastic left heart syndrome assigned postnatally, as described in the "Diagnoses Included for Case Classification of Hypoplastic Left Heart Syndrome" section below, without the presence of confirmatory information.
Diagnoses Included for Case Classification of Hypoplastic Left Heart Syndrome	
Inclusions	• Any diagnosis of hypoplastic left heart syndrome (HLHS), regardless of whether all components are present.
ICD-10-CM Codes	**Q23.4** Hypoplastic left heart syndrome **Hypoplastic left heart syndrome (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2325
SNOMED CT Codes	**62067003** Hypoplastic left heart syndrome (disorder) **Hypoplastic left heart syndrome (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2324
Additional Information	
Exclusions	• Hypoplasia or diminished size of the left ventricle alone without involvement of other structures on the left side of the heart or the aorta.
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential hypoplastic left heart syndrome: **ICD-10-CM:** **Q23.88** Other congenital malformations of aortic and mitral valves **Q23.9** Congenital malformation of aortic and mitral valves, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **93262004** Congenital hypoplasia of heart (disorder)

Additional Information	Hypoplastic left heart syndrome is among the congenital heart defects reported in association with Turner syndrome (45,X and variant karyotypes); genetic evaluation should be sought and documented when available. HLHS is one of the congenital heart defects most reliably identified by prenatal fetal echocardiography, due to the pronounced asymmetry of the cardiac chambers visible on the four-chamber view. A prenatal diagnosis by a pediatric cardiologist is therefore accepted as sufficient for confirmed case classification (see above). In pregnancies ending in stillbirth, spontaneous loss, or elective termination where no postnatal evaluation is available, a prenatal diagnosis by a pediatric cardiologist is sufficient for Confirmed classification; a prenatal diagnosis without a pediatric cardiologist interpreter should be classified as Accepted.

[1] Salemi JL, Rutkowski RE, Tanner JP, Matas JL, Kirby RS. Identifying algorithms to improve the accuracy of unverified diagnosis codes for birth defects. Public Health Rep. 2018;133(3):303-310. doi:10.1177/0033354918763168.

Interrupted aortic arch (IAA)

Interrupted aortic arch (IAA) *Cardiovascular*	
Description	Complete loss of communication (interruption) between the ascending and descending aorta, usually associated with a malalignment-type ventricular septal defect (VSD). Types of Interrupted Aortic Arch (IAA) are defined by where the interruption occurs along the arch from the conotruncus to the descending aorta. Type A interruption occurs distal to the left subclavian artery (between the left subclavian artery and the descending aorta). Type B occurs between the left common carotid and left subclavian arteries, is considered a conotruncal heart defect, and is the more common form of interrupted aortic arch. Type C occurs between the brachiocephalic innominate and left common carotid arteries.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	71
CDC/BPA Codes	**747.215** Interrupted aortic arch - Type A **747.216** Interrupted aortic arch - Type B **747.217** Interrupted aortic arch - Type C **747.285** Interrupted aortic arch, NOS
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with interrupted aortic arch. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of interrupted aortic arch from a fetal echocardiogram [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of interrupted aortic arch without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met:

	• Presence of a coded or written diagnosis of Interrupted aortic arch assigned postnatally, as described in the "Diagnoses Included for Case Classification of Interrupted Aortic Arch" section below.
Diagnoses Included for Case Classification of Interrupted Aortic Arch	
Inclusions	• IAA types A, B or C, or all IAA if type unknown or not otherwise specified (NOS).
ICD-10-CM Codes	**Q25.21** Interruption of aortic arch **Interrupted Aortic Arch (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2312
SNOMED CT Codes	**Interrupted Aortic Arch (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2311
Additional Information	
Exclusions	N/A
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential interrupted aortic arch: **ICD-10-CM:** **Q25.29** Other atresia of aorta **Q25.8** Other congenital malformations of other great arteries **Q25.9** Congenital malformation of great arteries, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **79439001** Congenital anomaly of aortic arch (disorder) **762253006** Common arterial trunk with pulmonary dominance co-occurrent with interrupted aortic arch (disorder)
Additional Information	IAA, especially type B, has the highest 22q11.2 deletion rate of all conotruncal defects, approximately 50%. The types of IAA correspond to SNOMED codes as the following: • Type A - 253681005 Interrupted aortic arch distal to left subclavian artery (disorder) • Type B - 253682003 Interrupted aortic arch between left subclavian and left common carotid artery (disorder) • Type C - 253683008 Interrupted aortic arch between left common carotid and brachiocephalic artery (disorder)

Pulmonary valve atresia

Pulmonary valve atresia *Cardiovascular*	
Description	Pulmonary valve atresia – Lack of patency, or failure of formation altogether, of the pulmonary valve, resulting in complete obstruction of blood flow from the right ventricle to the pulmonary artery. This group includes pulmonary atresia with intact ventricular septum. For pulmonary atresia with ventricular septal defect, see section on Tetralogy of Fallot.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	75
CDC/BPA Codes	**746.00** Pulmonary valve - Atresia or hypoplasia
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with pulmonary valve atresia. **Accepted:** The following criterion must be met: • Postnatal diagnosis of pulmonary valve atresia without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of pulmonary valve atresia assigned postnatally, as described in the "Diagnoses Included for Case Classification of Pulmonary Valve Atresia" section below.
Diagnoses Included for Case Classification of Pulmonary Valve Atresia	
Inclusions	• Pulmonary valve atresia with intact ventricular septum
ICD-10-CM Codes	**Q22.0** Pulmonary valve atresia **Pulmonary valve atresia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2318

SNOMED CT Codes	**Pulmonary valve atresia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2317
Additional Information	
Exclusions	• Atresia or stenosis of the main or branch (right or left) pulmonary arteries, not involving the pulmonary valve • Pulmonary stenosis that occurs as part of Tetralogy or Pentalogy of Fallot • Supra-valvular or sub-valvular pulmonic stenosis • Pulmonary valve stenosis (PS) (most cases of PS) Pulmonic stenosis (PS)
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential pulmonary valve atresia and stenosis: **ICD-10-CM:** **Q22.3** Other congenital malformations of pulmonary valve **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **123656005** Congenital atresia of cardiac valve (disorder) **204339005** Congenital pulmonary valve abnormality (disorder) **719955006** Atresia of pulmonary valve (disorder) **448643005** Abnormality of pulmonary valve (disorder) **236529001** Prune belly syndrome with pulmonic stenosis, intellectual disability and deafness (disorder)
Additional Information	These defects have important physiological and coding differences among systems as seen here in the table, which is also discussed in the Tetralogy of Fallot section. **CCHD** / **ICD-10** / **CDC/BPA** PA, IVS / Q22.0 / 746.00 PA, VSD (TOF) / Q21.3 / 747.31 TOF / Q21.3 / 745.20 - 21 Pulmonary valve atresia (PA) may occur with or without a coexisting ventricular septal defect (VSD). For pulmonary valve atresia without a VSD (intact ventricular septum (IVS)), the CDC/BPA code 746.00 (“Pulmonary valve - Atresia or hypoplasia”) is used, corresponding to the ICD-10-CM code Q22.0. Pulmonary atresia with a VSD is classified under Tetralogy of Fallot for surveillance purposes and is coded as Q21.3 in ICD-10-CM. Pulmonary atresia with a VSD is similar to severe forms of Tetralogy of Fallot (TOF), and is included in Tetralogy of Fallot for surveillance (see below). There is not an applicable code depicting valvular pulmonary atresia with VSD; hence in CDC/BPA the code 747.31 (“Pulmonary artery - Atresia with septal defect”) is used.

	While these conditions may be identified by prenatal ultrasound, they should not be included as confirmed cases without postnatal confirmation. Pulmonary atresia with intact ventricular septum (PA/IVS) is more likely to be detected prenatally than pulmonary valve stenosis (PVS), because PA/IVS produces more pronounced right heart changes visible on the standard four-chamber view. Mild and moderate PVS in particular may produce a normal-appearing four-chamber view on routine obstetric screening and may not be diagnosed until after delivery. The absence of either diagnosis on prenatal ultrasound does not exclude postnatal diagnosis.

Pulmonary valve stenosis

Pulmonary valve stenosis *Cardiovascular*	
Description	Pulmonary valve stenosis – Obstruction or narrowing of the pulmonary valve, which impairs blood flow from the right ventricle to the pulmonary artery. Clinically, pulmonary valve stenosis can vary from critical (functionally similar to atresia and requiring prompt intervention), to relatively mild.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	17
CDC/BPA Codes	**746.01** Pulmonary valve - Stenosis, congenital
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with pulmonary valve stenosis. **Accepted:** The following criterion must be met: • Postnatal diagnosis of pulmonary valve stenosis without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of pulmonary valve stenosis assigned postnatally, as described in the "Diagnoses Included for Case Classification of Pulmonary Valve Stenosis" section below.
Diagnoses Included for Case Classification of Pulmonary Valve Stenosis	
Inclusions	• Pulmonary valve stenosis (PS) (most cases of PS) Pulmonic stenosis (PS)
ICD-10-CM Codes	**Q22.1** Congenital pulmonary valve stenosis

	Pulmonary valve stenosis (ICD10CM) OID: 2.16.840.1.113762.1.4.1146.2320
SNOMED CT Codes	**Pulmonary valve stenosis (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2319
Additional Information	
Exclusions	• Atresia or stenosis of the main or branch (right or left) pulmonary arteries, not involving the pulmonary valve • Pulmonary stenosis that occurs as part of Tetralogy or Pentalogy of Fallot • Supra-valvular or sub-valvular pulmonic stenosis • Pulmonary valve atresia alone
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential pulmonary valve atresia and stenosis: **ICD-10-CM:** **Q22.3** Other congenital malformations of pulmonary valve **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **204339005** Congenital pulmonary valve abnormality (disorder) **56786000** Pulmonic valve stenosis (disorder) **448643005** Abnormality of pulmonary valve (disorder) **236529001** Prune belly syndrome with pulmonic stenosis, intellectual disability and deafness (disorder) **236530006** Pulmonic stenosis and congenital nephrosis (disorder) **472845002** Stenosis of fetal pulmonary valve (disorder)
Additional Information	Pulmonary valve stenosis (PVS) may occur with or without a coexisting ventricular septal defect (VSD). In CDC/BPA, 746.01 refers to pulmonary valve stenosis, corresponding to the ICD-10-CM code Q22.1. While this condition may be identified by prenatal ultrasound, it should not be included as a confirmed case without postnatal confirmation. Pulmonary atresia with intact ventricular septum (PA/IVS) is more likely to be detected prenatally than pulmonary valve stenosis (PVS), because PA/IVS produces more pronounced right heart changes visible on the standard four-chamber view. Mild and moderate PVS in particular may produce a normal-appearing four-chamber view on routine obstetric screening and may not be diagnosed until after delivery. The absence of either diagnosis on prenatal ultrasound does not exclude postnatal diagnosis.

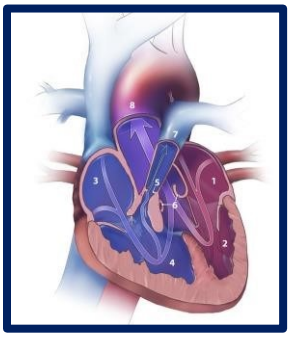
Single ventricle

Single ventricle *Cardiovascular*	
Description	A congenital heart defect in which only one morphologically distinct ventricle receives blood from both atria. This ventricle also serves as the dominant pumping chamber. The most common anatomic form is double-inlet left ventricle. Single ventricle is a complex lesion with multiple associated cardiac anomalies.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	70
CDC/BPA Codes	**745.30** Single ventricle / common ventricle, unspecified **745.31** Double-inlet left ventricle **745.32** Double-inlet right ventricle **745.39** Other specified single ventricle
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrate a univentricular anatomy consistent with single ventricle. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of single ventricle from a fetal echocardiogram [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of single ventricle without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of single ventricle assigned postnatally, as described in the "Diagnoses Included for Case Classification of Single Ventricle" section below.

Diagnoses Included for Case Classification of Single Ventricle	
Inclusions	Single (common) ventricle **without** a more specific diagnosis of hypoplastic ventricle or atrioventricular (AV) valve atresia. Included anatomic forms: • Double-inlet left ventricle (most common) • Double-inlet right ventricle • Double inlet to a ventricle of indeterminate morphology • Common ventricle / univentricular heart (no specified morphology) • Other specified forms of single ventricle
ICD-10-CM Codes	**Q20.4** Double inlet ventricle **Single Ventricle (Disorders) (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2327
SNOMED CT Codes	**Single Ventricle (Disorders) (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2326
Additional Information	
Exclusions	The following conditions produce single-ventricle physiology but are ascertained under a more specific defect and are excluded from this category: • Hypoplastic left heart syndrome (ascertained separately as HLHS) • Tricuspid atresia (ascertained separately) • Mitral atresia with normal aortic valve (when ascertained under its specific code) • Complete atrioventricular canal (AVSD) with severe malalignment of the AV valves to one side producing single-ventricle physiology (ascertained under AVSD) • Severe forms of double outlet right ventricle with a single functional ventricle (ascertained under DORV) • "Functional" single ventricle in a heart with two anatomic ventricles where one is very small due to AV valve atresia (ascertained under the primary defect)
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential single ventricle: **ICD-10-CM:** **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **443379009** Functional single ventricle SNOMED CT codes relating to associated AV-valve abnormalities in double-inlet ventricle (e.g., 253483002, 253486005, 253487001, 253489003, 253492004, 253493009, 253494003, 253495002, 253497005, 253498000, 253499008, 253500004, 253501000, 253502007, 253503002, 253504008, 253505009, 449040002, 449098005, 449099002, 449228007,

	460614005, 871583003, 871585005) describe co-occurring findings and should not be used as stand-alone case-finding codes for single ventricle.
Additional Information	Single ventricle anatomy is difficult to code because these hearts often carry multiple overlapping descriptors. Currently, imaging and other diagnostic methods can generally provide sufficient detail to define the primary ventricular morphology. Programs should record associated cardiac anomalies separately. Frequently co-occurring defects include transposed or malposed great arteries, pulmonary stenosis or atresia, coarctation of the aorta, interrupted aortic arch, and rudimentary outlet chambers. Pulse oximetry screening, cyanosis, or a cardiac murmur may raise suspicion but is not sufficient without imaging confirmation. Single ventricle is one of the 12 critical congenital heart defects (CCHDs) targeted by newborn pulse oximetry screening. In this document, "single ventricle" refers strictly to the anatomic univentricular heart. It must be distinguished from the broader clinical concept of a "functional single ventricle" (or univentricular physiology), which is an umbrella term for any heart that relies on one functional pumping chamber due to the severe underdevelopment of the other (e.g., Hypoplastic Left Heart Syndrome, Tricuspid Atresia, or severe unbalanced AV canal). Lesions resulting in a "functional" single ventricle should be excluded from this category and ascertained under their specific anatomic diagnoses.

Tetralogy of Fallot

Tetralogy of Fallot *Cardiovascular*	
Description	The simultaneous presence of a ventricular septal defect (VSD), right ventricular outflow tract obstruction (pulmonary and subpulmonary stenosis), a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	12
CDC/BPA Codes	**745.20** Tetralogy of Fallot (TOF) **745.21** Pentalogy of Fallot **747.31** Pulmonary artery - Atresia with septal defect
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates morphology consistent with Tetralogy of Fallot. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of Tetralogy of Fallot from a fetal echocardiogram [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of Tetralogy of Fallot without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** One of the following criteria must be met[1]: • Presence of more than one distinct coded or written diagnosis of Tetralogy of Fallot within an encounter, as described in the "Diagnoses Included for Case Classification of Tetralogy of Fallot" section below. • Presence of a coded or written diagnosis of Tetralogy of Fallot in more than one healthcare encounter, as described in the "Diagnoses

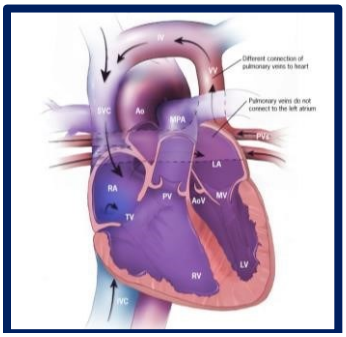
	Included for Case Classification of Tetralogy of Fallot" section below. **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of Tetralogy of Fallot assigned postnatally, as described in the "Diagnoses Included for Case Classification of Tetralogy of Fallot" section below, without the presence of confirmatory information.
Diagnoses Included for Case Classification of Tetralogy of Fallot	
Inclusions	• Pentalogy of Fallot – tetralogy of Fallot (TOF) with an associated inter-atrial communication, either a patent foramen ovale (PFO) or an atrial septal defect (ASD). • Tetralogy of Fallot (TOF) • Pulmonary atresia with VSD (see additional information below)
ICD-10-CM Codes	**Q21.3** Tetralogy of Fallot **Tetralogy of Fallot (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2329
SNOMED CT Codes	**Tetralogy of Fallot (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2328
Additional Information	
Exclusions	• Simultaneous occurrence of a VSD and pulmonary stenosis that has not been specifically diagnosed as Tetralogy of Fallot by the treating clinician. • Trilogy of Fallot, or Fallot's Triad – the simultaneous presence of an atrial septal defect, pulmonic stenosis, and right ventricular hypertrophy.
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Tetralogy of Fallot: **ICD-10-CM:** **Q21.8** Other congenital malformations of cardiac septa **Q21.9** Congenital malformation of cardiac septum, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **723336008** Fallot complex with intellectual disability and growth delay syndrome (disorder) **766976003** Pulmonary valve agenesis, tetralogy of Fallot, absence of ductus arteriosus syndrome (disorder) **253511007** Congenital abnormality of ventricles and ventricular septum (disorder) **93078006** Congenital hypertrophy of cardiac ventricle (disorder) **871668002** Congenital right ventricular anomaly (disorder) **768552007** Congenital ventricular septal defect (disorder)

	67278007 Congenital stenosis of pulmonary valve (disorder) **63934006** Overriding aorta (disorder) **89792004** Right ventricular hypertrophy (disorder) **253303001** Solitary aortic trunk with pulmonary atresia (disorder)
Additional Information	An infant with TOF may experience episodes of cyanosis or hypoxia that result from shunting of unoxygenated blood across the VSD from the right to the left ventricle. Children who have a coexisting VSD and pulmonary stenosis, but do not have TOF, may experience similar episodes. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean a child has been diagnosed with TOF. TOF is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants (approximately 1 in 7) with these defects have a deletion on the short arm of chromosome 22 (deletion 22q11.2). This deletion is diagnosed using fluorescent in situ hybridization (FISH) or other targeted 22q11.2 assays, and may be missed by routine karyotype analysis. TOF is on a spectrum with other defects having important physiological and coding differences among systems as seen in the table below. Pulmonary atresia with a ventricular septal defect (VSD) is similar to severe forms of TOF and is included here for surveillance. There is not an applicable code depicting valvular pulmonary atresia with VSD; hence in CDC/BPA the code 747.31 ("Pulmonary artery - Atresia with septal defect") is used. For pulmonary valve atresia without a VSD (intact ventricular septum (IVS)), the CDC/BPA code 746.00 ("Pulmonary valve - Atresia or hypoplasia") is used – see separate section on Pulmonary valve atresia.

CCHD	ICD-10	CDC/BPA
PVS	Q22.1	746.01
PA, IVS	Q22.0	746.00
PA, VSD (TOF)	Q21.3	747.31
TOF	Q21.3	745.20 - 21

[1] Salemi JL, Rutkowski RE, Tanner JP, Matas JL, Kirby RS. Identifying algorithms to improve the accuracy of unverified diagnosis codes for birth defects. Public Health Rep. 2018;133(3):303-310. doi:10.1177/0033354918763168.

Total anomalous pulmonary venous connection (TAPVC)

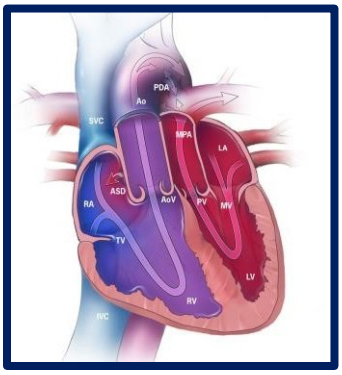
Total anomalous pulmonary venous connection (TAPVC) *Cardiovascular*	
Description	A condition in which all 4 pulmonary veins connect anomalously into the systemic venous circulation instead of the left atrium. The anomalous connection(s) can be to the right atrium or to systemic veins (e.g., the venae cavae) above or below the diaphragm. TAPVC often occurs with other cardiac anomalies, including heteroxy (laterality anomalies). 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	24
CDC/BPA Codes	**747.42** Total anomalous pulmonary venous return (TAPVR)
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates abnormal drainage anatomy of the entire pulmonary venous system consistent with total anomalous pulmonary venous connection. **Accepted:** The following criterion must be met: • Postnatal diagnosis of total anomalous pulmonary venous connection without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of total anomalous pulmonary venous connection assigned postnatally, as described in the "Diagnoses Included for Case Classification of Total Anomalous Pulmonary Venous Connection" section below.

<table>
<tr><th colspan="2">Diagnoses Included for Case Classification of Total Anomalous Pulmonary Venous Connection</th></tr>
<tr><td>Inclusions</td><td>• Total anomalous pulmonary venous connection (TAPVC)
• Total anomalous pulmonary venous return (TAPVR)
• Total anomalous pulmonary venous drainage (TAPVD)</td></tr>
<tr><td>ICD-10-CM Codes</td><td>Q26.2 Total anomalous pulmonary venous connection
Total Anomalous Pulmonary Venous Connection (ICD10CM) OID:
2.16.840.1.113762.1.4.1146.2335</td></tr>
<tr><td>SNOMED CT Codes</td><td>Total Anomalous Pulmonary Venous Connection (SNOMED) OID:
2.16.840.1.113762.1.4.1146.2334</td></tr>
<tr><th colspan="2">Additional Information</th></tr>
<tr><td>Exclusions</td><td>• Partial pulmonary anomalous venous return, if fewer than 4 veins are visibly connecting/draining anomalously
• Prenatal finding of total anomalous pulmonary venous connections without postnatal confirmation
• CDC/BPA code 747.41: Persistent left superior vena cava
• ICD-10 Q26.3: partial pulmonary anomalous venous return
• Summary table:
<table>
<tr><th>System</th><th>TAPVR code</th><th>PAPVR code</th></tr>
<tr><td>CDC/BPA</td><td>747.42</td><td>747.43</td></tr>
<tr><td>ICD-10-CM</td><td>Q26.2</td><td>Q26.3</td></tr>
</table></td></tr>
<tr><td>Other Codes for Case Ascertainment</td><td>Other codes that may be used for case ascertainment but require further review for potential total anomalous pulmonary venous connection:
<u>ICD-10-CM:</u>
Q26.8 Other congenital malformations of great veins
Q26.9 Congenital malformation of great vein, unspecified
O35.BXX0–O35.BXX9 Maternal care for (suspected) cardiac anomalies in the fetus

<u>SNOMED-CT:</u>
59631007 Anomalous pulmonary venous drainage (disorder)
111322000 Congenital anomaly of pulmonary veins (disorder)</td></tr>
<tr><td>Additional Information</td><td>The term anomalous venous connection, return, and drainage are often used interchangeably
Total anomalous pulmonary venous return can present as a critical neonatal emergency especially if the flow back to the heart is obstructed. Partial anomalous pulmonary venous return, depending on the anatomy, initially may remain clinically subtle
The medical record will likely subclassify TAPVR into several clinically relevant groups. The four main groups are distinguished by the site of the anomalous connection:
• Supracardiac (~50%)</td></tr>
</table>

	• Cardiac (20-25%) • Infracardiac (20-25%), • Mixed (5-10%) Infracardiac TAPVR is nearly invariably obstructed. Cardiac is typically not obstructed and has the best hemodynamic tolerance. Mixed often has the highest surgical complexity. Unlike other critical congenital heart defects, prenatal detection rates for TAPVR remain very low, particularly when it is isolated (not associated with other cardiac anomalies). If suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. Postnatal diagnosis may also be challenging due to the difficulty of imaging all 4 veins and may require several echocardiograms or more advanced imaging techniques. TAPVC often occurs with other cardiac anomalies, including heteroxy (laterality anomalies). When this happens, any concurrent abnormalities should be coded separately. The ICD-10-CM code Q26.8 (Other congenital malformations of great veins) can be used for case ascertainment. This code includes several anomalies of the venae cavae (absence, atresia, azygous continuation, etc.) as well as Scimitar 'syndrome', which is a form of PAPVR. For programs using ICD-9-CM codes or reviewing historical data, please note that the CDC/BPA code for this condition (747.42 Total anomalous pulmonary venous return (TAPVR)), is mismatched with the ICD-9-CM system. In ICD-9, this code is PAPVR (partial anomalous venous return), whereas 747.41 is TAPVR.

Transposition of the great arteries (TGA)

Transposition of the great arteries (TGA) *Cardiovascular*	
Description	Discordant connection between the ventricles and the great vessels 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). When the connection between atria and ventricles is concordant/normal (the left atrium to left ventricle, right atrium to right ventricle) the result is a critical cyanotic cardiac anomaly, due to the fact that the deoxygenated blood from the right ventricle is directed to the aorta (because of the discordant connection) instead of the lungs (via the pulmonary artery). TGA may occur with or without a ventricular septal defect. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	11 – Transposition of the Great Arteries (TGA) 74 – Dextro-transposition of the Great Arteries (d-TGA)
CDC/BPA Codes	**745.10** Transposition of great vessels (TGA, TGV) - Complete (no VSD) **745.11** Transposition of great vessels (TGA,TGV) - Incomplete (with VSD) **745.18** Transposition of great vessels (TGA, TGV) - Other specified, no mention of double outlet right ventricle **745.19** Transposition of great vessels (TGA, TGV) - Unspecified
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates ventriculo-arterial discordance consistent with transposition of the great arteries. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of transposition of the great arteries from a fetal echocardiogram [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of transposition of the great arteries without confirmatory imaging, surgery, or autopsy information.

Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of transposition of the great arteries assigned postnatally, as described in the "Diagnoses Included for Case Classification of Transposition of the Great Arteries" section below.
Diagnoses Included for Case Classification of Transposition of the Great Arteries	
Inclusions	• Complete or "dextro" transposition (d-TGA without a VSD) • Incomplete transposition (d-TGA with a VSD) • Transposition of the Great Arteries (TGA), not otherwise specified • Transposition of the Great Vessels (TGV)
ICD-10-CM Codes	**Q20.3** Discordant ventriculoarterial connection **Transposition of the Great Arteries [Unspecified] (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2310
SNOMED CT Codes	**D_Transposition of the Great Arteries (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2307 **Transposition of the Great Arteries [Unspecified] (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2309
Additional Information	
Exclusions	• Cases with codes for both DORV and TGA are counted in the DORV category • DORV subtype with malposed/transposed great arteries (CDC/BPA 745.14) are also counted in the DORV category • Corrected, or "levo" transposition (l-TGA)
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential transposition of the great arteries: **ICD-10-CM:** **Q20.8** Other congenital malformations of cardiac chambers and connections **Q20.9** Congenital malformation of cardiac chambers and connections, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **204300001** Incomplete great vessel transposition (disorder) **762250009** Congenital anomaly of great vessel (disorder) **253295000** Abnormal ventriculoarterial connection (disorder)

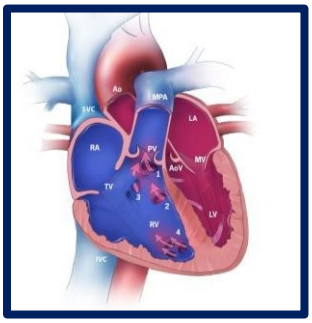
Additional Information	**Complete transposition of the great arteries (d-TGA)** In d-TGA, the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (ventriculoarterial discordance), while the atria connect normally to their respective ventricles (atrioventricular concordance). This creates two parallel circulations: deoxygenated blood recirculates through the body, and oxygenated blood recirculates through the lungs, with no exchange between them. Survival requires some communication between the two circulations to allow mixing of oxygenated and deoxygenated blood. This mixing may occur through a patent foramen ovale, atrial septal defect, ventricular septal defect (VSD), or patent ductus arteriosus. When a VSD is present, mixing is more adequate and cyanosis may be less severe at birth; when no such communication exists beyond a small PFO, profound cyanosis occurs immediately and urgent intervention (e.g., balloon atrial septostomy) is required. Older terminology used "complete TGA" for d-TGA without VSD and "incomplete TGA" for d-TGA with VSD. These terms are imprecise and are not recommended. When a VSD is present with d-TGA, it should be coded separately. **Association with 22q11.2 deletion** Transposition of the great arteries has been grouped with other outflow tract abnormalities known as conotruncal defects. However, the association between TGA and 22q11.2 deletion is considerably weaker than for other conotruncal defects such as interrupted aortic arch type B, truncus arteriosus, or tetralogy of Fallot. The 22q11.2 deletion may not be detected by routine karyotype and requires targeted testing such as FISH, MLPA, or chromosomal microarray.

Tricuspid valve atresia and stenosis

Tricuspid valve atresia and stenosis *Cardiovascular*	
Description	Tricuspid valve atresia: Lack of patency, or failure of formation altogether, of the tricuspid valve, resulting in complete obstruction of blood flow from the right atrium to the right ventricle. Tricuspid valve stenosis: Obstruction or narrowing of the tricuspid valve orifice, which impairs blood flow from the right atrium to the right ventricle.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	18
CDC/BPA Codes	**746.100** Tricuspid valve – Atresia **746.106** Tricuspid valve - Stenosis or hypoplasia, congenital **For CCHD Screening:** **746.100** Tricuspid valve – Atresia
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates absence or complete occlusion (atresia) or narrowing (stenosis) of the tricuspid valve orifice. **Accepted:** The following criterion must be met: • Postnatal diagnosis of tricuspid valve atresia or stenosis without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of tricuspid valve atresia or stenosis assigned postnatally, as described in the "Diagnoses Included for Case Classification of Tricuspid Valve Atresia and Stenosis" section below.

Diagnoses Included for Case Classification of Tricuspid Valve Atresia and Stenosis	
Inclusions	• Tricuspid atresia • Tricuspid stenosis
ICD-10-CM Codes	**Q22.4** Congenital tricuspid stenosis -This code is inclusive of congenital tricuspid atresia. **Tricuspid valve atresia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2337 **Tricuspid valve stenosis (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2339
SNOMED CT Codes	**Tricuspid valve atresia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2336 **Tricuspid valve stenosis (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2338
Additional Information	
Exclusions	• Tricuspid regurgitation without specific mention of tricuspid atresia or stenosis.
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential tricuspid valve atresia and stenosis: **ICD-10-CM:** **Q22.8** Other congenital malformations of tricuspid valve **Q22.9** Congenital malformation of tricuspid valve, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **4374004** Congenital anomaly of tricuspid valve (disorder) **123656005** Congenital atresia of cardiac valve (disorder) **123657001** Congenital stenosis of cardiac valve (disorder) **49915006** Tricuspid valve stenosis (disorder) **472841006** Stenosis of fetal tricuspid valve (disorder)
Additional Information	Tricuspid atresia and tricuspid stenosis are fundamentally different conditions in terms of hemodynamic severity, surgical pathway, and CCHD status. Tricuspid atresia is an absent right atrioventricular connection producing a functionally univentricular heart. It is a critical CHD that invariably requires staged palliation (typically Fontan pathway). Tricuspid stenosis is a narrowed but patent valve, which may range from mild to severe but still permits biventricular physiology. However, both ICD-9-CM and ICD-10-CM systems fail to separate tricuspid atresia and tricuspid stenosis. CDC/BPA recognized this limitation and is the one system that separates them for surveillance purposes. In ICD-10-CM, both tricuspid valve atresia and stenosis are included in Q22.4.

Ventricular septal defect

Ventricular septal defect *Cardiovascular*	
Description	An opening in the wall (septum) that separates the left and right ventricles of the heart. 
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	14
CDC/BPA Codes	**745.41** Eisenmenger syndrome **745.42** Gerbode defect **745.480** Ventricular septal defect (VSD) - Other specified **745.485** Ventricular septal defect (VSD) - Perimembranous or membranous **745.486** Ventricular septal defect (VSD) - Muscular, mid-muscular, apical **745.490** Ventricular septal defect (VSD) - NOS
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (echocardiography, cardiac catheterization with angiography, cardiac CT or MRI), surgery, or autopsy demonstrates opening in the septum consistent with ventricular septal defect. **Accepted:** The following criterion must be met: • Postnatal diagnosis of ventricular septal defect without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of ventricular septal defect assigned postnatally, as described in the "Diagnoses Included for Case Classification of Ventricular Septal Defect" section below.

Diagnoses Included for Case Classification of Ventricular Septal Defect	
Inclusions	• Ventricular septal defect (VSD) of all levels of severity
ICD-10-CM Codes	**Q21.0** Ventricular septal defect **Ventricular septal defect (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2529
SNOMED CT Codes	**Ventricular septal defect (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2528
Additional Information	
Exclusions	• Ventricular septal defects that occur as part of Tetralogy of Fallot or an atrioventricular septal defect • Inlet, subtricuspid, and canal-type VSDs are assumed to be part of an atrioventricular septal defect and should not be coded separately • Prenatal finding of VSD without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential ventricular septal defect: **ICD-10-CM:** **Q21.8** Other congenital malformations of cardiac septa **Q21.9** Congenital malformation of cardiac septum, unspecified **O35.BXX0–O35.BXX9** Maternal care for (suspected) cardiac anomalies in the fetus **SNOMED-CT:** **204312002** Ventricular septal defect between left ventricle and right atrium (disorder) **253483002** Abnormality of left atrioventricular valve in double inlet ventricle (disorder) **253486005** Left atrioventricular valve dysplasia (disorder) **253487001** Left atrioventricular valve hypoplasia (disorder) **253489003** Overriding left atrioventricular valve (disorder) **253492004** Straddling left atrioventricular valve (disorder) **253493009** Left atrioventricular valve leaflet abnormality (disorder) **253494003** Absent left atrioventricular valve leaflets (disorder) **253495002** Double orifice of left atrioventricular valve (disorder) **253497005** Left atrioventricular valve prolapse (disorder) **253498000** True cleft of left atrioventricular valve leaflet (disorder) **253499008** Accessory tissue on left atrioventricular valve leaflet (disorder) **253500004** Left atrioventricular valve leaflet dysplasia (disorder) **253501000** Abnormality of left atrioventricular valve chordae tendinae (disorder) **253502007** Left atrioventricular valve chordae too short (disorder) **253503002** Left atrioventricular valve chordae too long (disorder) **253504008** Left atrioventricular valve chordae to outlet septum (disorder)

	253505009 Arcade abnormality of left atrioventricular valve chordae (disorder) **253552003** Perimembranous ventricular septal defect with extension to right ventricular inlet (disorder) **253554002** Perimembranous ventricular septal defect with extension to right ventricular outlet (disorder) **253556000** Ventricular septal defect with malaligned outlet septum to right (disorder) **253559007** Ventricular septal defect with malaligned outlet septum to left (disorder) **253562005** Ventricular septal defect with absent outlet septum and overriding truncal valve (disorder) **253563000** Muscular ventricular septal defect in inlet septum (disorder) **253567004** Muscular ventricular septal defect in outlet septum (disorder) **253568009** Doubly committed subarterial ventricular septal defect (disorder) **253569001** Doubly committed subarterial ventricular septal defect with membranous septum extension (disorder) **253570000** Doubly committed subarterial ventricular septal defect with muscular posterior inferior rim (disorder) **447670001** Ventricular septal defect with anterior malaligned outlet septum with overriding pulmonary valve (disorder) **447671002** Ventricular septal defect with posterior malaligned outlet septum with overriding aortic valve (disorder) **447672009** Ventricular septal defect with posterior malaligned outlet septum with overriding pulmonary valve (disorder) **447698003** Ventricular septal defect with anterior malaligned outlet septum with overriding aortic valve (disorder) **447852001** Ventricular septal defect of inlet of right aspect of ventricular septum (disorder) **447941008** Residual ventricular septal defect (disorder) **448062009** Ventricular septal defect with malaligned outlet septum (disorder) **448475002** Absent pulmonary valve syndrome with ventricular septal defect of non Fallot type (disorder) **448780007** Ventricular septal defect with absent outlet septum and overriding truncal valve with extension of membranous septum (disorder) **448827000** Ventricular septal defect with absent outlet septum and overriding truncal valve with inferior muscular rim (disorder) **449040002** Hypoplasia of right atrioventricular valve annulus in double inlet ventricle (disorder) **449098005** Congenital abnormality of left atrioventricular valve chordae tendinae in double inlet ventricle (disorder) **449099002** Left atrioventricular valve stenosis in double inlet ventricle (disorder) **449178002** Doubly committed ventricular septal defect in double outlet ventriculoarterial connection (disorder) **449228007** Hypoplasia of left atrioventricular valve annulus in double inlet ventricle (disorder)

	460614005 Dilatation of left atrioventricular (not morphologically mitral) valve in double inlet ventricle (disorder) **762460002** Hemodynamically insignificant ventricular septal defect (disorder) **80387009** Roger's disease (disorder) **871579003** Perimembranous inlet ventricular septal defect with atrioventricular septal malalignment (disorder) **871583003** Congenital anomaly of left-sided atrioventricular valve in double inlet ventricle (disorder) **871585005** Congenital anomaly of right-sided atrioventricular valve in double inlet ventricle (disorder) **871600007** Outlet ventricular septal defect with posteriorly malaligned outlet septum (disorder) **871602004** Perimembranous outlet ventricular septal defect with anteriorly malaligned outlet septum (disorder) **871603009** Transposition of great arteries with concordant atrioventricular connections and ventricular septal defect (disorder) **871604003** Doubly committed juxta-arterial outlet ventricular septal defect with perimembranous extension and posteriorly malaligned outlet septum (disorder) **871605002** Transposition of great arteries with concordant atrioventricular connections and ventricular septal defect and left ventricular outflow tract obstruction (disorder) **871606001** Outlet ventricular septal defect with anteriorly malaligned outlet septum (disorder) **871607005** Doubly committed juxta-arterial outlet ventricular septal defect with perimembranous extension and anteriorly malaligned outlet septum (disorder) **871609008** Doubly committed juxta-arterial outlet ventricular septal defect with anteriorly malaligned outlet septum (disorder) **871610003** Doubly committed juxta-arterial outlet ventricular septal defect with perimembranous extension (disorder) **871611004** Doubly committed juxta-arterial outlet ventricular septal defect with posteriorly malaligned outlet septum (disorder) **871620008** Perimembranous inlet ventricular septal defect (disorder) **871636005** Doubly committed juxta-arterial ventricular septal defect with anteriorly malaligned fibrous outlet septum and muscular postero-inferior rim (disorder) **871648008** Perimembranous outlet ventricular septal defect with posteriorly malaligned outlet septum (disorder) **871658007** Muscular ventricular septal defect opening to right ventricular inlet (disorder) **871659004** Double inlet to solitary ventricle of indeterminate morphology (disorder)

Additional Information	VSDs are classified by the location of the defect along the ventricular septum. The main anatomic subtypes are muscular (the most common overall, particularly when small defects detected by echocardiography are included), perimembranous (the most common subtype among clinically significant VSDs requiring medical attention or intervention), inlet (not included here, classified under the atrioventricular septal defects), and outlet (also called supracristal, subpulmonary, or doubly committed subarterial). Many perimembranous and muscular VSDs close spontaneously during infancy, most commonly within the first 1–2 years of life. Spontaneous closure is rare for inlet and outlet VSDs. VSDs may be detected on prenatal ultrasound, but prenatal findings alone should not be used for case classification. Some VSDs detected prenatally close before delivery. Conversely, many VSDs — particularly small muscular defects — are not detectable on prenatal ultrasound because of the limited resolution of fetal imaging and the similar pressures between the ventricles during fetal life, which minimize flow across the defect. Postnatal confirmation is required.

Orofacial

Choanal atresia

Choanal atresia *Orofacial*	
Description	Congenital obstruction of the posterior openings of the nasal cavity (choanae) into the nasopharynx. The atresia can be unilateral or bilateral. Because babies are predominantly or obligate nose breathers, bilateral atresia is typically recognized at birth by respiratory distress.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	28
CDC/BPA Codes	**748.000** Choanal atresia, laterality unknown **748.001** Choanal atresia, left **748.002** Choanal atresia, right **748.003** Choanal atresia, unilateral NOS **748.004** Choanal atresia, bilateral
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal imaging (CT or MRI), surgery, or autopsy demonstrates obstruction of the choanae. • Postnatal physical examination documents a diagnosis of unilateral or bilaterial choanal atresia and no subsequent imaging, surgery, or autopsy refutes the diagnosis. **Accepted:** The following criterion must be met: • Postnatal diagnosis of choanal atresia without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of choanal atresia assigned postnatally, as described in the "Diagnoses Included for Case Classification of Choanal Atresia" section below.

Diagnoses Included for Case Classification of Choanal Atresia	
Inclusions	• Choanal atresia, type not specified • Membranous choanal atresia, with or without a bony rim • Completely bony choanal atresia
ICD-10-CM Codes	**Q30.0** Choanal atresia **Choanal Atresia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2497
SNOMED CT Codes	**Choanal Atresia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2496
Additional Information	
Exclusions	• Piriform/pyriform aperture atresia and stenosis • Choanal stenosis • Prenatal finding of choanal atresia without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential choanal atresia: **ICD-10-CM:** **Q30.8** Other congenital malformations of nose **Q30.9** Congenital malformation of nose, unspecified **O35.AXX0–O35.AXX9** Maternal care for (suspected) facial anomalies in the fetus **SNOMED-CT:** **47535005** Coloboma, heart defects, choanal atresia, retardation of growth and development, genitourinary problems, ear abnormalities syndrome (disorder) **722375007** Bamforth Lazarus syndrome (disorder) **1222709003** Syndromic congenital sodium diarrhea (disorder) **724141003** Microcephalic primordial dwarfism due to zinc finger protein 335 deficiency (disorder) **1367655003** Phosphoribosylaminoimidazole carboxylase deficiency disorder (disorder) **771072001** Monosomy 9p (disorder) **720496006** Anophthalmia plus syndrome (disorder) **1177166006** Frontonasal dysplasia, bifid nose, upper limb anomalies syndrome (disorder) **724144006** Embryofetopathy caused by methimazole (disorder) **1204421005** Lymphedema, posterior choanal atresia syndrome (disorder) **1281843005** Choanal atresia, athelia, hypothyroidism, delayed puberty, short stature syndrome (disorder) **1172697000** X-linked female restricted facial dysmorphism, short stature, choanal atresia, intellectual disability (disorder) **720009004** Intractable diarrhea with choanal atresia and eye anomaly syndrome (disorder) **720640005** Choanal atresia, hearing loss, cardiac defect, craniofacial dysmorphism syndrome (disorder)

Additional Information	Choanal atresia is one of the defects reported as part of the CHARGE association (SNOMED 47535005) which may also include colobomas, heart defects, retarded growth and development, genital hypoplasia, and ear anomalies and/or deafness. In addition to respiratory distress at birth in bilateral choanal atresia, both bilateral and unilateral choanal atresia may be strongly suspected by the inability to pass a feeding tube from the nasal passage(s) into the posterior pharynx. Additional imaging (e.g., scope, CT) will confirm the diagnosis. Piriform/pyriform aperture atresia and stenosis (an exclusion diagnosis) may also cause nasal obstruction and respiratory difficulty in babies, and imaging will help distinguish these conditions from choanal atresia. Piriform/pyriform aperture atresia and stenosis may present similarly to choanal atresia, but have important differences in management, and are excluded from this condition. Piriform/pyriform aperture atresia and stenosis are most often bilateral and are the result of bony overgrowth of the nasal process of the maxilla into the nasal cavity, which leads to a narrowing of the anterior nasal airway.

Cleft lip alone (without cleft palate)

Cleft lip alone (without cleft palate) *Orofacial*	
Description	A defect in the upper lip resulting from failure of fusion of the medial nasal and maxillary processes during embryonic development. For surveillance purposes, "cleft lip alone" requires the absence of a cleft of the hard or soft palate. A cleft involving only the alveolar ridge of the maxilla (primary palate) without a cleft of the secondary (hard or soft) palate is included under cleft lip alone. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	68
CDC/BPA Codes	**749.10** Cleft lip, unspecified **749.11** Cleft lip, unilateral complete **749.12** Cleft lip, unilateral incomplete **749.13** Cleft lip, bilateral complete **749.14** Cleft lip, bilateral incomplete **749.15** Cleft lip, median **749.19** Cleft lip, other
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Documented cleft lip (complete or incomplete, unilateral, bilateral or median) AND documented intact hard and soft palate on postnatal physical examination, imaging, surgery, or autopsy of the infant or in a clinical consultation report. **Accepted:** The following criterion must be met: • Postnatal diagnosis of cleft lip alone without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of cleft lip alone assigned postnatally, as described in the "Diagnoses Included for Case Classification of Cleft Lip Alone" section below.

Diagnoses Included for Case Classification of Cleft Lip Alone	
Inclusions	• Complete cleft lip — the defect extends through the full thickness of the lip and into the floor of the nose. • Incomplete cleft lip — the defect extends through part of the lip but does not reach the floor of the nose. • Unilateral (left, right) cleft lip. • Bilateral cleft lip. • Median (central) cleft lip. • Cleft lip with cleft of the alveolar process of the maxilla (primary palate) when the secondary palate (hard and/or soft) is intact. • Cheiloschisis; congenital fissure of lip; harelip; labium leporinum (These terms are archaic and should be avoided.)
ICD-10-CM Codes	**Q36.0** Cleft lip, bilateral **Q36.1** Cleft lip, median **Q36.9** Cleft lip, unilateral **Cleft Lip Alone (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2115
SNOMED CT Codes	**Cleft Lip Alone (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2114 **Fetal Cleft Lip (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2116
Additional Information	
Exclusions	• Cleft lip with cleft palate (any involvement of the hard or soft palate) • Cleft palate alone (without cleft lip) • Pseudocleft lip — an abnormal linear thickening, depressed groove, or scar-like pigmentary change of the lip without an actual cleft • Oblique facial clefts • Craniofacial (Tessier) clefts, macrostomia, and other radial facial clefts extending through facial and cranial bone • Congenital lip pits, fistulas, or sinuses without a cleft (Van der Woude features without a cleft) • Prenatal finding of cleft lip without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential cleft lip alone: **ICD-10-CM:** **O35.AXX0–O35.AXX9** Maternal care for (suspected) facial anomalies in the fetus **SNOMED-CT:** **722201004** Median cleft of upper lip, corpus callosum lipoma, cutaneous polyp syndrome (disorder) **723403008** Microbrachycephaly, ptosis, cleft lip syndrome (disorder) **725149008** Auricular abnormality, cleft lip, ocular abnormality syndrome (disorder)

	732247000 Cleft lip retinopathy syndrome (disorder)
Additional Information	Unilateral cleft lip is roughly twice as common as bilateral; left-sided clefts are more common than right-sided. Median cleft lip is rare and raises concern for midline CNS anomalies (e.g., holoprosencephaly) and may indicate a syndrome is present. Cleft lip can be identified on prenatal ultrasound (typically second trimester, mid-facial views). Prenatal ultrasound cannot reliably distinguish cleft lip alone from cleft lip with cleft palate because visualization of the secondary palate is limited. Cleft lip is identifiable on physical examination of the neonate. The distinction of cleft lip alone from cleft lip with cleft palate requires direct inspection of the hard and soft palate, which is often best documented by: • Plastic / craniofacial surgery consultation report • ENT evaluation • Digital or direct inspection of the oral cavity by a clinician • Cleft team documentation using LAHSAL, Kernahan-Stark, or other standardized notation The LAHSAL classification (Lip-Alveolus-Hard palate-Soft palate, recording Left and Right sides) is a useful way to capture completeness and laterality uniformly across programs[1]. A LAHSAL notation with entries only in the L or R "Lip" and/or "Alveolus" positions, with the Hard-palate and Soft-palate positions empty, corresponds to cleft lip alone.

[1] Shah SN, Khalid M, Khan MS. A review of classification systems for cleft lip and palate patients-I. Morphological classifications. Journal of Khyber College of Dentistry. 2011;1(2):95-99.

Cleft lip with cleft palate

Cleft lip with cleft palate *Orofacial*	
Description	An opening in the upper lip as well as the hard or soft palate resulting from failure of fusion of the medial nasal and maxillary processes during embryonic development. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	69
CDC/BPA Codes	**749.20** Cleft lip - Unilateral, with any cleft palate **749.21** Cleft lip - Bilateral, with any cleft palate **749.22** Cleft lip - Central, with any cleft palate **749.29** Cleft lip - NOS, with any cleft palate
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Documented cleft lip (complete or incomplete, unilateral, bilateral or median) AND documented cleft of the hard and/or soft palate on postnatal physical examination, imaging, surgery, or autopsy of the infant or in a clinical consultation report. **Accepted:** The following criterion must be met: • Postnatal diagnosis of cleft lip with cleft palate without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** One of the following criteria must be met[1]: • Presence of more than one distinct coded or written diagnosis of cleft lip with cleft palate within an encounter, as described in the "Diagnoses Included for Case Classification of Cleft Lip with Cleft Palate" section below. • Presence of a coded or written diagnosis of cleft lip with cleft palate in more than one healthcare encounter, as described in the "Diagnoses Included for Case Classification of Cleft Lip with Cleft Palate" section below. **Accepted:** The following criterion must be met:

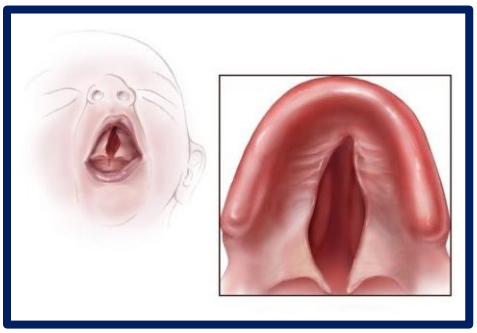
	• Presence of a coded or written diagnosis of cleft lip with cleft palate assigned postnatally, as described in the "Diagnoses Included for Case Classification of Cleft Lip with Cleft Palate" section below, without the presence of confirmatory information.
Diagnoses Included for Case Classification of Cleft Lip with Cleft Palate	
Inclusions	• Cleft lip with cleft of the hard and soft palate • Cleft lip with cleft of the hard palate • Cleft lip with cleft of the soft palate • Cleft lip with cleft palate, not otherwise specified • Cheilopalatoschisis
ICD-10-CM Codes	**Q37.0** Cleft hard palate with bilateral cleft lip **Q37.1** Cleft hard palate with unilateral cleft lip **Q37.2** Cleft soft palate with bilateral cleft lip **Q37.3** Cleft soft palate with unilateral cleft lip **Q37.4** Cleft hard and soft palate with bilateral cleft lip **Q37.5** Cleft hard and soft palate with unilateral cleft lip **Q37.8** Unspecified cleft palate with bilateral cleft lip **Q37.9** Unspecified cleft palate with unilateral cleft lip **Cleft Lip with Cleft Palate (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2120
SNOMED CT Codes	**Cleft Lip with Cleft Palate (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2119
Additional Information	
Exclusions	• Pseudocleft lip with cleft palate – An abnormal linear thickening, depressed grove, or scar-like pigmentary change on the skin of the lip without an actual cleft • Oblique facial clefts with cleft palate • Cleft palate without an associated cleft lip • Cleft lip without an associated cleft palate • Craniofacial (Tessier) clefts or Macrostomia - Clefts that extend through radiating axes of facial and cranial bone • Congenital lip pits, fistulas, or sinuses without clefts in both the lip and hard or soft palate (Van der Woude features without a cleft) • Cleft of the alveolar process of the maxilla (primary palate) without involvement of the hard or soft palate is classified as cleft lip alone, not cleft lip with cleft palate • Prenatal finding of cleft lip with cleft palate without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential cleft lip with cleft palate: **ICD-10-CM:** **O35.AXX0–O35.AXX9** Maternal care for (suspected) facial anomalies in the fetus **SNOMED-CT:**

	716248001 Cleft lip and cleft palate with ectodermal dysplasia syndrome **719456001** Cleft lip and cleft palate with intestinal malrotation and cardiopathy syndrome **1187039001** Cleft lip and palate, craniofacial dysmorphism, congenital heart defect, hearing loss syndrome **716007007** Cleft palate and cleft lip with deafness and sacral lipoma syndrome **1208338004** Dysraphism, cleft lip and palate, limb reduction defect syndrome **783159001** Holzgreve syndrome **721836009** Hypertelorism with microtia and facial clefting syndrome **771013004** Pilotto syndrome **719042007** Uveal coloboma with cleft lip and palate and intellectual disability syndrome **733605002** XY type gonadal dysgenesis with associated anomalies syndrome **764697003** Verloove Vanhorick Brubakk syndrome (disorder)
Additional Information	Unilateral cleft lip is roughly twice as common as bilateral; left-sided clefts are more common than right-sided. Median cleft lip is rare and raises concern for midline CNS anomalies (e.g., holoprosencephaly) and may indicate a syndrome is present. Cleft lip is identifiable on physical examination of the neonate. Cleft palate is usually identifiable on physical examination of the hard and soft palate; however, submucous cleft palate and bifid uvula may be difficult to diagnose by physical examination during the first year of life. These findings are often best documented by: • Plastic / craniofacial surgery consultation report • ENT evaluation • Digital or direct inspection of the oral cavity by a clinician • Cleft team documentation using LAHSAL, Kernahan-Stark, or other standardized notation The LAHSAL classification (Lip-Alveolus-Hard palate-Soft palate, recording Left and Right sides) is a useful way to capture completeness and laterality uniformly across programs[2]. A LAHSAL notation with entries in the L or R "Lip" and/or "Alveolus" positions as well as the Hard-palate and/or Soft-palate positions corresponds to cleft lip with cleft palate.

[1] Salemi JL, Rutkowski RE, Tanner JP, Matas JL, Kirby RS. Identifying algorithms to improve the accuracy of unverified diagnosis codes for birth defects. Public Health Rep. 2018;133(3):303-310. doi:10.1177/0033354918763168.

[2] Shah SN, Khalid M, Khan MS. A review of classification systems for cleft lip and palate patients-I. Morphological classifications. Journal of Khyber College of Dentistry. 2011;1(2):95-99.

Cleft palate alone

Cleft palate alone *Orofacial*	
Description	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. Lips are intact. 
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	26
CDC/BPA Codes	**749.00** Cleft hard palate - Unilateral, without cleft lip **749.01** Cleft hard palate - Bilateral, without cleft lip **749.02** Cleft hard palate - Central, without cleft lip **749.03** Cleft hard palate - NOS, without cleft lip **749.04** Cleft soft palate - Unilateral, without cleft lip **749.05** Cleft soft palate - Bilateral, without cleft lip **749.06** Cleft soft palate - Central, without cleft lip **749.07** Cleft soft palate - NOS, without cleft lip **749.09** Cleft palate - NOS, hard or soft not specified, without cleft lip
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Documented cleft of the hard and/or soft palate AND documented absence of cleft lip on postnatal physical examination, imaging, surgery, or autopsy of the infant or in a clinical consultation report. **Accepted:** The following criterion must be met: • Postnatal diagnosis of cleft palate alone without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** One of the following criteria must be met[1]: • Presence of more than one distinct coded or written diagnosis of cleft palate alone within an encounter, as described in the "Diagnoses Included for Case Classification of Cleft Palate Alone" section below. • Presence of a coded or written diagnosis of cleft palate alone in more than one healthcare encounter, as described in the "Diagnoses

	Included for Case Classification of Cleft Palate Alone" section below. **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of cleft palate alone assigned postnatally, as described in the "Diagnoses Included for Case Classification of Cleft Palate Alone" section below, without the presence of confirmatory information.
Diagnoses Included for Case Classification of Cleft Palate Alone	
Inclusions	• Cleft palate, type not specified • Cleft hard palate • Cleft soft palate • Submucous cleft palate – A cleft in the soft palate that is covered by the mucosa or a thin muscle layer
ICD-10-CM Codes	**Q35.1** Cleft hard palate **Q35.3** Cleft soft palate **Q35.5** Cleft hard palate with cleft soft palate **Q35.9** Cleft palate, unspecified **Cleft Palate Alone (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2118
SNOMED CT Codes	**Cleft Palate Alone (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2117
Additional Information	
Exclusions	• Cleft palate that coexists with a cleft lip • Cleft of the alveolar process of the maxilla (primary palate) without involvement of the hard or soft palate is classified as cleft lip alone, not cleft palate • Cleft uvula • Tessier cleft • Prenatal finding of cleft without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential cleft palate: **ICD-10-CM** **O35.AXX0–O35.AXX9** Maternal care for (suspected) facial anomalies in the fetus **SNOMED-CT:** **128336000** Congenital anomaly of palate (disorder) **18910001** Cleft uvula (disorder) **109548000** Bilateral cleft of primary palate (disorder) **109546001** Cleft of primary palate (disorder) **783204009** Ankyloblepharon filiforme adnatum with cleft palate syndrome (disorder) **722375007** Bamforth Lazarus syndrome (disorder) **717909004** Bilateral microtia with deafness and cleft palate syndrome (disorder)

	718574003 Cleft palate with coloboma of eye and deafness syndrome (disorder) **719466009** Cleft palate with short stature and vertebral anomaly syndrome (disorder) **719468005** Cleft palate with stapes fixation and oligodontia syndrome (disorder) **1335869007** Cleft palate, congenital heart defect, intellectual disability syndrome (disorder) **763130006** Cleft palate, large ears, small head syndrome (disorder) **403772000** Cleft palate lateral synechia syndrome (disorder) **763278004** Facial dysmorphism, cleft palate, loose skin syndrome (disorder) **773628009** Frontonasal dysplasia, severe microphthalmia, severe facial clefting syndrome (disorder) **773749003** Genitopalatocardiac syndrome (disorder) **717822006** Goldberg Shprintzen megacolon syndrome (disorder) **718576001** Hydrocephalus with cleft palate and joint contracture syndrome (disorder) **719408007** Lethal omphalocele with cleft palate syndrome (disorder) **773282001** Macrosomia, microphthalmia, cleft palate syndrome (disorder) **722463001** Macular coloboma, cleft palate, hallux valgus syndrome (disorder) **715471007** Mesomelic dysplasia with cleft palate and camptodactyly syndrome (disorder) **719394002** Microcephalus cleft palate syndrome (disorder) **54036001** Oto-palato-digital syndrome, type I (disorder) **42432003** Oto-palato-digital syndrome, type II (disorder) **771186004** Poikiloderma, alopecia, retrognathism, cleft palate syndrome (disorder) **718766002** Spondyloepiphyseal dysplasia, craniosynostosis, cleft palate, cataract and intellectual disability syndrome (disorder) **766761000** X-linked cleft palate and ankyloglossia (disorder) **897570002** Distal arthrogryposis type 3 (disorder) **723439002** Native American myopathy (disorder) **726670008** Weaver Williams syndrome (disorder) **720636001** Cholestasis with pigmentary retinopathy and cleft palate syndrome (disorder) **1216940001** Joint contractures, developmental delay, Pierre Robin syndrome (disorder) **4602007** Robin sequence (disorder) **719823007** Ventricular extrasystoles with syncope, perodactyly and Robin sequence syndrome (disorder) **723461007** Pierre Robin sequence faciodigital anomaly syndrome (disorder) **723998001** Short stature, Pierre Robin sequence, cleft mandible, hand anomalies, clubfoot syndrome (disorder) **763744009** Intellectual disability, brachydactyly, Pierre Robin syndrome (disorder) **770681000** Robin sequence and oligodactyly syndrome (disorder)
Additional Information	Cleft palate is usually identifiable on physical examination of the hard and soft palate; however, submucous cleft palate and bifid uvula may be

	difficult to diagnose by physical examination during the first year of life. These findings are often best documented by: • Plastic / craniofacial surgery consultation report • ENT evaluation • Digital or direct inspection of the oral cavity by a clinician • Cleft team documentation using LAHSAL, Kernahan-Stark, or other standardized notation The LAHSAL classification (Lip-Alveolus-Hard palate-Soft palate, recording Left and Right sides) is a useful way to capture completeness and laterality uniformly across programs[2]. A LAHSAL notation with entries in the Hard-palate and/or Soft-palate positions and with no entries in the L or R "Lip" and/or "Alveolus" positions corresponds to cleft palate without cleft lip. Pierre-Robin sequence consists of a group of conditions affecting the jaw/mouth – a small lower jaw/chin (micrognathia) causes displacement of the tongue backwards (glossoptosis) and a U-shaped cleft palate[3]. Studies point towards multiple causes of this sequence. Cleft uvula was included in previous versions of this condition but is now excluded due to its mild presentation and high prevalence.

[1] Salemi JL, Rutkowski RE, Tanner JP, Matas JL, Kirby RS. Identifying algorithms to improve the accuracy of unverified diagnosis codes for birth defects. Public Health Rep. 2018;133(3):303-310. doi:10.1177/0033354918763168.
[2] Shah SN, Khalid M, Khan MS. A review of classification systems for cleft lip and palate patients-I. Morphological classifications. Journal of Khyber College of Dentistry. 2011;1(2):95-99.
[3] Johns Hopkins Medicine. Pierre Robin sequence. Johns Hopkins Medicine. Accessed June 24, 2026. https://www.hopkinsmedicine.org/health/conditions-and-diseases/pierre-robin-sequence.

Gastrointestinal

Biliary atresia

Biliary atresia *Gastrointestinal*	
Description	Congenital agenesis, absence, hypoplasia, obstruction or stricture of the lumen of the extrahepatic bile ducts.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	33
CDC/BPA Codes	**751.65** Hepatic or bile ducts - Agenesis or atresia
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal imaging, surgery, or autopsy documents biliary atresia. • Finding of blocked bile duct(s) via biliary excretion study, such as HIDA (hepatobiliary iminodiacetic acid) scan. **Accepted:** The following criterion must be met: • Postnatal diagnosis of biliary atresia without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of biliary atresia assigned postnatally, as described in the "Diagnoses Included for Case Classification of Biliary Atresia" section below.
Diagnoses Included for Case Classification of Biliary Atresia	
Inclusions	• Agenesis, absence, hypoplasia, obstruction or stricture of the bile duct(s)
ICD-10-CM Codes	**Q44.2** Atresia of bile ducts **Q44.3** Congenital stenosis and stricture of bile ducts **Biliary Atresia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2538
SNOMED CT Codes	**Biliary Atresia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2537

Additional Information	
Exclusions	• Congenital or neonatal hepatitis • Intrahepatic biliary atresia (absence or paucity of bile ducts within the liver) not associated with extrahepatic biliary atresia • Prenatal finding of biliary atresia without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential biliary atresia: **ICD-10-CM:** **Q44.79** Other congenital malformations of liver **O35.DXX0 – O35.DXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal gastrointestinal anomalies **SNOMED-CT:** **235916001** Ichthyosis congenita with biliary atresia (disorder) **717156002** Biliary atresia with splenic malformation syndrome (disorder) **4711003** Congenital anomaly of bile ducts (disorder) **253807009** Intrahepatic biliary atresia (disorder) **276723008** Intrahepatic biliary hypoplasia (disorder)
Additional Information	The liver contains intrahepatic bile ducts that join and coalesce to form the right and left hepatic ducts. These exit the liver at the hilum and unite to form the common hepatic duct, which is the first segment of the extrahepatic biliary system. The extrahepatic bile ducts include the common hepatic duct, the cystic duct (which drains the gallbladder), and the common bile duct (formed by the junction of the common hepatic duct and the cystic duct), which empties bile into the duodenum. The more common physiologic neonatal jaundice is due to indirect (unconjugated) hyperbilirubinemia, is typically treated with phototherapy, and does not indicate biliary atresia. Biliary atresia presents with direct (conjugated) hyperbilirubinemia, which does not respond to phototherapy. While biliary atresia may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of biliary atresia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Esophageal atresia

Esophageal atresia *Gastrointestinal*	
Description	Esophageal atresia – A condition in which the esophagus ends in a blind pouch and fails to connect with the stomach.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	29
CDC/BPA Codes	**750.30** Esophageal atresia - Without tracheoesophageal (T-E) fistula (TEF) **750.31** Esophageal atresia - With tracheoesophageal (T-E) fistula (TEF) **750.330** Bronchoesophageal fistula - With or without esophageal atresia **750.350** Esophageal web
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal CT or MRI scan, surgery, or autopsy documents esophageal atresia. • Postnatal X-ray documentation of failure of a feeding tube to pass from the pharynx into the stomach. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of esophageal atresia [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of esophageal atresia without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of esophageal atresia assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Esophageal Atresia" section below, without the presence of confirmatory information.

Diagnoses Included for Case Classification of Esophageal Atresia	
Inclusions	• Esophageal atresia alone • Esophageal atresia with tracheoesophageal fistula (proximal, distal, or both) • Esophageal web
ICD-10-CM Codes	**Q39.0** Atresia of esophagus without fistula **Q39.1** Atresia of esophagus with tracheo-esophageal fistula **Q39.4** Esophageal web **Esophageal Atresia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.3313
SNOMED CT Codes	**Esophageal Atresia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.3314
Additional Information	
Exclusions	• Tracheal atresia • Tracheoesophageal cleft • TE fistula without esophageal atresia • Esophageal stenosis or stricture
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Esophageal atresia/tracheoesophageal fistula: **ICD-10-CM:** **Q39.8** Other congenital malformations of esophagus **Q39.9** Congenital malformation of esophagus, unspecified **O35.DXX0 – O35.DXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal gastrointestinal anomalies **SNOMED-CT:** **69771008** Congenital anomaly of esophagus (disorder) **14532008** Congenital anomaly of trachea (disorder) **27742002** Vertebral anomalies/dysgenesis, anal atresia, tracheo-esophageal fistula, esophageal atresia, renal anomalies, radial dysplasia association (disorder)
Additional Information	Tracheoesophageal (TE) fistula – An abnormal communication between the esophagus and the trachea. This is almost always associated with some form of esophageal atresia. To aid in surveillance standardization between programs, TE fistula without esophageal atresia is not included in this condition because it is rarely identified in the window monitored by birth defect programs. TE fistula and esophageal atresia are included as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, anal atresia, renal defects, and radial limb anomalies. Prenatal ultrasound findings suggestive of esophageal atresia include if the stomach or stomach bubble cannot be visualized, or the presentation of polyhydramnios.

Rectal and large intestinal atresia/stenosis

Rectal and large intestinal atresia/stenosis *Gastrointestinal*	
Description	Complete or partial occlusion or complete absence of the lumen of one or more segments of the large intestine and/or rectum.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	30
CDC/BPA Codes	**751.20** Large intestine - Stenosis, atresia or absence **751.21** Rectum - Stenosis, atresia or absence, with fistula **751.22** Rectum - Stenosis, atresia or absence without mention of fistula **751.23** Anus - Stenosis, atresia or absence, with fistula **751.24** Anus - Stenosis, atresia or absence, without mention of fistula
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal imaging, surgery, or autopsy documents rectal or large intestinal atresia/stenosis. • Documentation of physical exam diagnosing anal atresia/imperforate anus. • Documented barium enema test finding of obstruction, atresia or stenosis. **Accepted:** The following criterion must be met: • Postnatal diagnosis of rectal or large intestinal atresia/stenosis without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of rectal or large intestinal atresia/stenosis assigned postnatally, as described in the "Diagnoses Included for Case Classification of Rectal and Large Intestinal Atresia/Stenosis" section below.

Diagnoses Included for Case Classification of Rectal and Large Intestinal Atresia/Stenosis	
Inclusions	• Anal atresia or stenosis • Colonic atresia or stenosis • Imperforate anus • Large intestinal atresia or stenosis (include types: I, II, IIIa, IV, and not stated) • Rectal atresia or stenosis
ICD-10-CM Codes	**Q42.0** Congenital absence, atresia and stenosis of rectum with fistula **Q42.1** Congenital absence, atresia and stenosis of rectum without fistula **Q42.2** Congenital absence, atresia and stenosis of anus with fistula **Q42.3** Congenital absence, atresia and stenosis of anus without fistula **Q42.8** Congenital absence, atresia and stenosis of other parts of large intestine **Q42.9** Congenital absence, atresia and stenosis of large intestine, part unspecified **Rectal and Large Intestinal Atresia/Stenosis (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2601
SNOMED CT Codes	**Rectal and Large Intestinal Atresia/Stenosis (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2600
Additional Information	
Exclusions	• Apple peel intestinal atresia (Type IIIb) • Duodenal atresia or stenosis • Ileal atresia or stenosis • Jejunal atresia or stenosis • Small intestinal atresia or stenosis • Prenatal finding of rectal or large intestinal atresia/stenosis without postnatal confirmation • Anteriorly displaced anus • Absence of large intestine – While included in the descriptions for CDC/BPA or ICD10CM codes relevant for this condition, this is separate from atresia/stenosis. The inclusions in code descriptions are unlikely to affect surveillance estimates due to the rarity of this finding.
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Rectal and large intestinal atresia/stenosis: **ICD-10-CM:** **Q43.2** Other congenital functional disorders of colon **Q43.8** Other specified congenital malformations of intestine **Q43.9** Congenital malformation of intestine, unspecified **O35.DXX0 – O35.DXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal gastrointestinal anomalies **SNOMED-CT:** **86993003** Congenital anomaly of rectum (disorder)

	737197004 Congenital stenosis of large intestine (disorder) **27742002** Vertebral anomalies/dysgenesis, anal atresia, tracheo-esophageal fistula, esophageal atresia, renal anomalies, radial dysplasia association (disorder)
Additional Information	Large intestinal and rectal atresia or stenosis may be suspected by the clinical presentation of failure to pass meconium or stool, abdominal distension, and/or bilious vomiting. These conditions may occur with or without a fistula. Anal atresia is one of the defects reported as part of the VATE/ VACTERL, association, which may also include vertebral and cardiac defects, TE fistula/esophageal atresia, renal defects, and limb anomalies. Prenatal ultrasound is not a reliable diagnostic procedure for colonic and rectal atresia, with false negatives and false positives. Prenatal diagnoses should not be included in surveillance data without postnatal confirmation.

Small intestinal atresia/stenosis

Small intestinal atresia/stenosis *Gastrointestinal*	
Description	Complete or partial occlusion of the lumen of one or more segments of the small intestine. Small intestinal atresias are often assigned a type descriptor in the surgical or autopsy report, depending upon the severity of the atresia (types include I, II, IIIA, IIIB, and VI).
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	67
CDC/BPA Codes	**751.10** Duodenum - Stenosis, atresia or absence **751.11** Jejunum - Stenosis, atresia or absence of jejunum **751.12** Ileum - Stenosis, atresia or absence **751.190** Small intestine, NOS - Stenosis, atresia or absence **751.195** Small intestine, NOS - Stenosis, atresia or absence, with fistula
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal surgery or autopsy documents small intestinal atresia/stenosis. **Accepted:** The following criterion must be met: • Postnatal diagnosis of small intestinal atresia/stenosis without confirmatory surgery or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of small intestinal atresia/stenosis assigned postnatally, as described in the "Diagnoses Included for Case Classification of Small Intestinal Atresia/Stenosis" section below.
Diagnoses Included for Case Classification of Small Intestinal Atresia/Stenosis	
Inclusions	• Duodenal atresia or stenosis (also include duodenal web, membrane, diaphragm, or windsock); include all types: I, II, IIIA, IIIB, VI, and not stated • Jejunal atresia or stenosis (also include jejunal web or membrane); include all types: I, II, IIIA, IIIB, VI, and not stated

	• Ileal atresia or stenosis (also include ileal web or membrane); include all types: I, II, IIIA, IIIB, VI, and not stated • Small intestinal atresia or stenosis, not otherwise specified; include all types: I, II, IIIA, IIIB, VI, and not stated
ICD-10-CM Codes	**Q41.0** Congenital absence, atresia and stenosis of duodenum **Q41.1** Congenital absence, atresia and stenosis of jejunum **Q41.2** Congenital absence, atresia and stenosis of ileum **Q41.8** Congenital absence, atresia and stenosis of other specified parts of small intestine **Q41.9** Congenital absence, atresia and stenosis of small intestine, part unspecified **Small Intestinal Atresia and Stenosis (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2632
SNOMED CT Codes	**Small Intestinal Atresia and Stenosis (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2633
Additional Information	
Exclusions	• Intestinal atresia/stenosis in an infant with cystic fibrosis • Sirenomelia • Anal atresia or stenosis • Anal stenosis, anteriorly displaced anus • Colonic atresia or stenosis • Imperforate anus • Large intestinal atresia or stenosis • Rectal atresia or stenosis • Prenatal finding of small intestinal atresia/stenosis without further postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Small intestinal atresia/stenosis: **ICD-10-CM:** **Q43.2** Other congenital functional disorders of colon **Q43.8** Other specified congenital malformations of intestine **Q43.9** Congenital malformation of intestine, unspecified **SNOMED-CT:** **55193002** Congenital anomaly of small intestine (disorder) There are no codes specific to diagnosis of Small intestinal atresia/stenosis in the fetus. Fetal-indication obstetric codes that may encompass findings of Small intestinal atresia/stenosis include: **O35.DXX0 – O35.DXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal gastrointestinal anomalies

Additional Information	The diagnosis may be suspected by clinical presentation of abdominal distension, vomiting, lack of passage of meconium, “double bubble” sign on abdominal ultrasound, dilated loops of bowel on abdominal x-ray, or failure of contrast to advance on upper GI or barium enema studies. Approximately one-third of all infants with duodenal atresia or stenosis have Down syndrome. While these conditions may be suspected by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation; postnatal diagnosis of the small intestinal atresia or stenosis requires a surgical or autopsy report (i.e., ultrasound or abdominal x-ray studies, such as an upper GI or barium enema, are not sufficient). In addition, the absence of small intestinal atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Genitourinary

Bladder exstrophy

Bladder exstrophy *Genitourinary*	
Description	A congenital defect of the lower abdominal wall and the anterior wall of the urinary bladder in which the posterior bladder wall is exposed (everted) through the abdominal wall. The pubic rami are widely separated (pubic diastasis), and the abdominal-wall muscles and fascia are disrupted in the midline. Epispadias is almost uniformly present.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	35
CDC/BPA Codes	**753.50** Exstrophy of urinary bladder
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal imaging (abdominal / pelvic ultrasound, contrast studies (e.g., voiding cystourethrogram), CT or MRI of the pelvis, pelvic x-ray for pubic-diastasis measurement), surgery, or autopsy demonstrates bladder exstrophy without concurrent omphalocele and imperforate anus (i.e., not cloacal exstrophy). • Postnatal physical examination documents a diagnosis of bladder exstrophy with a patent anus and no subsequent imaging, surgery, or autopsy refutes the diagnosis. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of bladder exstrophy [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of bladder exstrophy without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of bladder exstrophy assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Bladder Exstrophy" section below.

Diagnoses Included for Case Classification of Bladder Exstrophy	
Inclusions	• Classic bladder exstrophy • Ectopia vesicae • Extroversion of the urinary bladder • Vesical exstrophy • Supravesical fissure of the urinary bladder • Covered (closed) exstrophy, pseudoexstrophy, duplicate exstrophy, superior vesical fistula, inferior vesicle, and other variants of bladder exstrophy • Bladder Exstrophy–Epispadias–Cloacal exstrophy Complex (BEEC), the bladder-exstrophy subset
ICD-10-CM Codes	**Q64.10** Exstrophy of urinary bladder, unspecified **Q64.11** Supravesical fissure of urinary bladder **Q64.19** Other exstrophy of urinary bladder **Bladder Exstrophy (ICD10CT) OID:** 2.16.840.1.113762.1.4.1146.2492
SNOMED CT Codes	**Bladder Exstrophy (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2491
Additional Information	
Exclusions	• Cloacal exstrophy (bladder exstrophy accompanied by omphalocele and imperforate anus) • Isolated epispadias • Ambiguous genitalia without documented bladder exstrophy • Congenital absence of the bladder • Urachal anomalies without bladder exstrophy • Omphalocele on prenatal ultrasound together with suspected bladder exstrophy cannot be reliably distinguished from cloacal exstrophy prenatally; postnatal confirmation is required
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential bladder exstrophy: **ICD-10-CM:** **Q64.70** Unspecified congenital malformation of bladder and urethra **Q64.79** Other congenital malformations of bladder and urethra **O35.EXX0–O35.EXX9** Maternal care for (suspected) genitourinary anomalies in the fetus **SNOMED-CT:** **37939008** Congenital anomaly of urinary bladder (disorder) **1230270001** Exstrophy epispadias complex (disorder)

Additional Information	In classic bladder exstrophy, the urinary tract is open anteriorly from the urethral meatus to the umbilicus. The pubic rami are separated. The abdominal muscles and fascia are disrupted. The external genitalia may be bifid or duplicated. Classic bladder exstrophy occurs more frequently in males, whereas variants occur more frequently in females. Prenatal ultrasound findings suggestive of bladder exstrophy include non-visualization of a fluid-filled bladder on serial examinations, a low-set abdominal-wall mass, a low-inserted umbilical cord, and widened pubic rami. Classic bladder exstrophy is recognized on physical examination at delivery because the exposed posterior bladder wall, pubic diastasis, and abnormal external genitalia are visible. Epispadias accompanies essentially all cases of bladder exstrophy and should not be recorded as a separate defect. Ambiguous genitalia is frequently documented in bladder-exstrophy cases in the absence of a clearly formed scrotum, and should not be recorded as a separate defect. The Bladder Exstrophy–Epispadias–Cloacal exstrophy Complex (BEEC) spans a continuum from epispadias through bladder exstrophy to cloacal exstrophy. For surveillance, each endpoint is ascertained and counted under its specific category (epispadias, bladder exstrophy, or cloacal exstrophy). When both bladder exstrophy and cloacal exstrophy features are documented, the case is counted under cloacal exstrophy only.

Cloacal exstrophy

Cloacal exstrophy *Genitourinary*	
Description	A complex anterior abdominal-wall and pelvic malformation consisting of the four-component OEIS complex: **O**mphalocele, bladder **E**xstrophy, **I**mperforate anus, and **S**pinal defects. Cloacal exstrophy represents the most severe end of the Bladder Exstrophy–Epispadias–Cloacal exstrophy Complex (BEEC) and presents with exposed hindgut (cecal plate) situated between two hemi-bladders.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	66
CDC/BPA Codes	**751.555** Cloacal exstrophy
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal imaging (abdominal and pelvic ultrasound, contrast studies, CT or MRI of the abdomen, pelvis and spine, Spinal MRI (to assess tethered cord, lipomeningocele, myelomeningocele)), surgery, or autopsy demonstrates the four OEIS components or the full anatomic picture of cloacal exstrophy. • Postnatal physical examination documents omphalocele, bladder exstrophy, and imperforate anus together and no subsequent imaging, surgery, or autopsy refutes the diagnosis. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of cloacal exstrophy [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of cloacal exstrophy without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of cloacal exstrophy assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Cloacal Exstrophy" section below.

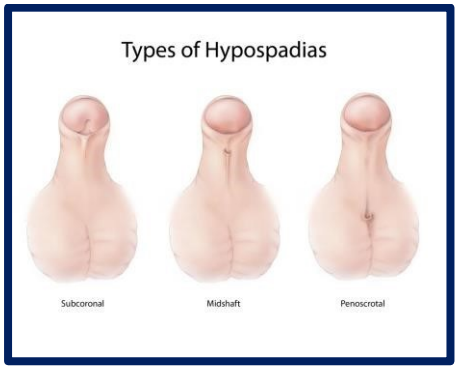
Diagnoses Included for Case Classification of Cloacal Exstrophy	
Inclusions	• Cloacal exstrophy • OEIS complex (Omphalocele, Exstrophy of the bladder, Imperforate anus, Spinal defects) • The cloacal-exstrophy subset of the Bladder Exstrophy–Epispadias–Cloacal exstrophy Complex (BEEC)
ICD-10-CM Codes	**Q64.12** Cloacal exstrophy of urinary bladder **Cloacal Exstrophy (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2499
SNOMED CT Codes	**Cloacal Exstrophy (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2498
Additional Information	
Exclusions	• Bladder exstrophy without both omphalocele and imperforate anus • Persistent cloaca (urorectal septum malformation sequence, URSMS) — this is a distinct entity in which urinary, intestinal, and genital tracts empty into a common cloaca without exstrophy • Isolated omphalocele without bladder exstrophy and imperforate anus • Isolated imperforate anus
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential bladder exstrophy: **ICD-10-CM:** **Q64.70** Unspecified congenital malformation of bladder and urethra **Q64.79** Other congenital malformations of bladder and urethra **O35.EXX0–O35.EXX9** Maternal care for (suspected) genitourinary anomalies in the fetus **SNOMED-CT:** **37939008** Congenital anomaly of urinary bladder (disorder) **1230270001** Exstrophy epispadias complex (disorder)
Additional Information	In classic cloacal exstrophy, the exposed hindgut plate is positioned between two exstrophied hemi-bladders, with the umbilical cord inserted at the superior margin of a low omphalocele. Pubic diastasis is wide. The external genitalia of either sex may be bifid or duplicated. Cloacal exstrophy may be suspected prenatally when ultrasound shows a combination of non-visualized bladder, infraumbilical abdominal-wall defect, omphalocele, and/or spinal anomaly (OEIS). Prenatal diagnosis cannot reliably distinguish cloacal exstrophy from bladder exstrophy in all cases. Ambiguous genitalia is frequently noted in cloacal-exstrophy cases. Epispadias is uniformly present. Spinal defects in cloacal exstrophy most commonly include lipomeningocele, myelomeningocele, or sacral dysgenesis. When both bladder exstrophy and cloacal exstrophy features are present, the case is counted only under cloacal exstrophy.

Congenital posterior urethral valves

Congenital posterior urethral valves *Genitourinary*	
Description	Posterior urethral valves (PUV) are tissue folds of the posterior urethra and function as valves obstructing urine outflow. Congenital PUV is an abnormal congenital obstructing membrane that is located within the posterior male urethra; this valve is the most common cause of bladder outlet obstruction in male children. Congenital PUV can also be found in virilized females and rarely in normal females. Obstruction could vary from mild to severe.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	65
CDC/BPA Codes	**753.60** Posterior urethral valves, congenital
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal finding of congenital PUV from contrast study, such as voiding cystourethrogram (VCUG). • Postnatal direct visualization of congenital PUV using cytoscopy or urethral endoscopy. • Postnatal surgery or autopsy documents congenital PUV. **Accepted:** The following criterion must be met: • Postnatal diagnosis of congenital PUV without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of congenital posterior urethral valves assigned postnatally, as described in the "Diagnoses Included for Case Classification of Congenital Posterior Urethral Valves" section below.
Diagnoses Included for Case Classification of Congenital Posterior Urethral Valves	
Inclusions	• Posterior urethral valves of all levels of severity
ICD-10-CM Codes	**Q64.2** Congenital posterior urethral valves

	Congenital Posterior Urethral Valves (ICD10CM) OID: 2.16.840.1.113762.1.4.1146.2490
SNOMED CT Codes	**Congenital Posterior Urethral Valves (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2489
Additional Information	
Exclusions	• Inhibition of urinary flow resulting solely from neurologic impairment • Prenatal finding of Congenital PUV without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Congenital Posterior Urethral Valves: **ICD-10-CM:** **Q64.8** Other specified congenital malformations of urinary system **Q64.9** Congenital malformation of urinary system, unspecified **O35.EXX0 – O35.EXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal genitourinary anomalies **SNOMED-CT:** **297163001** Congenital urethral valve (disorder) **13806003** Congenital anomaly of urethra (disorder)
Additional Information	Congenital PUV can present at birth with abdominal masses, distended bladder, hydronephrosis, or with respiratory distress, oligohydramnios, and Potter facies. When urine flow is obstructed, the portion of the genitourinary tract proximal to the affected area may become enlarged and dilated with urine. Mild lesions may produce only partial or intermittent urinary obstruction without permanent damage. More severe lesions may substantially or completely obstruct urine flow, resulting in permanent damage to proximal structures, and sometimes impaired kidney function, if not relieved by surgery. Congenital PUV may be suspected prenatally by routine obstetric ultrasonography. However, some resolve spontaneously prior to birth. For this reason, PUV should not be included in surveillance data without postnatal confirmation. Conversely, the absence of genitourinary obstruction on prenatal ultrasound does not necessarily mean that an obstructive defect such as PUV will not be diagnosed after delivery. Congenital PUV may not present until later in infancy or early childhood if the obstruction is not complete or is intermittent. These defects should still be counted in surveillance data if they are discovered outside the newborn period.

Hypospadias

Hypospadias *Genitourinary*	
Description	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. 
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	50
CDC/BPA Codes	**752.600** Hypospadias without chordee - Degree not specified **752.605** Hypospadias without chordee - First-degree (glandular or coronal) **752.606** Hypospadias without chordee - Second-degree (penile) **752.607** Hypospadias without chordee - Third-degree (perineal or scrotal) **752.620** Chordee, congenital, with hypospadias - Degree not specified **752.625** Chordee, congenital, with first-degree hypospadias (glandular or coronal) **752.626** Chordee, congenital, with second-degree hypospadias (penile) **752.627** Chordee, congenital, with third-degree hypospadias (perineal or scrotal)
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal diagnosis of hypospadias with or without confirmatory exam, imaging, surgery, or autopsy information. **Accepted:** N/A
Tier 2 Surveillance	**Confirmed:** The following criterion must be met[1,2]: • Presence of a coded or written diagnosis of hypospadias assigned postnatally, as described in the "Diagnoses Included for Case Classification of Hypospadias" section below. **Accepted:**

	N/A
Diagnoses Included for Case Classification of Hypospadias	
Inclusions	• First-degree hypospadias – The urethral meatus is located on the glans of the penis. Also called primary, 1°, glandular, or coronal hypospadias. • Second-degree hypospadias – The urethral meatus is located on the shaft of the penis. Also called secondary, 2°, subcoronal or penile shaft hypospadias. • Third-degree hypospadias – The urethral meatus is located at the base of the penis on the scrotum or perineum. Also called tertiary, 3°, scrotal, penoscrotal, or perineal hypospadias. • Hypospadias, degree not specified • Hypospadias of any type with chordee • Megameatus
ICD-10-CM Codes	**Q54.0** Hypospadias, balanic **Q54.1** Hypospadias, penile **Q54.2** Hypospadias, penoscrotal **Q54.3** Hypospadias, perineal **Q54.8** Other hypospadias **Q54.9** Hypospadias, unspecified **Hypospadias (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2517
SNOMED CT Codes	**Hypospadias (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2516
Additional Information	
Exclusions	• Chordee alone without associated hypospadias • Ambiguous genitalia • Epispadias • Prenatal finding of hypospadias without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential hypospadias: **ICD-10-CM:** **Q54.4** Congenital chordee **Q55.8** Other specified congenital malformations of male genital organs **Q55.9** Congenital malformation of male genital organ, unspecified **Q64.8** Other specified congenital malformations of urinary system **Q64.9** Congenital malformation of urinary system, unspecified **O35.EXX0 – O35.EXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal genitourinary anomalies **SNOMED-CT:** **13806003** Congenital anomaly of urethra (disorder) **716092007** Radial hypoplasia and triphalangeal thumb with hypospadias and maxillary diastema syndrome (disorder)

	716096005 Hypospadias and intellectual disability syndrome Goldblatt type (disorder) **722452004** Preaxial deficiency, postaxial polydactyly, hypospadias syndrome (disorder) **763889002** Spina bifida and hypospadias syndrome (disorder) **770561007** Lower limb malformation hypospadias syndrome (disorder)
Additional Information	Chordee indicates a ventral (downward) curve of the penis, which may result from cutaneous or fibrous restriction. It is present in approximately 35% to 50% of cases of hypospadias. In mild forms of first-degree hypospadias, the foreskin may appear hooded but there may be no overt clinical symptoms. Milder forms such as balanic (glanular) hypospadias are easily missed at delivery and might be discovered during circumcision. In contrast, third-degree hypospadias may be described as ambiguous genitalia. In this instance, it is important to search the medical record for detailed information (including chromosome, molecular, and hormone analyses; genetics and endocrinology consultations; surgery or autopsy reports) that may clarify the anatomy and/or indicate whether an underlying genetic condition or endocrinopathy associated with ambiguous genitalia is present. Ambiguous genitalia should not be coded if hypospadias is the only diagnosis. Hypospadias generally should not be coded if a normal female karyotype (46,XX) is reported. While this condition may be diagnosed by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of hypospadias on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery.

[1] Salemi JL, Rutkowski RE, Tanner JP, Matas JL, Kirby RS. Identifying algorithms to improve the accuracy of unverified diagnosis codes for birth defects. Public Health Rep. 2018;133(3):303-310. doi:10.1177/0033354918763168.
[2] Salemi JL, Tanner JP, Sampat D, Anjohrin SB, Correia JA, Watkins SM, et al. The accuracy of hospital discharge diagnosis codes for major birth defects: evaluation of a statewide registry with passive case ascertainment. J Public Health Manag Pract. 2016;22(3):E9-E19. doi:10.1097/PHH.0000000000000291.

Renal agenesis/hypoplasia

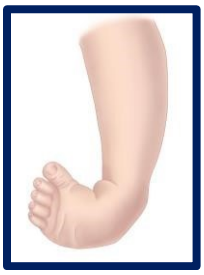
Renal agenesis/hypoplasia *Genitourinary*	
Description	Renal agenesis – Complete absence of the kidney Renal hypoplasia – Incomplete development of the kidney
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	34
CDC/BPA Codes	**753.000** Kidneys - Absence, agenesis, dysplasia, or hypoplasia - Bilateral **753.009** Kidneys - Absence, agenesis, dysplasia, or hypoplasia - NOS **753.010** Kidney - Absence, agenesis, dysplasia, or hypoplasia, laterality unknown **753.011** Kidney - Absence, agenesis, dysplasia, or hypoplasia, unilateral, left **753.012** Kidney - Absence, agenesis, dysplasia, or hypoplasia, unilateral, right **753.013** Kidney - Absence, agenesis, dysplasia, or hypoplasia, unilateral, NOS
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging, surgery, or autopsy documents renal agenesis/hypoplasia. **Accepted:** One of the following criteria must be met: • Prenatal imaging documents bilateral renal agenesis and no information from further postnatal evaluation is available. Prenatal ultrasound findings suggestive of bilateral renal agenesis include oligohydramnios with non-visualization of both kidneys. • Postnatal diagnosis of renal agenesis/hypoplasia without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of renal agenesis/hypoplasia assigned postnatally, as described in the "Diagnoses Included for Case Classification of Renal agenesis/hypoplasia" section below.

Diagnoses Included for Case Classification of Renal Agenesis/Hypoplasia	
Inclusions	• Renal agenesis, dysgenesis, aplasia, or hypoplasia (bilateral, unilateral, or not specified) • Potter syndrome secondary to renal agenesis/hypoplasia
ICD-10-CM Codes	**Q60.0** Renal agenesis, unilateral **Q60.1** Renal agenesis, bilateral **Q60.2** Renal agenesis, unspecified **Q60.3** Renal hypoplasia, unilateral **Q60.4** Renal hypoplasia, bilateral **Q60.5** Renal hypoplasia, unspecified **Q60.6** Potter's syndrome **Renal Agenesis or Hypoplasia (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2501
SNOMED CT Codes	**Renal Agenesis or Hypoplasia (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2500
Additional Information	
Exclusions	• Cystic renal dysplasia • Cystic kidney disease • Multicystic kidney • Multicystic dysplastic kidney • Polycystic kidney • Renal cysts • Renal dysplasia • Small kidney
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Renal agenesis/hypoplasia: **ICD-10-CM:** **Q64.8** Other specified congenital malformations of urinary system **Q64.9** Congenital malformation of urinary system, unspecified **O35.EXX0 – O35.EXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal genitourinary anomalies **SNOMED-CT:** **44513007** Congenital anomaly of the kidney (disorder) **204949001** Renal dysplasia (disorder) **204950001** Bilateral renal dysplasia (disorder) **717742006** Primary renal dysplasia (disorder) **717744007** Secondary renal dysplasia (disorder) **717745008** Bilateral secondary renal dysplasia (disorder) **765775002** Dysplasia of left kidney (disorder) **765776001** Dysplasia of right kidney (disorder) **24814002** Potter's facies (disorder) **1237470001** Lethal fetal brain malformation, duodenal atresia, bilateral renal hypoplasia syndrome (disorder)

	733116005 Aniridia, renal agenesis, psychomotor retardation syndrome (disorder)
Additional Information	Bilateral renal agenesis is often suspected on physical examination after delivery because of the Potter phenotype: low-set cartilage-deficient ears, prominent epicanthal folds, flattened "parrot-beaked" nose, recessed chin, limb contractures, malformed hands, and clubbed feet. Bilateral renal hypoplasia may or may not be recognized after delivery, depending on the severity and degree of residual kidney function. Unilateral renal agenesis or hypoplasia may not be symptomatic at delivery if the contralateral kidney is not impaired. Renal agenesis and hypoplasia may be unilateral or bilateral. If the defect coding system includes unique codes for these different types, the location should be coded. Bilateral renal agenesis, or any condition that significantly impairs the function of both kidneys in utero, may lead to the oligohydramnios sequence (Potter syndrome) due to lack of fetal urine production and the resulting decreased amniotic fluid volume. The sequence includes minor facial dysmorphism (flat face, small chin, large ears), pulmonary hypoplasia, and joint contractures. Bilateral renal agenesis is incompatible with long-term survival unless a kidney transplant is performed. In contrast, unilateral renal agenesis/hypoplasia may not be diagnosed until weeks, months, or even years after birth if the contralateral kidney function is normal. Some unilateral cases may be diagnosed only as incidental findings during evaluation for other conditions, and some may never be recognized.

Musculoskeletal

Clubfoot

Clubfoot *Musculoskeletal*	
Description	An abnormality consisting of plantar flexion (downward pointing of the foot and toes), inversion (varus, or internal rotation), and metatarsus adductus (deviation of the forefoot toward the body) of the foot. An abnormally high arch (pes cavus) and midfoot flexion crease usually are also present. 
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	64
CDC/BPA Codes	**754.500** Talipes equinovarus, Laterality Unk **754.501** Talipes equinovarus, Left **754.502** Talipes equinovarus, Right **754.503** Talipes equinovarus, Unilateral NOS **754.504** Talipes equinovarus, Bilateral **754.730** Clubfoot, NOS, Laterality Unk **754.731** Clubfoot, NOS, Left **754.732** Clubfoot, NOS, Right **754.733** Clubfoot, NOS, Unilateral NOS **754.734** Clubfoot, NOS, Bilateral
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Documented clubfoot on postnatal physical examination, imaging, surgery, or autopsy of the infant or in a clinical consultation report. **Accepted:** The following criterion must be met: • Postnatal diagnosis of clubfoot without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met:

	• Presence of a coded or written diagnosis of clubfoot assigned postnatally, as described in the "Diagnoses Included for Case Classification of Clubfoot" section below.
Diagnoses Included for Case Classification of Clubfoot	
Inclusions	• Talipes equinovarus (including congenital, idiopathic, and neurogenic) • Talipes not otherwise specified • Clubfoot not otherwise specified
ICD-10-CM Codes	**Q66.00** Congenital talipes equinovarus, unspecified foot **Q66.01** Congenital talipes equinovarus, right foot **Q66.02** Congenital talipes equinovarus, left foot **Clubfoot (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2543
SNOMED CT Codes	**Clubfoot (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2541
Additional Information	
Exclusions	• Talipes equinovalgus / Talipes calcaneovarus / Talipes calcaneovalgus / Talipes varus / Talipes valgus • Vertical talus • Metatarsus adductus alone / Metatarsus varus alone • Pes varus / Pes valgus / Pes planus • Rocker-bottom foot • Positional or postural clubfoot
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential clubfoot: **ICD-10-CM:** **Q66.89** Other specified congenital deformities of feet **Q66.90** Congenital deformity of feet, unspecified, unspecified foot **Q66.91** Congenital deformity of feet, unspecified, right foot **Q66.92** Congenital deformity of feet, unspecified, left foot **O35.HXX0–O35.HXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal lower extremities anomalies **SNOMED-CT:** **63387002** Larsen syndrome (disorder) **66651005** Triploidy syndrome (disorder) **766987006** Moebius syndrome (disorder) **63051000119101** Fetal clubfoot (disorder)

Additional Information	Clubfoot can occur on either side alone or in both feet. The calf muscles on the affected side are permanently small. While in some instances the affected foot can be moved passively to a normal or near-normal position (so-called positional clubfoot), more commonly there is a component of rigidity which can be severe. Clubfoot often occurs alone but can be associated with other musculoskeletal abnormalities such as torticollis or developmental dysplasia of the hip, and with genetic syndromes such as triploidy, Larsen syndrome, or Moebius sequence. Neurogenic clubfoot results from impaired innervation of the foot during development. Examples of conditions that can result in such impairment include spina bifida, arthrogryposis, sacral agenesis, spinal muscular atrophy, and other paralytic states.

Craniosynostosis

Craniosynostosis *Musculoskeletal*	
Description	Premature closure (fusion) of one or several cranial sutures (connective tissue membranes that separate the bones of the developing skull).
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	63
CDC/BPA Codes	**756.000** Craniosynostosis, NOS, laterality unknown **756.001** Craniosynostosis, NOS, left **756.002** Craniosynostosis, NOS, right **756.003** Craniosynostosis, NOS, unilateral NOS **756.004** Craniosynostosis, NOS, bilateral **756.005** Craniosynostosis – Sagittal **756.006** Craniosynostosis – Metopic **756.010** Craniosynostosis – Coronal, laterality unknown **756.011** Craniosynostosis – Coronal, left **756.012** Craniosynostosis – Coronal, right **756.013** Craniosynostosis – Coronal, unilateral NOS **756.014** Craniosynostosis – Coronal, bilateral **756.020** Craniosynostosis – Lambdoidal, laterality unknown **756.021** Craniosynostosis – Lambdoidal, left **756.022** Craniosynostosis – Lambdoidal, right **756.023** Craniosynostosis – Lambdoidal, unilateral NOS **756.024** Craniosynostosis – Lambdoidal, bilateral **756.030** Craniosynostosis – Other types
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal documented closure of one or more cranial sutures through diagnostic imaging (skull X-ray or tomography), surgery, or autopsy. • Postnatal physical examination by a pediatric neurologist or craniofacial specialist documents a diagnosis of craniosynostosis and no subsequent imaging, surgery, or autopsy refutes the diagnosis. **Accepted:** The following criterion must be met: • Postnatal diagnosis of craniosynostosis without confirmatory exam, imaging, surgery, or autopsy information.

Tier 2 Surveillance	**<u>Confirmed:</u>** N/A **<u>Accepted:</u>** The following criterion must be met: • Presence of a coded or written diagnosis of Craniosynostosis assigned postnatally, as described in the "Diagnoses Included for Case Classification of Craniosynostosis" section below.
Diagnoses Included for Case Classification of Craniosynostosis	
Inclusions	• Craniosynostosis subtypes are typically named by the cranial sutures involved: sagittal, coronal, lambdoidal, or metopic craniosynostoses are the most common conditions. Mixed or multiple sutures can be involved, and rarely basilar or squamosal sutures fuse prematurely. • Cloverleaf skull Cranial shapes resulting from craniosynostosis: • Oxycephaly/Turricephaly/Acrocephaly – tall, tower-like skull (sometimes pointed) resulting from premature fusion of coronal and usually sagittal sutures • Trigonocephaly – triangular-shaped skull resulting from premature closure of metopic suture
ICD-10-CM Codes	**Q75.001** Craniosynostosis, unspecified type, unilateral **Q75.002** Craniosynostosis, unspecified type, bilateral **Q75.009** Craniosynostosis, unspecified **Q75.01** Sagittal craniosynostosis **Q75.021** Coronal craniosynostosis, unilateral **Q75.022** Coronal craniosynostosis, bilateral **Q75.029** Coronal craniosynostosis, unspecified **Q75.03** Metopic craniosynostosis **Q75.041** Lambdoid craniosynostosis, unilateral **Q75.042** Lambdoid craniosynostosis, bilateral **Q75.049** Lambdoid craniosynostosis, unspecified **Q75.051** Cloverleaf skull **Q75.052** Pansynostosis **Q75.058** Other multi-suture craniosynostosis **Q75.08** Other single-suture craniosynostosis
SNOMED CT Codes	**57219006** Craniosynostosis syndrome (disorder) **711039004** Acrobrachycephaly (disorder) **205260006** Acrocephalopolysyndactyly (disorder) **403767009** Acrocephalopolysyndactyly type II (disorder) **403768004** Acrocephalopolysyndactyly type III (disorder) **268262006** Acrocephalosyndactyly (disorder) **205258009** Acrocephalosyndactyly type I (disorder) **70410008** Acrocephalosyndactyly type V (disorder) **48069004** Acrocephaly (disorder) **722281001** Agammaglobulinemia, microcephaly, craniosynostosis, severe dermatitis syndrome (disorder)

62964007 Antley-Bixler syndrome (disorder)
77608001 Baller-Gerold syndrome (disorder)
254021002 Bicoronal craniosynostosis (disorder)
720815000 Capra DeMarco syndrome (disorder)
720606005 Cardiocranial syndrome Pfeiffer type (disorder)
254022009 Cloverleaf skull syndrome (disorder)
717771007 Cloverleaf skull with multiple congenital anomalies syndrome (disorder)
783181006 Cloverleaf skull, asphyxiating thoracic dysplasia syndrome (disorder)
8611000119100 Complex craniosynostosis (disorder)
1003602001 Congenital synostosis of all sutures of skull (disorder)
1003586008 Congenital synostosis of coronal suture of skull (disorder)
1003594001 Congenital synostosis of left coronal suture of skull (disorder)
1003600009 Congenital synostosis of left side of lambdoid suture of skull (disorder)
1003595000 Congenital synostosis of right coronal suture of skull (disorder)
1003599006 Congenital synostosis of right side of lambdoid suture of skull (disorder)
720755009 Craniofacial dyssynostosis syndrome (disorder)
720757001 Craniofrontonasal dysplasia with Poland anomaly syndrome (disorder)
725098001 Craniomicromelic syndrome (disorder)
784350004 Craniorhiny (disorder)
773332008 Craniosynostosis and dental anomalies syndrome (disorder)
720816004 Craniosynostosis and intracranial calcification syndrome (disorder)
720817008 Craniosynostosis Boston type (disorder)
732250002 Craniosynostosis fibular aplasia syndrome (disorder)
763684005 Craniosynostosis Herrmann Opitz type (disorder)
720818003 Craniosynostosis Philadelphia type (disorder)
720813007 Craniosynostosis with Dandy-Walker malformation and hydrocephalus syndrome (disorder)
720812002 Craniosynostosis, anal anomaly, porokeratosis syndrome (disorder)
1269224009 Craniosynostosis, microretrognathia, severe intellectual disability syndrome (disorder)
28861008 Crouzon syndrome (disorder)
702361006 Crouzon syndrome with acanthosis nigricans (disorder)
720819006 Curry Jones syndrome (disorder)
703528008 Cutis gyrata syndrome of Beare and Stevenson (disorder)
39401000 Dolichocephalic dwarfism (disorder)
725030006 Familial scaphocephaly syndrome McGillivray type (disorder)
778008009 Fibroblast growth factor receptor 2-related bent bone dysplasia (disorder)
440350001 Fibroblast growth factor receptor 3-related craniosynostosis (disorder)
109413005 Fronto-malar faciosynostosis (disorder)
715434005 Holoprosencephaly craniosynostosis syndrome (disorder)
721227001 Hunter McAlpine craniosynostosis syndrome (disorder)
109409003 Interfrontal craniofaciosynostosis (disorder)
109418001 Interparietal craniosynostosis (disorder)
709105005 Jackson-Weiss syndrome (disorder)
773672007 Lethal occipital encephalocele, skeletal dysplasia syndrome (disorder)
72239002 Long narrow head (disorder)
721974000 Lowry MacLean syndrome (disorder)
1179283004 Metopic ridging, ptosis, facial dysmorphism syndrome (disorder)

	787407003 Muenke syndrome (disorder) **1365683002** Non-syndromic bicoronal and metopic craniosynostosis (disorder) **1363589008** Non-syndromic bicoronal and sagittal craniosynostosis (disorder) **1365889000** Non-syndromic bilambdoid craniosynostosis (disorder) **1363596005** Non-syndromic metopic and sagittal craniosynostosis (disorder) **1231181001** Non-syndromic metopic craniosynostosis (disorder) **1365719004** Non-syndromic unicoronal and sagittal craniosynostosis (disorder) **1365886007** Non-syndromic unifrontosphenoidal craniosynostosis (disorder) **1365888008** Non-syndromic unilambdoid craniosynostosis (disorder) **1365887003** Non-syndromic unisquamosal craniosynostosis (disorder) **722051004** Obesity, colitis, hypothyroidism, cardiac hypertrophy, developmental delay syndrome (disorder) **109417006** Parieto-occipital craniosynostosis (disorder) **1003877009** Pfeiffer syndrome type 1 (disorder) **1003916008** Pfeiffer syndrome type 2 (disorder) **1003918009** Pfeiffer syndrome type 3 (disorder) **715867000** Pseudoaminopterin syndrome (disorder) **83015004** Saethre-Chotzen syndrome (disorder) **719069008** Shprintzen Goldberg craniosynostosis syndrome (disorder) **255581000119100** Simple craniosynostosis (disorder) **734173003** Skeletal abnormality, cutis laxa, craniostenosis, ambiguous genitalia, retardation, facial abnormality syndrome (disorder) **109416002** Spheno-fronto-parietal craniofaciosynostosis (disorder) **718766002** Spondyloepiphyseal dysplasia, craniosynostosis, cleft palate, cataract and intellectual disability syndrome (disorder) **28740008** Trigonocephaly (disorder) **715409005** Trigonocephaly C syndrome (disorder) **719948009** Trigonocephaly with bifid nose and acral anomaly syndrome (disorder) **719949001** Trigonocephaly with broad thumb syndrome (disorder) **733066002** Trigonocephaly, short stature, developmental delay syndrome (disorder) **254020001** Unicoronal craniosynostosis (disorder)
Additional Information	
Exclusions	• Deformational plagiocephaly without synostosis • Other abnormal head shapes without craniosynostosis
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential craniosynostosis: **ICD-10-CM:** **Q75.8** Other specified congenital malformations of skull and face bones **Q75.9** Congenital malformation of skull and face bones, unspecified **Q67.4** Other congenital deformities of skull, face and jaw **O35.AXX0–O35.AXX9** Maternal care for (suspected) facial anomalies in the fetus **SNOMED-CT:** **72239002** Long narrow head (disorder) **13649004** Brachycephaly (disorder) **21850008** Plagiocephaly (disorder)

Additional Information	Cranial shapes that may or may not result from craniosynostosis: • Dolichocephaly/Scaphocephaly – long, wedge-shaped skull with a prominent forehead and occiput resulting from premature closure of sagittal suture • Brachycephaly – high, wide, short skull resulting from premature fusion of coronal sutures • Plagiocephaly – asymmetric skull shape which can result from unilateral closure of coronal and/or lambdoidal suture Craniosynostosis is seen in many syndromes such as the acrocephalosyndactylies, in which there are limb abnormalities such as syndactyly. A particularly severe form of craniosynostosis of multiple sutures is called cloverleaf skull or Kleeblattschädel; this condition is usually associated with a syndrome diagnosis. Syndromes with craniosynostosis as a necessary component include but are not limited to Antley-Bixler syndrome, Baller-Gerold Syndrome, Capra DeMarco syndrome, Crouzon syndrome, Curry Jones syndrome, Jackson-Weiss syndrome, Lowry MacLean syndrome, Muenke syndrome, Pfeiffer syndrome, Pseudoaminopterin syndrome, and Saethre-Chotzen syndrome. Craniosynostosis can be identified or suspected on prenatal ultrasound; however, rates of prenatal detection of craniosynostosis are low, particularly for non-syndromic craniosynostosis. Prenatally identified craniosynostosis should be counted as an accepted case without postnatal confirmation.

Diaphragmatic hernia

Diaphragmatic hernia *Musculoskeletal*	
Description	Incomplete formation of the diaphragm through which a portion of the abdominal contents herniate into the thoracic cavity.
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	43
CDC/BPA Codes	**756.600** Absent diaphragm **756.610** Diaphragmatic hernia - Congenital, NOS **756.615** Diaphragmatic hernia - Bochdalek **756.616** Diaphragmatic hernia - Morgagni **756.617** Hemidiaphragm
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal imaging (X-ray, contrast study of the bowel, CT, or MRI scan), surgery, or autopsy demonstrates herniation of abdominal contents into the thoracic cavity. **Accepted:** One of the following criteria must be met: • A prenatal diagnosis of diaphragmatic hernia [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of diaphragmatic hernia without confirmatory imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of diaphragmatic hernia assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Diaphragmatic Hernia" section below.

Diagnoses Included for Case Classification of Diaphragmatic Hernia	
Inclusions	• Absence of the diaphragm • Bochdalek hernia – Herniation through a defect in the posterolateral portion of the diaphragm • Diaphragmatic hernia, type not specified • Hemidiaphragm • Morgagni hernia – Herniation through a defect in the anterior portion of the diaphragm • Paraesophageal hernia – Herniation through a defect in the central portion of the diaphragm surrounding the esophagus
ICD-10-CM Codes	**Q79.0** Congenital diaphragmatic hernia **Diaphragmatic hernia (Disorders) (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2540
SNOMED CT Codes	**Diaphragmatic hernia (Disorders) (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2539
Additional Information	
Exclusions	Eventration of the diaphragm – Weakness in, or absence of, the muscles of the diaphragm which allows upward displacement of a portion of the abdominal contents. However, there is no true herniation of contents through the diaphragm into the thoracic cavity.
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential diaphragmatic hernia: **ICD-10-CM:** **Q79.1** Other congenital malformations of diaphragm **Q40.1** Congenital hiatus hernia **K44.0** Diaphragmatic hernia with obstruction, without gangrene **K44.1** Diaphragmatic hernia with gangrene **K44.9** Diaphragmatic hernia without obstruction or gangrene **O35.FXX0–O35.FXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal musculoskeletal anomalies of trunk **SNOMED-CT:** **39839004** Diaphragmatic hernia (disorder) **286972004** Complicated diaphragmatic hernia (disorder) **196904003** Irreducible diaphragmatic hernia (disorder) **196903009** Obstructed diaphragmatic hernia (disorder) and child concepts
Additional Information	Children with diaphragmatic hernia often have accompanying abnormalities of the heart, intestine, and lungs, including hypoplastic lungs, which result from the abnormal location of abdominal organs within the thoracic cavity during development.

Gastroschisis

Gastroschisis *Musculoskeletal*	
Description	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. The opening is separated from the umbilicus by a small bridge of skin, and the herniating organs are not covered by a protective membrane. Gastroschisis usually occurs on the right side of the umbilicus, although it may occur on the left. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	40
CDC/BPA Codes	**756.71** Gastroschisis
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** The following criterion must be met: • Postnatal diagnosis of gastroschisis with or without confirmatory exam, imaging, surgery, or autopsy information. **Accepted:** The following criterion must be met: • Prenatal ultrasound documents gastroschisis [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available.
Tier 2 Surveillance	**Confirmed:** The following criterion must be met[1,2,3,4]: • Presence of a coded or written diagnosis of gastroschisis assigned postnatally, as described in the "Diagnoses Included for Case Classification of Gastroschisis" section below. **Accepted:** N/A

Diagnoses Included for Case Classification of Gastroschisis	
Inclusions	• Gastroschisis
ICD-10-CM Codes	**Q79.3** Gastroschisis **Gastroschisis (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2132
SNOMED CT Codes	**Gastroschisis (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2131
Additional Information	
Exclusions	• Omphalocele
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential gastroschisis: **ICD-10-CM:** **Q79.59** Other congenital malformations of abdominal wall **Q79.8** Other congenital malformations of musculoskeletal system **Q79.9** Congenital malformation of musculoskeletal system, unspecified **O35.FXX0–O35.FXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal musculoskeletal anomalies of trunk **SNOMED-CT:** **363024001** Congenital anomaly of abdomen (disorder) **253818009** Congenital malformation of anterior abdominal wall (disorder)
Additional Information	The distinction between gastroschisis and omphalocele is important because they have different etiologies and different implications for treatment and long-term survival. In gastroschisis, the umbilicus and cord are normal and separated from the abdominal wall defect by a small bridge of skin. The herniating organs are not covered by a protective membrane. However, they may appear matted and covered by a thick fibrous material as a result of prolonged exposure to amniotic fluid in utero. In omphalocele, abdominal organs herniate through the umbilicus into the umbilical cord. There is no bridge of skin between the abdominal wall defect and the umbilicus and cord. While the herniating organs are covered by a protective membrane, this may rupture before, during, or after delivery. Gastroschisis may be one of the defects reported as part of the Limb-Body Wall complex. This is a disruption complex of the lateral body wall, which may also include limb reductions, neural tube defects, heart defects, and other anomalies. Maternal serum alpha-fetoprotein (MSAFP) and/or amniotic fluid alpha-fetoprotein (AFAFP) may be elevated with gastroschisis. However, these screening tests alone are not sufficient to diagnose the condition. It may be difficult to distinguish gastroschisis from omphalocele on prenatal ultrasound, and the terms sometimes are used interchangeably.

[1] Contreras D, Miura-Akagi B, Gibbons M, Ramirez GM. Gastroschisis: is the combination of diagnosis and procedure code sufficient for case-confirmation? Birth Defects Res. 2026;118(2):e70018. doi:10.1002/bdr2.70018.
[2] Howley M, Williford E, St. Louis A, Michalski A, Browne M, Fisher S. A comparison of active and passive surveillance strategies for selected birth defects in New York. Birth Defects Res. 2024;116:e2399. doi:10.1002/bdr2.2399.
[3] Salemi JL, Rutkowski RE, Tanner JP, Matas JL, Kirby RS. Identifying algorithms to improve the accuracy of unverified diagnosis codes for birth defects. Public Health Rep. 2018;133(3):303-310. doi:10.1177/0033354918763168.
[4] Salemi JL, Tanner JP, Sampat D, Anjohrin SB, Correia JA, Watkins SM, et al. The accuracy of hospital discharge diagnosis codes for major birth defects: evaluation of a statewide registry with passive case ascertainment. J Public Health Manag Pract. 2016;22(3):E9-E19. doi:10.1097/PHH.0000000000000291.

Limb deficiencies (reduction defects)

Limb deficiencies (reduction defects) *Musculoskeletal*	
Description	Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), fingers (phalanges), thigh (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges). Most limb deficiencies can be further grouped as terminal transverse, longitudinal preaxial, longitudinal postaxial, longitudinal axial, and intercalary.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	62
CDC/BPA Codes	**755.200** Absent upper limb, laterality unknown **755.201** Absent upper limb, left **755.202** Absent upper limb, right **755.203** Absent upper limb, unilateral NOS **755.204** Absent upper limb, bilateral **755.210** Absent upper arm and forearm, laterality unknown **755.211** Absent upper arm and forearm, left **755.212** Absent upper arm and forearm, right **755.213** Absent upper arm and forearm, unilateral NOS **755.214** Absent upper arm and forearm, bilateral **755.220** Absent upper arm only or forearm only, laterality unknown **755.221** Absent upper arm only or forearm only, left **755.222** Absent upper arm only or forearm only, right **755.223** Absent upper arm only or forearm only, unilateral NOS **755.224** Absent upper arm only or forearm only, bilateral **755.230** Absent forearm and hand, laterality unknown **755.231** Absent forearm and hand, left **755.232** Absent forearm and hand, right **755.233** Absent forearm and hand, unilateral NOS **755.234** Absent forearm and hand, bilateral **755.240** Absent hand or fingers, laterality unknown **755.241** Absent hand or fingers, left **755.242** Absent hand or fingers, right **755.243** Absent hand or fingers, unilateral NOS **755.244** Absent hand or fingers, bilateral **755.250** Split-hand malformation, laterality unknown **755.251** Split-hand malformation, left **755.252** Split-hand malformation, right **755.253** Split-hand malformation, unilateral NOS **755.254** Split-hand malformation, bilateral **755.260** Preaxial longitudinal reduction defect - Upper limb, laterality unknown

755.261 Preaxial longitudinal reduction defect - Upper limb, left
755.262 Preaxial longitudinal reduction defect - Upper limb, right
755.263 Preaxial longitudinal reduction defect - Upper limb, unilateral NOS
755.264 Preaxial longitudinal reduction defect - Upper limb, bilateral
755.265 Longitudinal reduction defect, type not specified - Upper limb
755.270 Postaxial longitudinal reduction defect - Upper limb, laterality unknown
755.271 Postaxial longitudinal reduction defect - Upper limb, left
755.272 Postaxial longitudinal reduction defect - Upper limb, right
755.273 Postaxial longitudinal reduction defect - Upper limb, unilateral NOS
755.274 Postaxial longitudinal reduction defect - Upper limb, bilateral
755.280 Other specified reduction defect - Upper limb, laterality unknown
755.281 Other specified reduction defect - Upper limb, left
755.282 Other specified reduction defect - Upper limb, right
755.283 Other specified reduction defect - Upper limb, unilateral NOS
755.284 Other specified reduction defect - Upper limb, bilateral
755.285 Transverse reduction defect - Upper limb, NOS
755.290 Unspecified reduction defect - Upper limb, laterality unknown
755.291 Unspecified reduction defect - Upper limb, left
755.292 Unspecified reduction defect - Upper limb, right
755.293 Unspecified reduction defect - Upper limb, unilateral NOS
755.294 Unspecified reduction defect - Upper limb, bilateral
755.300 Absent lower limb, laterality unknown
755.301 Absent lower limb, left
755.302 Absent lower limb, right
755.303 Absent lower limb, unilateral NOS
755.304 Absent lower limb, bilateral
755.310 Absent thigh and lower leg, laterality unknown
755.311 Absent thigh and lower leg, left
755.312 Absent thigh and lower leg, right
755.313 Absent thigh and lower leg, unilateral NOS
755.314 Absent thigh and lower leg, bilateral
755.320 Absent femur only or lower leg only, laterality unknown
755.321 Absent femur only or lower leg only, left
755.322 **A**bsent femur only or lower leg only, right
755.323 Absent femur only or lower leg only, unilateral NOS
755.324 Absent femur only or lower leg only, bilateral
755.330 Absent lower leg and foot, laterality unknown
755.331 Absent lower leg and foot, left
755.332 Absent lower leg and foot, right
755.333 Absent lower leg and foot, unilateral NOS
755.334 Absent lower leg and foot, bilateral
755.340 Absent foot or toes, laterality unknown
755.341 Absent foot or toes, left
755.342 Absent foot or toes, right
755.343 Absent foot or toes, unilateral NOS
755.344 Absent foot or toes, bilateral
755.350 Split-foot malformation, laterality unknown
755.351 Split-foot malformation, left

755.352 Split-foot malformation, right
755.353 Split-foot malformation, unilateral NOS
755.354 Split-foot malformation, bilateral
755.360 Longitudinal reduction defect, NOS - Lower limb, laterality unknown
755.361 Longitudinal reduction defect, NOS - Lower limb, left
755.362 Longitudinal reduction defect, NOS - Lower limb, right
755.363 Longitudinal reduction defect, NOS - Lower limb, unilateral NOS
755.364 Longitudinal reduction defect, NOS - Lower limb, bilateral
755.365 Preaxial longitudinal reduction defect - Lower limb
755.366 Postaxial longitudinal reduction defect - Lower limb
755.380 Other specified reduction defect - Lower limb, laterality unknown
755.381 Other specified reduction defect - Lower limb, left
755.382 Other specified reduction defect - Lower limb, right
755.383 Other specified reduction defect - Lower limb, unilateral NOS
755.384 Other specified reduction defect - Lower limb, bilateral
755.385 Transverse reduction defect, NOS - Lower limb
755.390 Unspecified reduction defect - Lower limb, laterality unknown
755.391 Unspecified reduction defect - Lower limb, left
755.392 Unspecified reduction defect - Lower limb, right
755.393 Unspecified reduction defect - Lower limb, unilateral NOS
755.394 Unspecified reduction defect - Lower limb, bilateral
755.400 Absent limb, NOS, Laterality Unknown
755.401 Absent limb, NOS, Left
755.402 Absent limb, NOS, Right
755.403 Absent limb, NOS, Unilateral NOS
755.404 Absent limb, NOS, Bilateral
755.410 Phocomelia, NOS, Laterality Unk
755.411 Phocomelia, NOS, Left
755.412 Phocomelia, NOS, Right
755.413 Phocomelia, NOS, Unilateral NOS
755.414 Phocomelia, NOS, Bilateral
755.420 Transverse reduction defect, NOS, Laterality Unk
755.421 Transverse reduction defect, NOS, Left
755.422 Transverse reduction defect, NOS, Right
755.423 Transverse reduction defect, NOS, Unilateral NOS
755.424 Transverse reduction defect, NOS, Bilateral
755.430 Longitudinal reduction defect, NOS, Laterality Unk
755.431 Longitudinal reduction defect, NOS, Left
755.432 Longitudinal reduction defect, NOS, Right
755.433 Longitudinal reduction defect, NOS, Unilateral NOS
755.434 Longitudinal reduction defect, NOS, Bilateral
755.440 Absent digits, NOS, Laterality Unk
755.441 Absent digits, NOS, Left
755.442 Absent digits, NOS, Right
755.443 Absent digits, NOS, Unilateral NOS
755.444 Absent digits, NOS, Bilateral
755.480 Other specified reduction defect of unspecified limb, Laterality Unk
755.481 Other specified reduction defect of unspecified limb, Left

	755.482 Other specified reduction defect of unspecified limb, Right **755.483** Other specified reduction defect of unspecified limb, Unilateral NOS **755.484** Other specified reduction defect of unspecified limb, Bilateral **755.490** Unspecified reduction defect of unspecified limb, Laterality Unk **755.491** Unspecified reduction defect of unspecified limb, Left **755.492** Unspecified reduction defect of unspecified limb, Right **755.493** Unspecified reduction defect of unspecified limb, Unilateral NOS **755.494** Unspecified reduction defect of unspecified limb, Bilateral
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal physical examination documents a diagnosis of limb deficiency and no subsequent imaging, surgery, or autopsy refutes the diagnosis. • Postnatal imaging (X-ray, CT, MRI), surgery, or autopsy confirms limb deficiency. **Accepted:** One of the following criteria must be met: • Prenatal ultrasound documents amelia (absence of an entire limb) [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of limb deficiency without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of limb deficiency assigned postnatally, as described in the "Diagnoses Included for Case Classification of Limb Deficiencies (Reduction Defects)" section below.
Diagnoses Included for Case Classification of Limb Deficiencies (Reduction Defects)	
Inclusions	***Transverse limb deficiency (reduction)*** – Complete or partial absence of the distal (furthest from the body) structures of the arm or leg in a transverse (cross-wise) plane at the point where the deficiency begins. Structures proximal to the point where the deficiency begins remain essentially intact. This group includes amelia (absent of all or most of a limb) and terminal transverse deficiencies ***Amelia*** – Complete absence of one or more limbs. The most proximal portion of the proximal limb segment (proximal humerus or femur) may

be present, and this form is sometimes called peromelia. In amelia, the upper limb (humerus, radius, ulna, wrist, hand and fingers) or lower limb (femur, tibia, fibula, ankle, foot, and toes) is absent. Amelia is very rare.

Transverse terminal deficiency (reduction) – Complete absence of the distal structures of the arm with the proximal structures intact. This term usually refers to deficiency below the elbow, or complete absence of the distal structures of the leg with the proximal structures intact.

Terms in medical records that can indicate a transverse deficiency include the following:

- Acheiria – Absence of a hand
- Adactyly – Absence of digits (fingers or toes), excluding isolated missing thumb (see below)
- Aphalangia – Absence of phalanges. Fingers and toes typically contain 3 phalanges each. The thumb (pollex) and big toe (hallux) typically contain 2 phalanges.

Congenital amputation, type not specified

Longitudinal limb deficiency (reduction). Absence of limb components in the upper or lower limbs in parallel with the long axis of limbs. They involve the lower leg (below the knee) or the distal arm (below the elbow). These deficiencies are described as preaxial if the affected bone(s) are on the side of the thumb or first toe; postaxial if on the fifth finger side/ on the fifth toe side, or central parts of hands and feet. Longitudinal limb deficiencies, compared to transverse deficiencies, are more frequently associated with genetic syndromes and multiple malformation complexes (e.g., preaxial deficiencies in the VACTERL association).

- ***Preaxial limb deficiency***. Absence of preaxial limb components, including missing thumb with or without missing or hypoplastic radius (upper limb), and missing first toe with or without missing or hypoplastic tibia (lower limb).
- ***Postaxial limb deficiency***. Absence of postaxial limb components, including missing 5th (or 4th-5th) fingers with or without missing or hypoplastic ulna (upper limb), and missing 5th (or 4th-5th) toes with or without missing or hypoplastic fibula (lower limb).
- ***Axial deficiency, or split hand/split foot malformation, SHSF*.** Absence of one or more of the central rays of the hands or feet (or both). In the hand, the finding is of missing or severely hypoplastic second through fourth fingers and their associated metacarpal bones. In the feet, the finding is of missing or severely hypoplastic second through fourth toes and their associated metatarsal bones.
    - **Note:** Older terms (e.g., lobster claw hand) are now discouraged in favor of SH/SF malformation.

Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the upper limb or lower limb

	with all or part of the distal segment present. For example, the radius, ulna, and distal humerus are missing with presence of a hand that attaches to the remnant of the humerus. • **Note:** Phocomelia was term originally used for intercalary limb deficiencies. This term is now discouraged. ***Unspecified deficiency (reduction defect) of the upper limb or lower limb not elsewhere coded or of unspecified type*** – Complete or partial absence of the upper limb or lower limb that does not fall within the above categories or for which there is no specific description. Additional terms that point to limb deficiencies, also now discouraged in favor of more descriptive terms • Ectrodactly. Used in the past to indicate longitudinal or sometimes transverse limb deficiencies involving fingers or toes. • Ectromelia. Similar term, indicating missing limb segments. ***Syndromes with a limb deficiency*.** Limb deficiency is a cardinal (strongly associated) or common finding in many genetic and non-genetic syndromes. Examples of genetic syndromes in which limb deficiencies are cardinal findings include DK phocomelia syndrome, Limb mammary syndrome, Roberts-SC phocomelia syndrome, Oromandibular-Limb Hypogenesis spectrum, Holt-Oram syndrome and TAR (thrombocytopenia with absent radius) syndrome, among others. Non-genetic conditions with limb deficiencies include the include thalidomide embryopathy and the amnion rupture or amniotic band sequence (considered a disruption and not a developmental disorder). Although a limb deficiency is a cardinal feature of these and other syndromic presentations, it is crucial to identify and characterize the specific type of limb deficiency for appropriate coding, classification, and analysis.
ICD-10-CM Codes	**Q71.00** Congenital complete absence of unspecified upper limb **Q71.01** Congenital complete absence of right upper limb **Q71.02** Congenital complete absence of left upper limb **Q71.03** Congenital complete absence of upper limb, bilateral **Q71.10** Congenital absence of unspecified upper arm and forearm with hand present **Q71.11** Congenital absence of right upper arm and forearm with hand present **Q71.12** Congenital absence of left upper arm and forearm with hand present **Q71.13** Congenital absence of upper arm and forearm with hand present, bilateral **Q71.20** Congenital absence of both forearm and hand, unspecified upper limb **Q71.21** Congenital absence of both forearm and hand, right upper limb **Q71.22** Congenital absence of both forearm and hand, left upper limb **Q71.23** Congenital absence of both forearm and hand, bilateral **Q71.30** Congenital absence of unspecified hand and finger **Q71.31** Congenital absence of right hand and finger **Q71.32** Congenital absence of left hand and finger

Q71.33 Congenital absence of hand and finger, bilateral
Q71.40 Longitudinal reduction defect of unspecified radius
Q71.41 Longitudinal reduction defect of right radius
Q71.42 Longitudinal reduction defect of left radius
Q71.43 Longitudinal reduction defect of radius, bilateral
Q71.50 Longitudinal reduction defect of unspecified ulna
Q71.51 Longitudinal reduction defect of right ulna
Q71.52 Longitudinal reduction defect of left ulna
Q71.53 Longitudinal reduction defect of ulna, bilateral
Q71.60 Lobster-claw hand, unspecified hand
Q71.61 Lobster-claw right hand
Q71.62 Lobster-claw left hand
Q71.63 Lobster-claw hand, bilateral
Q71.891 Other reduction defects of right upper limb
Q71.892 Other reduction defects of left upper limb
Q71.893 Other reduction defects of upper limb, bilateral
Q71.899 Other reduction defects of unspecified upper limb
Q71.90 Unspecified reduction defect of unspecified upper limb
Q71.91 Unspecified reduction defect of right upper limb
Q71.92 Unspecified reduction defect of left upper limb
Q71.93 Unspecified reduction defect of upper limb, bilateral
Q72.00 Congenital complete absence of unspecified lower limb
Q72.01 Congenital complete absence of right lower limb
Q72.02 Congenital complete absence of left lower limb
Q72.03 Congenital complete absence of lower limb, bilateral
Q72.10 Congenital absence of unspecified thigh and lower leg with foot present
Q72.11 Congenital absence of right thigh and lower leg with foot present
Q72.12 Congenital absence of left thigh and lower leg with foot present
Q72.13 Congenital absence of thigh and lower leg with foot present, bilateral
Q72.20 Congenital absence of both lower leg and foot, unspecified lower limb
Q72.21 Congenital absence of both lower leg and foot, right lower limb
Q72.22 Congenital absence of both lower leg and foot, left lower limb
Q72.23 Congenital absence of both lower leg and foot, bilateral
Q72.30 Congenital absence of unspecified foot and toe(s)
Q72.31 Congenital absence of right foot and toe(s)
Q72.32 Congenital absence of left foot and toe(s)
Q72.33 Congenital absence of foot and toe(s), bilateral
Q72.40 Longitudinal reduction defect of unspecified femur
Q72.41 Longitudinal reduction defect of right femur
Q72.42 Longitudinal reduction defect of left femur
Q72.43 Longitudinal reduction defect of femur, bilateral
Q72.50 Longitudinal reduction defect of unspecified tibia
Q72.51 Longitudinal reduction defect of right tibia
Q72.52 Longitudinal reduction defect of left tibia
Q72.53 Longitudinal reduction defect of tibia, bilateral
Q72.60 Longitudinal reduction defect of unspecified fibula
Q72.61 Longitudinal reduction defect of right fibula

	Q72.62 Longitudinal reduction defect of left fibula **Q72.63** Longitudinal reduction defect of fibula, bilateral **Q72.70** Split foot, unspecified lower limb **Q72.71** Split foot, right lower limb **Q72.72** Split foot, left lower limb **Q72.73** Split foot, bilateral **Q72.891** Other reduction defects of right lower limb **Q72.892** Other reduction defects of left lower limb **Q72.893** Other reduction defects of lower limb, bilateral **Q72.899** Other reduction defects of unspecified lower limb **Q72.90** Unspecified reduction defect of unspecified lower limb **Q72.91** Unspecified reduction defect of right lower limb **Q72.92** Unspecified reduction defect of left lower limb **Q72.93** Unspecified reduction defect of lower limb, bilateral **Q73.0** Congenital absence of unspecified limb(s) **Q73.1** Phocomelia, unspecified limb(s) **Q73.8** Other reduction defects of unspecified limb(s) **Limb Reduction (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2148 See below for the organization of ICD10CM limb deficiency codes by type and laterality.
SNOMED CT Codes	**Limb Reduction (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2147
Additional Information	
Exclusions	• Shortened arms, forearms, hands, upper and/or lower legs, feet, toes or fingers that have all of their component parts, including those that are part of a generalized chondrodystrophy, osteodystrophy, or dwarfism • Hypoplastic nails • Polydactyly and syndactyly without limb reduction — these are not true limb deficiencies but are commonly miscoded as such • Prenatal finding of limb deficiency that is less severe than amelia (absence of an entire limb) without postnatal confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential limb deficiency: **ICD-10-CM:** **Q74.8** Other specified congenital malformations of limb(s) **Q74.9** Unspecified congenital malformation of limb(s) **Q71.811** Congenital shortening of right upper limb **Q71.812** Congenital shortening of left upper limb **Q71.813** Congenital shortening of upper limb, bilateral **Q71.819** Congenital shortening of unspecified upper limb **Q72.811** Congenital shortening of right lower limb **Q72.812** Congenital shortening of left lower limb **Q72.813** Congenital shortening of lower limb, bilateral **Q72.819** Congenital shortening of unspecified lower limb

O35.GXX0–O35.GXX9 Maternal care for other (suspected) fetal abnormality and damage, fetal upper extremities anomalies

O35.HXX0–O35.HXX9 Maternal care for other (suspected) fetal abnormality and damage, fetal lower extremities anomalies

SNOMED-CT:

720430002 Acrofacial dysostosis Rodriguez type (disorder)
720414005 Acrorenal mandibular syndrome (disorder)
34748004 Adams-Oliver syndrome (disorder)
1145447009 Aplasia of bone of extremity (disorder)
1145448004 Aplasia of bone of lower limb (disorder)
1145451006 Aplasia of bone of upper limb (disorder)
733030003 Congenital hypoplasia of ulna and split foot syndrome (disorder)
721880009 Congenital microgastria with limb reduction defect syndrome (disorder)
1172589000 Congenital omphalocele, diaphragmatic hernia, cardiovascular anomalies, radial ray defect syndrome (disorder)
897586009 Congenital radial deviation of bilateral fingers (disorder)
724619002 Congenital radial deviation of finger (disorder)
732250002 Craniosynostosis fibular aplasia syndrome (disorder)
722429003 Distal limb deficiency with micrognathia syndrome (disorder)
719021005 DK phocomelia syndrome (disorder)
699867001 Duane-radial ray syndrome (disorder)
1208338004 Dysraphism, cleft lip and palate, limb reduction defect syndrome (disorder)
1230098009 Femur fibula ulna complex (disorder)
783156008 Fibular aplasia, tibial campomelia, oligo-syndactyly syndrome (disorder)
721296004 Fuhrmann syndrome (disorder)
766032007 Holoprosencephaly, ectrodactyly, cleft lip, cleft palate syndrome (disorder)
19092004 Holt-Oram syndrome (disorder)
716106000 Limb body wall complex (disorder)
721972001 Limb mammary syndrome (disorder)
68551007 Limb reduction-ichthyosis syndrome (disorder)
715533002 Microcephaly, microphthalmia, ectrodactyly of lower limbs and prognathism syndrome (disorder)
85589009 Radial aplasia-thrombocytopenia syndrome (disorder)
783137003 Radial deficiency, tibial hypoplasia syndrome (disorder)
766765009 Radio-renal syndrome (disorder)
48718006 Roberts-SC phocomelia syndrome (disorder)
770681000 Robin sequence and oligodactyly syndrome (disorder)
726724005 Splenogonadal fusion, limb defect, micrognathia syndrome (disorder)

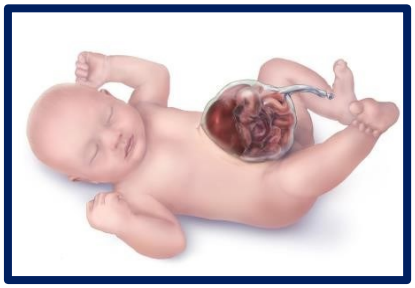
	766821006 Spondyloepimetaphyseal dysplasia, short limb, abnormal calcification syndrome (disorder) **1172635005** Split-foot malformation, mesoaxial polydactyly syndrome (disorder) **205387007** Constriction ring syndrome of lower limb with amputation (disorder) **205817005** Aglossia-adactyly syndrome (disorder) **702313004** Tetra-amelia syndrome (disorder) **715506001** Phocomelia, ectrodactyly, deafness and sinus arrhythmia syndrome (disorder) **715531000** Tibial aplasia and ectrodactyly syndrome (disorder) **716099003** Absence deformity of leg and congenital cataract syndrome (disorder) **716249009** Tetraamelia with multiple malformation syndrome (disorder) **719685004** Absent thumb with short stature and immunodeficiency syndrome (disorder) **720498007** Aphalangy and syndactyly with microcephaly syndrome (disorder) **720952001** Aplasia of fibula and ectrodactyly syndrome (disorder) **723611008** Split hand, split foot malformation with sensorineural hearing loss syndrome (disorder) **732927000** Split hand, obstructive uropathy, spina bifida, diaphragmatic defect syndrome (disorder) **733068001** Absent tibia, polydactyly, arachnoid cyst syndrome (disorder) **733118006** Aphalangy, hemivertebra, urogenital, intestinal dysgenesis syndrome (disorder) **763743003** Intellectual disability, spasticity, ectrodactyly syndrome (disorder) **771177009** Ectrodactyly polydactyly syndrome (disorder) **771264005** Absent radius, anogenital anomalies syndrome (disorder) **890221004** Acrocardiofacial syndrome (disorder)

Additional Information	Summary clinical notes may include terminology that is outdated or confusing. Historically, terms such as "phocomelia" or "ectrodactyly" have been used or misused for different types of limb deficiencies. It is crucial to look for a complete description of all structures that are present and absent to verify the diagnosis. Often the radiology report provides the clearest window into the condition. IN some cases of transverse limb deficiency (reduction) , rudimentary or nubbin toes may be present at the distal end of the truncated limb. Their presence alone does not change the classification of the defect as transverse. Joint contractures or clubfoot/clubhand are commonly seen in association with longitudinal limb deficiencies. Thalidomide use during early pregnancy is known to cause intercalary deficiency as well as other limb deficiency types, including longitudinal deficiency. Limb deficiency, typically preaxial, is a common component of the VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia. Oromandibular-Limb Hypogenesis spectrum, which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), and sixth and seventh cranial nerve palsies (Moebius sequence). Limb deficiencies (reductions) are usually recognizable on physical examination of the neonate. However, precise morphologic diagnosis may only be distinguished by imaging, surgery, or autopsy. While limb deficiency may be identified by prenatal ultrasound, these cases should not be included in birth defects surveillance data without postnatal confirmation.

Organization of ICD10CM Codes by Type of Limb Deficiency

Morphological Group	Laterality	Upper Limb (Q71)	Lower Limb (Q72)	Unspecified Limb (Q73)
Intercalary	Right	**Q71.11**	**Q72.11**	—
(formerly Phocomelia)	Left	**Q71.12**	**Q72.12**	—
	Bilateral	**Q71.13**	**Q72.13**	—
	Unspecified	**Q71.10**	**Q72.10**	**Q73.1**
Transverse Terminal	Right	**Q71.31**	**Q72.31**	—
(Total absence/Aplasia)	Left	**Q71.32**	**Q72.32**	—
	Bilateral	**Q71.33**	**Q72.33**	—
	Unspecified	**Q71.30**	**Q72.30**	**Q73.0**
Preaxial	Right	**Q71.41** (Rad)	**Q72.51** (Tib)	—
(Radial or Tibial)	Left	**Q71.42**	**Q72.52**	—
	Bilateral	**Q71.43**	**Q72.53**	—
	Unspecified	**Q71.40**	**Q72.50**	**Q73.8**
Postaxial	Right	**Q71.51** (Uln)	**Q72.61** (Fib)	—
(Ulnar or Fibular)	Left	**Q71.52**	**Q72.62**	—
	Bilateral	**Q71.53**	**Q72.63**	—
	Unspecified	**Q71.50**	**Q72.60**	**Q73.8**
Axial	Right	**Q71.61**	**Q72.71**	—
(Central/SHFM)	Left	**Q71.62**	**Q72.72**	—
	Bilateral	**Q71.63**	**Q72.73**	—
	Unspecified	**Q71.60**	**Q72.70**	**Q73.8**
Unspecified	—	**Q71.90**	**Q72.90**	**Q73.9**

Omphalocele

Omphalocele *Musculoskeletal*	
Description	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. 
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	41
CDC/BPA Codes	**756.70** Omphalocele
NBDPN Case Definition	
Tier 1 Surveillance	**Confirmed:** One of the following criteria must be met: • Postnatal physical examination indicates herniation of abdominal organs into the umbilical cord and no subsequent imaging, surgery, or autopsy refutes the diagnosis. • Postnatal imaging, surgery or autopsy demonstrates presence of omphalocele. **Accepted:** One of the following criteria must be met: • Prenatal ultrasound documents omphalocele [in a pregnancy that results in stillbirth, spontaneous loss, or elective termination] and no information from further postnatal evaluation is available. • Postnatal diagnosis of omphalocele without confirmatory exam, imaging, surgery, or autopsy information.
Tier 2 Surveillance	**Confirmed:** One of the following criteria must be met: • Presence of a coded or written diagnosis of omphalocele assigned postnatally within the first three months after birth by an inpatient setting, as described in the "Diagnoses Included for Case Classification of Omphalocele" section below. • Presence of a coded or written diagnosis of omphalocele assigned postnatally, as described in the "Diagnoses Included for Case Classification of Omphalocele" section below, AND death of the infant within the first year of life.

	Accepted: The following criterion must be met: • Presence of a coded or written diagnosis of omphalocele assigned postnatally, as described in the "Diagnoses Included for Case Classification of Omphalocele" section below, without the presence of confirmatory information.
Diagnoses Included for Case Classification of Omphalocele	
Inclusions	• Exomphalos • Omphalocele
ICD-10-CM Codes	**Q79.2** Exomphalos **Omphalocele (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2570
SNOMED CT Codes	**Omphalocele (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2569
Additional Information	
Exclusions	• Gastroschisis • Umbilical hernia • Cloacal Exstrophy
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential omphalocele: **ICD-10-CM:** **Q79.59** Other congenital malformations of abdominal wall **Q79.8** Other congenital malformations of musculoskeletal system **Q79.9** Congenital malformation of musculoskeletal system, unspecified **O35.FXX0–O35.FXX9** Maternal care for other (suspected) fetal abnormality and damage, fetal musculoskeletal anomalies of trunk **SNOMED-CT:** **363024001** Congenital anomaly of abdomen (disorder) **253818009** Congenital malformation of anterior abdominal wall (disorder) **81780002** Beckwith-Wiedemann syndrome (disorder) **716230005** Shprintzen Goldberg omphalocele syndrome (disorder) **719408007** Lethal omphalocele with cleft palate syndrome (disorder) **770900000** Familial omphalocele syndrome with facial dysmorphism (disorder) **1172589000** Congenital omphalocele, diaphragmatic hernia, cardiovascular anomalies, radial ray defect syndrome (disorder)
Additional Information	The distinction between omphalocele and gastroschisis is important because they have different etiologies and different implications for treatment and long-term survival. In omphalocele, abdominal organs herniate through the umbilicus into the umbilical cord. There is no bridge of skin between the abdominal wall defect and the umbilicus and cord. While the herniating organs are covered by a protective membrane, this may rupture before, during, or after delivery.

	In gastroschisis, the umbilicus and cord are normal and separated from the abdominal wall defect by a small bridge of skin. The herniating organs are not covered by a protective membrane. However, they may appear matted and covered by a thick fibrous material as a result of prolonged exposure to amniotic fluid in utero. Omphalocele is one of the defects reported as part of the Omphalocele-Exstrophy-Imperforate Anus-Spina Bifida (OEIS) complex. Maternal serum alpha-fetoprotein (MSAFP) and/or amniotic fluid alpha-fetoprotein (AFAFP) may be elevated with omphalocele. However, these screening tests alone are not sufficient to diagnose the condition. In contrast to omphalocele, umbilical hernias are completely covered by normal skin. It may be difficult to distinguish gastroschisis from omphalocele on prenatal ultrasound, and the terms sometimes are used interchangeably.

Chromosomal

Deletion 22 q11

Deletion 22 q11 *Chromosomal*	
Description	Chromosome abnormality resulting from genomic microdeletions within a critical region on the long arm of chromosome 22 (22q11.2).
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	61
CDC/BPA Codes	**758.37** Deletion in band 11 of long arm of chromosome 22
NBDPN Case Definition	

Tier 1 Surveillance	**Confirmed:**
	The following criterion must be met:
	• Prenatal or postnatal cytogenetic or molecular-cytogenetic testing demonstrates deletion at the 22q11.2 site using one of the following methods:
	  ○ Chromosomal microarray analysis (CMA /array-CGH).
	  ○ Fluorescence in situ hybridization (FISH) with probe targeted to the 22q11.2 site.
	  ○ Multiplex ligation-dependent probe amplification (MLPA) testing.
	Accepted:
	The following criterion must be met:
	• Prenatal or postnatal diagnosis of Deletion 22q11 without confirmatory laboratory information.

Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of Deletion 22q11 assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Deletion 22q11" section below.

Diagnoses Included for Case Classification of Deletion 22q11	
Inclusions	• Deletion 22q11.2 syndrome • Chromosome deletion 22q11.2 • DiGeorge syndrome with chromosome 22q11.2 deletion • Thymic aplasia syndrome with chromosome 22q11.2 deletion • Velo-cardio-facial (VCF) syndrome with chromosome 22q11.2 deletion • Conotruncal anomaly face (CTAF) syndrome with chromosome 22q11.2 deletion • Cayler cardiofacial (asymmetric crying facies) syndrome with chromosome 22q11.2 deletion • Shprintzen syndrome with chromosome 22q11.2 deletion • Sedlackova (velofacial hypoplasia) syndrome with chromosome 22q11.2 deletion • Takao syndrome with chromosome 22q11.2 deletion
ICD-10-CM Codes	**Q93.81** Velo-cardio-facial syndrome
SNOMED CT Codes	**767263007** 22q11.2 deletion syndrome (disorder)
Additional Information	
Exclusions	• Named phenotypes without cytogenetic abnormalities • Cases identified only by prenatal screening (e.g., NIPT) • TBX1 mutations without cytogenetic abnormalities • Deletion 22q13.3 • Duplication 22q11.2 • Shprintzen-Goldberg syndrome
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Deletion 22q11: **ICD-10-CM:** **D82.1** Di George's syndrome **O35.10X0–O35.10X9** Maternal care for (suspected) chromosomal abnormality in fetus, unspecified **O35.19X0–O35.19X9** Maternal care for (suspected) chromosomal abnormality in fetus, other chromosomal abnormality **SNOMED-CT:** **19550003** 22q partial monosomy (disorder) **726399005** Deletion of part of chromosome 22 (disorder) **LOINC:** **82246-0** Chromosome region 22q11.2 deletion and duplication mutation analysis in Blood or Tissue by FISH **82245-2** Chromosome region 22q11.2 deletion and duplication mutation analysis in Amniotic fluid or Chorionic villus sample by FISH

Additional Information	The deletion 22q11.2 syndrome phenotype can include cardiac abnormalities, abnormal or dysmorphic facial features, thymic aplasia, cleft palate or velopharyngeal insufficiency, or hypocalcemia due to hypoparathyroidism; the "CATCH" acronym appeared in the literature previously to describe these cardinal features, but this term is no longer used. Chromosome 22q11.2 deletions can be found with any of these features in isolation, and is sometimes not diagnosed until adulthood, e.g., in subtly affected parents of children with deletion 22q11.2 syndrome phenotypes or defects. The term "DiGeorge syndrome" was used originally (before 22q11.2 deletions were described) for children with the combination of thymic and parathyroid defects; the ICD-9-CM code 279.11 or ICD-10CM code D82.1 is sometimes still found in medical records with this diagnosis, but should be used in combination with the chromosomal codes listed above for individuals with documented 22q11.2 deletions. "Atypical" smaller 22q11.2 deletions seen on chromosomal microarrays are often labelled with specific letters (e.g., "C-D" deletion) or numbers describing the chromosomal loci.

Trisomy 13

Trisomy 13 *Chromosomal*	
Description	The presence of three copies of all or a large part of chromosome 13.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	44
CDC/BPA Codes	**758.10** Trisomy 13 - karyotype documented **758.11** Trisomy D, NOS **758.12** Translocation trisomy 13 **758.13** Translocation trisomy D, NOS **758.19** Trisomy 13, NOS
NBDPN Case Definition	

Tier 1 Surveillance	**Confirmed:**
	The following criterion must be met:
	• Prenatal or postnatal cytogenetic or molecular-cytogenetic testing demonstrates complete, partial, or mosaic Trisomy 13 using one of the following methods:
	  ○ Karyotype analysis.
	  ○ Chromosomal microarray analysis (CMA /array-CGH).
	  ○ Fluorescence in situ hybridization (FISH).
	  ○ Multiplex ligation-dependent probe amplification (MLPA) testing.
	Accepted:
	The following criterion must be met:
	• Prenatal or postnatal diagnosis of Trisomy 13 without confirmatory laboratory information.

Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of Trisomy 13 assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Trisomy 13" section below.

Diagnoses Included for Case Classification of Trisomy 13	
Inclusions	• Patau syndrome • Mosaic Patau syndrome • Mosaic trisomy 13 • Translocation Patau syndrome • Translocation trisomy 13 • Trisomy 13, not otherwise specified • Trisomy D1, not otherwise specified
ICD-10-CM Codes	**Q91.4** Trisomy 13, nonmosaicism (meiotic nondisjunction) **Q91.5** Trisomy 13, mosaicism (mitotic nondisjunction) **Q91.6** Trisomy 13, translocation **Q91.7** Trisomy 13, unspecified **Trisomy 13 (Disorders) (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2503
SNOMED CT Codes	**Trisomy 13 (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2502 **Fetal Trisomy 13 (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2504
Additional Information	
Exclusions	• Cases identified only by prenatal screening (e.g., NIPT) • Balanced translocations involving chromosome 13
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Trisomy 13: **ICD-10-CM:** **Q92.9** Trisomy and partial trisomy of autosomes, unspecified **O35.11X0–O35.11X9** Maternal care for (suspected) chromosomal abnormality in fetus, Trisomy 13 **SNOMED-CT:** **17760001** Anomaly of chromosome pair 13 (disorder) **716091000** Holoprosencephaly and postaxial polydactyly syndrome (disorder) **270521004** Trisomy and partial trisomy of autosome (disorder) **LOINC:** **77013-1** Fetal Chromosome 13 trisomy [Presence] based on Plasma cell-free DNA by Sequencing **75979-5** Fetal Chromosome 13 trisomy [Interpretation] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA Qualitative **75981-1** Fetal Chromosome 13 trisomy [Presence] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA **75975-3** Fetal Chromosome 13+18+21 trisomy [Presence] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA

Additional Information	When the two copies of chromosome 13 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 13 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+13 or 47,XY,+13. This is the most common type of trisomy 13 and is associated with advanced maternal age, particularly of 35 years or greater. Translocation trisomy 13 occurs when two separate copies of chromosome 13 are present, but a third copy of part of chromosome 13 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 13. Mosaic trisomy 13 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 13. In this instance, the karyotype is written as 46,XY/47,XY,+13, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 13 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis or preferably postnatal blood or tissue samples. Approximately 80% of infants with trisomy 13 do not survive beyond the first month of life. Major malformations associated with trisomy 13 may include holoprosencephaly, microcephaly, meningomyelocele, cleft lip and/or palate, microphthalmia, retinal dysplasia, polydactyly, heart defects (most commonly a VSD), omphalocele, and genitourinary defects, among others. Among children who survive the newborn period, severe developmental delay is virtually always present as may be deafness, visual impairment, minor motor seizures, and apneic spells. Infants with mosaic trisomy 13 may be less severely affected with variable degrees of developmental delay and longer survival. Infants with partial trisomy for the proximal segment of chromosome 13 (13pter→q14) exhibit a nonspecific pattern of abnormalities with near-normal survival. Approximately 25% of infants with partial trisomy for the distal segment of chromosome 13 (13q14→qter) die during early postnatal life. Children who survive exhibit severe developmental delay and specific abnormalities. Major malformations that occur with trisomy 13 in the same infant should be coded separately, as their presence varies among affected individuals.

Trisomy 18

Trisomy 18 *Chromosomal*	
Description	The presence of three copies of all or a large part of chromosome 18.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	46
CDC/BPA Codes	**758.20** Trisomy 18 - karyotype documented **758.21** Trisomy E, NOS **758.22** Translocation trisomy 18 **758.23** Translocation trisomy E, NOS **758.29** Trisomy 18, NOS
NBDPN Case Definition	

Tier 1 Surveillance	**Confirmed:**
	The following criterion must be met:
	• Prenatal or postnatal cytogenetic or molecular-cytogenetic testing demonstrates complete, partial, or mosaic Trisomy 18 using one of the following methods:
	  ○ Karyotype analysis.
	  ○ Chromosomal microarray analysis (CMA /array-CGH).
	  ○ Fluorescence in situ hybridization (FISH).
	  ○ Multiplex ligation-dependent probe amplification (MLPA) testing.
	Accepted:
	The following criterion must be met:
	• Prenatal or postnatal diagnosis of Trisomy 18 without confirmatory laboratory information.

Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of Trisomy 18 assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Trisomy 18" section below.

Diagnoses Included for Case Classification of Trisomy 18	
Inclusions	• Edwards syndrome • Mosaic Edwards syndrome • Mosaic trisomy 18 • Translocation Edwards syndrome • Translocation trisomy 18 • Trisomy 18, not otherwise specified
ICD-10-CM Codes	**Q91.0** Trisomy 18, nonmosaicism (meiotic nondisjunction) **Q91.1** Trisomy 18, mosaicism (mitotic nondisjunction) **Q91.2** Trisomy 18, translocation **Q91.3** Trisomy 18, unspecified **Trisomy 18 (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2521 **Fetal Trisomy 18 (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2520
SNOMED CT Codes	**Trisomy 18 (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2518 **Fetal Trisomy 18 (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2519
Additional Information	
Exclusions	• Named phenotypes without cytogenetic abnormalities • Cases identified only by prenatal screening (e.g., NIPT) • Balanced translocations involving chromosome 18
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Trisomy 18: **ICD-10-CM:** **Q92.9** Trisomy and partial trisomy of autosomes, unspecified **O35.12X0–O35.12X9** Maternal care for (suspected) chromosomal abnormality in fetus, Trisomy 18 **SNOMED-CT:** **59033006** Anomaly of chromosome pair 18 (disorder) **270521004** Trisomy and partial trisomy of autosome (disorder) **LOINC:** **77012-3** Fetal Chromosome 18 trisomy [Presence] based on Plasma cell-free DNA by Sequencing **75978-7** Fetal Chromosome 18 trisomy [Interpretation] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA Qualitative **75982-9** Fetal Chromosome 18 trisomy [Presence] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA **75975-3** Fetal Chromosome 13+18+21 trisomy [Presence] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA
Additional Information	When the two copies of chromosome 18 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 18 will be present in the fetus. In this instance, the karyotype is written as

	47,XX,+18 or 47,XY,+18. This is the most common type of trisomy 18 and is associated with advanced maternal age, particularly of 35 years or greater. Translocation trisomy 18 occurs when two separate copies of chromosome 18 are present, but a third copy of part of chromosome 18 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 18. Mosaic trisomy 18 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 18. In this instance, the karyotype is written as 46,XY/47,XY,+18, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 18 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis or preferably postnatal blood or tissue samples. Most pregnancies affected with trisomy 18 result in spontaneous abortion. Approximately 50% of liveborn infants with trisomy 18 do not survive beyond the first week of life. Only 5% to 10% survive beyond the first year of life. Major malformations associated with trisomy 18 may include microcephaly, micrognathia, cleft lip and/or palate, heart defects, omphalocele, and renal defects, among others. Minor anomalies associated with trisomy 18 may include low-set malformed auricles (external ears), overlapping of the index and fifth fingers over the third and fourth fingers, absent distal crease on the fifth finger, hirsutism (excess hair) of the forehead and back, lateral deviation of the hands, a hypoplastic thumb, a single transverse palmar crease, and rocker-bottom feet, among others. Developmental delay is virtually always present, as may be hypertonicity, a weak cry, growth retardation, hypoplasia of skeletal muscle and subcutaneous fat, and clenched hands. Infants with mosaic trisomy 18 may be less severely affected, with variable degrees of developmental delay and longer survival. Infants with trisomy of only the short arm of chromosome 18 (partial trisomy 18) exhibit a nonspecific pattern of abnormalities with mild to no developmental delay. Infants with trisomy of the short arm, centromere, and proximal third of the long arm of chromosome 18 exhibit features of trisomy 18 but not the entire spectrum of abnormalities. Infants with trisomy of only one-third to one-half of the long arm of chromosome 18 exhibit features of trisomy 18 but have longer survival and less severe developmental delays. Major malformations that occur with trisomy 18 in the same infant should be coded separately, as their presence varies among affected individuals.

Trisomy 21 (Down syndrome)

Trisomy 21 (Down syndrome) *Chromosomal*	
Description	The presence of three copies of all or a large part of chromosome 21. 
NBDPN Data Management	
NBDPN Defect Tier	Core
NBDPN Birth Defect Case Definition Code	45
CDC/BPA Codes	**758.00** Trisomy 21 - karyotype documented **758.01** Trisomy G, NOS - karyotype documented **758.02** Translocation trisomy 21 **758.03** Translocation trisomy G, NOS **758.04** Mosaic trisomy 21 syndrome **758.09** Trisomy 21, NOS
NBDPN Case Definition	

Tier 1 Surveillance	**Confirmed:**
	One of the following criteria must be met:
	• Prenatal or postnatal cytogenetic or molecular-cytogenetic testing demonstrates complete, partial, or mosaic Trisomy 21 using one of the following methods:
	  ○ Karyotype analysis
	  ○ Chromosomal microarray analysis (CMA /array-CGH).
	  ○ Fluorescence in situ hybridization (FISH).
	  ○ Multiplex ligation-dependent probe amplification (MLPA) testing.
	• Postnatal diagnosis of Trisomy 21 without confirmatory laboratory information.
	Accepted:
	The following criterion must be met:
	• Prenatal diagnosis of Trisomy 21 without confirmatory laboratory information.

Tier 2 Surveillance	**Confirmed:** The following criterion must be met[1,2]: • Presence of a coded or written diagnosis of Trisomy 21 assigned postnatally, as described in the "Diagnoses Included for Case Classification of Trisomy 21" section below. **Accepted:** The following criterion must be met: • Presence of a coded or written diagnosis of Trisomy 21 assigned prenatally, as described in the "Diagnoses Included for Case Classification of Trisomy 21" section below.
Diagnoses Included for Case Classification of Trisomy 21	
Inclusions	• Down syndrome • Mosaic Down syndrome • Mosaic trisomy 21 • Translocation Down syndrome • Translocation trisomy 21 • Trisomy 21, not otherwise specified
ICD-10-CM Codes	**Q90.0** Trisomy 21, nonmosaicism (meiotic nondisjunction) **Q90.1** Trisomy 21, mosaicism (mitotic nondisjunction) **Q90.2** Trisomy 21, translocation **Q90.9** Down syndrome, unspecified **Down Syndrome (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2125 **Fetal Down Syndrome (ICD10CM) OID:** 2.16.840.1.113762.1.4.1146.2127
SNOMED CT Codes	**Down Syndrome (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2124 **Fetal Down Syndrome (SNOMED) OID:** 2.16.840.1.113762.1.4.1146.2126
Additional Information	
Exclusions	• Cases identified only by prenatal screening (e.g., NIPT) • Balanced translocations involving chromosome 21 • "Downs facies" without associated trisomy 21
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Trisomy 21: **ICD-10-CM:** **Q92.9** Trisomy and partial trisomy of autosomes, unspecified **O35.13X0–O35.13X9** Maternal care for (suspected) chromosomal abnormality in fetus, Trisomy 21 **SNOMED-CT:** **70156005** Anomaly of chromosome pair 21 (disorder) **270521004** Trisomy and partial trisomy of autosome (disorder) **248199009** Down's facies (finding)

	415683005 Fetal complete trisomy 21 syndrome suspected (situation) **227035005** 21q partial distal trisomy (disorder) **228050004** 21q partial trisomy (disorder) **371045000** Translocation Down syndrome (disorder) **LOINC:** **43306-0** Chromosome 21 trisomy [Percentile] by Cytogenetics **77011-5** Fetal Chromosome 21 trisomy [Presence] based on Plasma cell-free DNA by Sequencing **21771-1** Chromosome 21 trisomy [Presence] in Blood or Tissue by Cytogenetics **75976-1** Fetal Chromosome 21 trisomy [Interpretation] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA Qualitative **75983-7** Fetal Chromosome 21 trisomy [Presence] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA **75975-3** Fetal Chromosome 13+18+21 trisomy [Presence] based on Plasma cell-free DNA by Dosage of chromosome-specific cfDNA
Additional Information	When the two copies of chromosome 21 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 21 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+21 or 47,XY,+21. This is the most common type of trisomy 21 and is associated with advanced maternal age, particularly of 35 years or greater. Translocation trisomy 21 occurs when two separate copies of chromosome 21 are present, but a third copy of part of chromosome 21 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 21. Mosaic trisomy 21 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 21. In this instance, the karyotype is written as 46,XY/47,XY,+21, for example. Because the placenta may contain mosaic cell lines not present in the fetus, mosaic trisomy 21 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis or preferably postnatal blood or tissue samples. Infants with Down syndrome have a typical appearance and other characteristics, including decreased muscle tone (hypotonia), a weak startle (Moro) reflex, hyperflexible joints, a flattened facial profile, upward slanting eyes, abnormally shaped external ears (auricles), loose skin on the back of the neck, dysplasia of the pelvic bones, incurving of the fifth finger (clinodactyly), and a single transverse crease in the palm of the hand (Simian crease). Developmental delay is virtually always present. Major malformations associated with Down syndrome include heart defects (most notably atrioventricular septal defects), gastrointestinal defects, and vertebral abnormalities, among others. Major malformations that occur with Down syndrome in the same infant should be coded separately, as their presence may varies among affected individuals. Mongolism is an outdated term for Down syndrome.

[1] Salemi JL, Rutkowski RE, Tanner JP, Matas JL, Kirby RS. Identifying algorithms to improve the accuracy of unverified diagnosis codes for birth defects. Public Health Rep. 2018;133(3):303-310. doi:10.1177/0033354918763168.
[2] Salemi JL, Tanner JP, Sampat D, Anjohrin SB, Correia JA, Watkins SM, et al. The accuracy of hospital discharge diagnosis codes for major birth defects: evaluation of a statewide registry with passive case ascertainment. J Public Health Manag Pract. 2016;22(3):E9-E19. doi:10.1097/PHH.0000000000000291.

Turner syndrome

Turner syndrome *Chromosomal*	
Description	A chromosomal disorder in phenotypic females associated with complete or partial absence of one X chromosome in some or all cells. The classic karyotype is 45,X (monosomy X); additional karyotypes include mosaicism (e.g., 45,X/46,XX; 45,X/46,XY), isochromosome Xq (46,X,i(Xq)), ring X chromosome, and partial deletions of Xp or Xq. The phenotype ranges from nonviable hydrops fetalis to a near-typical appearance; common features include short stature, gonadal dysgenesis, and characteristic congenital heart and renal anomalies.
NBDPN Data Management	
NBDPN Defect Tier	Enhanced
NBDPN Birth Defect Case Definition Code	60
CDC/BPA Codes	**758.60** Turner syndrome, unspecified **758.61** Karyotype 45,X **758.62** Mosaic Turner syndrome **758.63** Structurally abnormal X chromosome (isochromosome, ring, deletion) **758.69** Other gonadal dysgenesis
NBDPN Case Definition	

Tier 1 Surveillance	**Confirmed:**
	All of the following criteria must be met:
	• Phenotypic female (or non-viable fetus with female or indeterminate phenotype).
	• Prenatal or postnatal cytogenetic or molecular-cytogenetic testing demonstrates one of the following:
	  ○ 45,X, or a structurally abnormal X chromosome (iso(Xq), ring X, partial Xp deletion) consistent with a Turner karyotype on an appropriate prenatal or postnatal specimen.
	  ○ Mosaicism involving 45,X on a postnatal specimen.
	
	Acceptable testing methods include:
	• Postnatal G-banded karyotype on peripheral blood lymphocytes. Per ACMG laboratory guidance, a minimum of 30 metaphases should be analyzed (detects ≥10% mosaicism at 95% confidence).
	• Karyotype on a second tissue — skin fibroblasts, buccal mucosa cells, or bladder epithelial cells — when clinical suspicion is high but blood karyotype is 46,XX. FISH on buccal mucosa is specifically recommended by the ACMG Turner syndrome

	laboratory guideline for detection of tissue-limited mosaicism not seen in blood. • Fluorescence in situ hybridization (FISH) with X and Y centromeric probes on blood or buccal specimens (also used to detect cryptic Y material that may warrant gonadectomy). • Chromosomal microarray analysis (CMA / array-CGH). Note that CMA may be less sensitive than sequencing or karyotype for low-level mosaicism. • Karyotype, FISH, or CMA on prenatal specimens obtained by amniocentesis or chorionic villus sampling (CVS). **Accepted:** All of the following criteria must be met: • Phenotypic female or indeterminate phenotype. • Prenatal or postnatal diagnosis of Turner syndrome without confirmatory laboratory information.
Tier 2 Surveillance	**Confirmed:** N/A **Accepted:** All of the following criteria must be met: • Phenotypic female or indeterminate phenotype. • Presence of a coded or written diagnosis of Turner syndrome assigned prenatally or postnatally, as described in the "Diagnoses Included for Case Classification of Turner Syndrome" section below.
Diagnoses Included for Case Classification of Turner Syndrome	
Inclusions	• Turner syndrome, including 45,X monosomy • Turner syndrome with mosaicism (45,X / 46,XX; 45,X / 46,XY; 45,X / 47,XXX; 45,X with autosomal translocation; or combinations) • Turner syndrome with a ring X chromosome, 46,X,r(X) • Turner syndrome with an isochromosome Xq, 46,X,i(Xq) • Turner syndrome with partial Xp deletion producing the Turner phenotype • Forms of gonadal dysgenesis with 45,X or 45,X-variant cell line and Turner phenotype
ICD-10-CM Codes	**Q96.0** Karyotype 45, X **Q96.1** Karyotype 46, X iso (Xq) **Q96.2** Karyotype 46, X with abnormal sex chromosome, except iso (Xq) **Q96.3** Mosaicism, 45,X / 46,XX or XY **AND** phenotype is female or indeterminate **Q96.4** Mosaicism, 45,X / other cell line(s) with abnormal sex chromosome **Q96.8** Other variants of Turner's syndrome

	Q96.9 Turner's syndrome, unspecified **O35.14X0 - O35.14X9** Maternal care for (suspected) chromosomal abnormality in the fetus, Monosomy X / Turner syndrome
SNOMED CT Codes	**710008008** Monosomy X (disorder) **710010005** Mosaic Turner syndrome (disorder) **302960008** Mosaicism 45,X / 46,XX (disorder) **710019006** Mosaicism 45,X or other cell line with abnormal sex chromosome (disorder) **205686009** Karyotype 46, X iso (Xq) (disorder) **205687000** Karyotype 46, X with abnormal sex chromosome except iso (Xq) (disorder) **254280007** Turner's phenotype, partial X deletion karyotype (disorder) **254281006** Turner's phenotype — ring chromosome karyotype (disorder) **205684007** Turner's phenotype, karyotype normal (disorder)
Additional Information	
Exclusions	• 46,XY males (including 46,XY/45,X mosaicism) — mixed gonadal dysgenesis in a phenotypic male • Xq24 deletions without Turner phenotype • Distal Xp22.3 deletions (e.g., short-stature homeobox gene region) without Turner phenotype • Noonan syndrome ("male Turner"; RASopathy) — a separate autosomal dominant condition • Prenatal screening findings (e.g., NIPT suggestive of monosomy X) without postnatal karyotype or molecular confirmation • Clinical phenotype without cytogenetic confirmation
Other Codes for Case Ascertainment	Other codes that may be used for case ascertainment but require further review for potential Turner Syndrome: **ICD-10-CM:** **O35.10X0–O35.10X9** Maternal care for (suspected) chromosomal abnormality in fetus, unspecified **O35.19X0–O35.19X9** Maternal care for (suspected) chromosomal abnormality in fetus, other chromosomal abnormality **SNOMED-CT:** **111312006** Anomaly of chromosome X (disorder) **125491000119103** Fetal Turner syndrome (disorder) **LOINC:** **81748-6** Chromosome region Yp11.3 deletion AndOr rearrangement in Blood or Tissue by FISH

Additional Information	Phenotype is highly variable. At one extreme, affected fetuses present with non-immune hydrops (cystic hygroma, pleural effusions, generalized edema) and rarely survive to term. At the other extreme, infants may appear phenotypically typical at birth and be diagnosed later for short stature or primary amenorrhea. FISH performed prenatally can detect 45,X but does not reliably detect structural X abnormalities; postnatal karyotype should be obtained when Turner syndrome is clinically suspected despite negative FISH. Mosaic Turner syndrome identified prenatally should be confirmed postnatally on a specimen obtained from the infant or fetus. Associated congenital anomalies that should be counted separately for surveillance purposes when meeting surveillance case definitions include: • Congenital heart defects (occur in approximately 30% of cases) — bicuspid aortic valve, coarctation of the aorta, hypoplastic left heart, partial anomalous pulmonary venous return. • Renal anomalies (occur in approximately 30% of cases) — horseshoe kidney, duplex collecting system, unilateral renal agenesis. • Lymphedema (residual fetal lymphatic distention): dorsal hand/foot puffiness, neck webbing, low posterior hairline. Etiologies differ by karyotype. The 45,X karyotype arises from a random error of meiosis or early mitosis (non-disjunction). Mosaic karyotypes arise from post-zygotic mitotic errors. Structural X abnormalities and partial deletions are very rarely inherited. The terms Bonnevie-Ullrich syndrome and Ullrich-Turner syndrome are historical synonyms for Turner syndrome.