Appendix 3.2

NBDPN Abstractor's Instructions

Appendix 3.2
NBDPN Abstractor's Instructions

Appendix 3.2
NBDPN Abstractor's Instructions

Format for Birth Defect Descriptions

Defect Name

Description	Description of the defect.
Inclusions	Other names or conditions that should be included in the code for the defect.
Exclusions	Other names or conditions that should not be included in the code for the defect.
ICD-9-CM Codes	Applicable ICD-9-CM codes for the defect.
CDC/BPA Codes	Applicable CDC/BPA codes for the defect.
Diagnostic Methods	Postnatal procedures by which the defect may be accurately and reliably diagnosed.
Prenatal Diagnoses Not Confirmed Postnatally	Guidance on whether cases with only a prenatal diagnosis should be included in the defect code.
Additional Information	Tips and useful information about the defect.

Anencephalus

Description	Partial or complete absence of the brain and skull.
Inclusions	Acrania – Absence of skull bones with some brain tissue present. Absent brain, with or without skull bones present. Anencephalus Anencephaly Craniorachischisis – Anencephaly continuous with an open posterior spinal defect with no meninges covering the neural tissue.
Exclusions	Encephalocele Iniencephaly Rachischisis – When used alone, this term refers only to the spinal defect and should be coded as spina bifida without anencephalus.
ICD-9-CM Codes	740.0 – 740.1
CDC/BPA Codes	740.00 – 740.10
Diagnostic Methods	Anencephalus is easily recognized on physical examination at delivery.
Prenatal Diagnoses Not Confirmed Postnatally	Anencephalus may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data.

Additional Information:
Anencephalus is one of a group of defects that result from failure of the neural tube to close.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with anencephalus. However, these screening tests alone are not sufficient to diagnose the condition.

In cases where both anencephalus and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephalus and spina bifida should be coded.

Spina Bifida without Anencephalus

Description	Incomplete closure of the vertebral spine (usually posteriorly) through which spinal cord tissue and/or the membranes covering the spine (meninges) herniate.
Inclusions	Lipomeningocele Lipomyelomeningocele Meningocele – Herniation of meninges only. Meningomyelocele, Myelomeningocele – Herniation of meninges and spinal cord tissue. Myelocystocele Myelodysplasia Myeloschisis Open spina bifida Rachischisis – Open spina bifida without meninges covering the spinal cord tissue. Spina bifida aperta Spina bifida cystica
Exclusions	Closed spina bifida Diastematomyelia Diplomyelia Hydromyelia Spina bifida with coexisting anencephalus – Code only as anencephalus. Spina bifida occulta Syringomyelia Tethered spinal cord
ICD-9-CM Codes	741.0 or 741.9 without 740.0 – 740.1
CDC/BPA Codes	741.00 – 741.99 without 740.00 – 740.10
Diagnostic Methods	The majority of defects result in a direct opening on the infant's back that is easily recognized on physical examination at delivery. However, the exact nature of the defect (meningocele vs. myelomeningocele) may only be distinguished by CT or MRI scan, at surgery, or at autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	Spina bifida may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of spina bifida on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Spina bifida is one of a group of defects that result from failure of the neural tube to close.

Open lesions (spina bifida cystica, spina bifida aperta) are those with no covering or with only meninges covering the neural tissue. They usually leak cerebrospinal fluid. Closed lesions are covered by normal skin.

Closed lesions, or spina bifida occulta, do not produce an opening in the infant's back and may result only in a defect of the vertebral spine without significant herniation of neural tissue or neurologic impairment. When asymptomatic, it may be detected as an incidental finding on an x-ray or other test performed for a different indication.

Hydrocephalus and Arnold-Chiari malformation of the brain frequently, though not always, result from spina bifida. When present, there is no need to code them separately from the spina bifida.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated in spina bifida. However, these screening tests alone are not sufficient to diagnose the condition.

In cases where both anencephalus and spina bifida are present but are not continuous (i.e., not craniorachischisis), both anencephalus and spina bifida should be coded.

If the defect coding system includes unique codes for different levels of spina bifida (cervical; thoracic; lumbar; sacral) and a defect involves more than one level (cervicothoracic; thoracolumbar; lumbosacral), the highest level at which it occurs should be coded (i.e., cervical; thoracic; lumbar). The highest level of involvement determines the degree of associated neurologic impairment.

Hydrocephalus without Spina Bifida

Description	An increase in the amount of cerebrospinal fluid (CSF) within the brain resulting in enlargement of the cerebral ventricles and increased intracranial pressure.
Inclusions	Aqueductal stenosis – Narrowing or incomplete patency of the aqueduct of Sylvius between the third and fourth ventricles. This is the most common type of obstructive hydrocephalus (see below). Atresia of the foramina of Magendie and Luschka – Incomplete patency of the openings in the roof of the fourth ventricle through which CSF normally flows out of the brain. Communicating hydrocephalus – Impaired absorption of CSF, leading to an increased amount of CSF within the brain. Dandy-Walker malformation Hydranencephaly Hydrocephalus, type not specified Obstructive (noncommunicating) hydrocephalus – Obstruction of the flow of CSF within or out of the brain.
Exclusions	Hydrocephalus that results from a prior intracranial hemorrhage. This may be seen particularly in preterm infants. Hydrocephalus that occurs in association with spina bifida. Only the appropriate spina bifida code should be used. Ventriculomegaly
ICD-9-CM Codes	742.3 without 741.0 or 741.9
CDC/BPA Codes	742.30 – 742.39 without 741.00 – 741.99
Diagnostic Methods	While severe cases may be suspected by physical examination at delivery, hydrocephalus may be conclusively diagnosed only through direct visualization of the brain by cranial ultrasound, CT or MRI scan, surgery, or autopsy. While a child's head circumference may be increased for age, this measurement alone is not sufficient to make the diagnosis.
Prenatal Diagnoses Not Confirmed Postnatally	While hydrocephalus may be identified by prenatal ultrasound, it generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the diagnosis on prenatal ultrasound, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Severe cases may be

included without postnatal confirmation. Live-born children who survive should always have confirmation of the defect postnatally before being included.

Additional Information:
Hydrocephalus has a variety of etiologies, including infection, hemorrhage, and tumors, as well as anatomic lesions of the brain such as agenesis of the corpus callosum, encephalocele, cysts, and some bone dysplasias. In many cases, the etiology is not known.

In its true form, Dandy-Walker malformation is a malformation of the cerebellum and not a form of hydrocephalus. However, the term Dandy-Walker variant has been used to denote atresia of the foramina of Magendie and Luschka, dilatation of the cisterna magna (the space between the cerebellum and the brainstem), or cerebellar cysts, all of which have the appearance of increased fluid in the posterior fossa of the brain. It is, somewhat incorrectly, included in the defect codes for hydrocepahlus.

In hydranencephaly, the cerebral hemispheres are largely replaced by fluid-filled sacs within a normal skull. Hydranencephaly is not a true form of hydrocephalus. It is, somewhat incorrectly, included in the defect codes for hydrocepahlus.

Ventriculomegaly refers to enlargement of the cerebral ventricles, as measured by ultrasound (either prenatal or postnatal), CT or MRI scan. The distinction between hydrocephalus and ventriculomegaly has not been clearly defined, and these terms may be used interchangeably. Ventriculomegaly may be described as mild, moderate, or severe. How these designations correlate with the presence of true hydrocephalus, particularly when seen on prenatal ultrasound, also has not been clearly defined.

Encephalocele

Description	Herniation of brain tissue and/or meninges through a defect in the skull. The hernia sac is usually covered by skin.
Inclusions	Cephalocele Cranial meningocele – Herniation of meninges only. Encephalocele Encephalomyelocele - Herniation through a defect in a portion of both the skull and the upper spine. Encephalocystomeningocele Hydranencephalocele Meningoencephalocele Ventriculocele
Exclusions	NA
ICD-9-CM Codes	742.0
CDC/BPA Codes	742.00 – 742.09
Diagnostic Methods	Most cases of encephalocele are recognizable on physical examination after delivery. However, they may be conclusively diagnosed only through direct visualization of the brain by cranial ultrasound, CT or MRI scan, surgery, or autopsy. This is particularly true for internal herniations through the sphenoid, maxillary, or ethmoid bones, the orbit, or pharynx.
Prenatal Diagnoses Not Confirmed Postnatally	Encephalocele may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of a small encephalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Encephaloceles are often included as one of a group of defects that result from failure of the neural tube to close.

Maternal serum alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) and amniotic fluid acetylcholinesterase (ACHE) may be elevated with encephaloceles. However, these screening tests alone are not sufficient to diagnose the condition.

Occipital encephalocele is a component of Meckel-Gruber syndrome.

Microcephalus

Description	A cranial vault that is smaller than normal for age. The size of the cranial vault is an indicator of the size of the underlying brain.
Inclusions	Microcephalus Microcephaly Primary or True Microcephalus
Exclusions	Microcephalus that is secondary to a birth or delivery complication or to a postnatal insult or trauma.
ICD-9-CM Codes	742.1
CDC/BPA Codes	742.10
Diagnostic Methods	Microcephaly is usually easily diagnosed on physical examination by measurement of the occipitofrontal circumference (OFC, head circumference). However, there is difference of opinion as to what the lower limit of a normal head circumference should be (see below). Cranial ultrasound, CT or MRI scans may also reflect the diagnosis and contribute to the diagnosis of any underlying brain abnormalities.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of microcephalus on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Microcephalus may be defined variously as an OFC less than the 10^{th}, 5^{th}, or 3^{rd} percentile, or less than 2 or 3 standard deviations below the mean for age. There is no single accepted standard. Reference graphs differ in terms of the cut-off values displayed and the reference population used. Reference graphs for postnatal OFC growth usually are displayed separately for males and females.

In addition, it must be recognized that a proportion of normal children will have an OFC below any single cut-off value (i.e., 5% of the population has an OFC below the 5^{th} percentile by definition). For this reason, only children who have been given a clinical diagnosis of microcephalus should be included in birth defects surveillance data. The diagnosis should not be assigned based on the OFC measurement at birth without corroborating evidence from the medical record that the child carries the diagnosis of microcephalus.

Microcephalus itself is not a primary malformation, but a sign that the brain is small. It has a wide variety of causes. It is a component of a number of genetic syndromes. It also may result from a primary brain abnormality or a prenatal, perinatal, or postnatal insult. Examples of the latter include intrauterine infection, such as rubella or cytomegalovirus (CMV); *in utero* exposure to

alcohol and some medications, such as isotretinoin or dilantin; hypoxia during delivery; chronic hypoxia complicating prematurity; postnatal meningitis; head trauma. Only cases of microcephalus that have onset before delivery should be included in surveillance data. Unfortunately, the timing of onset and the etiology often are not known.

Anophthalmia/Microphthalmia

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.
Inclusions	Anophthalmia Microphthalmia Nanophthalmia – Microphthalmia with normal internal eye (intraocular) structures. This is a distinct genetic condition.
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.
ICD-9-CM Codes	743.0, 743.1
CDC/BPA Codes	743.00 – 743.10
Diagnostic Methods	These conditions are usually recognized on physical examination after delivery, especially by an ophthalmologist. However, the anteroposterior diameter of the globe may be measured only by ultrasound, CT or MRI scan, or at autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anophthalmia or microphthalmia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Microphthalmia may occur in association with colobomas (gaps) in the uvea, iris, choroid and/or optic nerve (colobomatous microphthalmia).

Anophthalmia and microphthalmia often are accompanied by malformations of the brain and face, and frequently are components of genetic syndromes.

Congenital Cataract

Description	An opacity of the lens of the eye that has its origin prenatally.
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
Exclusions	Any of the above types of cataract that has its origin after birth Corneal opacities
ICD-9-CM Codes	743.30 – 743.34
CDC/BPA Codes	743.320 – 743.326
Diagnostic Methods	Some cataracts are readily apparent on physical examination. Others are visible with an ophthalmoscope. However, they may be conclusively diagnosed only through examination by an ophthalmologist.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of a cataract on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

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.

Aniridia

Description	Hypoplasia of the iris of both eyes.
Inclusions	Aniridia Hypoplasia of the iris
Exclusions	Axenfeld-Rieger syndrome Chandler syndrome Coloboma of the iris Iris atrophy Peters anomaly Rieger syndrome
ICD-9-CM Codes	743.45
CDC/BPA Codes	743.42
Diagnostic Methods	Aniridia may be apparent on physical examination. However, it may be conclusively diagnosed only through examination by an ophthalmologist.
Prenatal Diagnoses Not Confirmed Postnatally	Aniridia should not be included in surveillance data unless diagnosed postnatally.

Additional Information:
Aniridia is usually associated with other abnormalities of the eye, including a persistent pupillary membrane; displaced lens; glaucoma; corneal and retinal abnormalities; hypoplasia of the optic nerve. While there is often near-total absence of the iris, it is never completely absent. Aniridia may be a component of Peters anomaly (abnormal development of the cornea and anterior chamber of the eye).

Aniridia has been associated with Wilms tumor of the kidney and certain chromosomal abnormalities. It may be inherited or may occur sporadically.

When aniridia occurs with a cataract in the same infant, both conditions should be coded.

Anotia/Microtia

Description	Anotia – Total absence of the external ear and canal. Microtia – Malformation or hypoplasia of the external ear (auricle, pinna).
Inclusions	Anotia Microtia
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. However, these defects are sometimes designated as Type I Microtia. Isolated absence, atresia, stenosis or malformation of the ear canal with a normal external ear. Congenital absence of the ear not diagnosed as anotia or microtia.
ICD-9-CM Codes	744.01, 744.23
CDC/BPA Codes	744.01, 744.21
Diagnostic Methods	Anotia and microtia are usually easily recognized on physical examination after delivery. However, abnormalities of the middle and inner ear may be conclusively diagnosed only by CT or MRI scan, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anotia or microtia on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery.

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 B – Generally small ears that retain most of the overall structure of the normal auricle. These should not be coded as microtia.

Type II B – 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 B – The ear is a rudiment of soft tissue and the auricle has no resemblance to a normal pinna.

Type IV B – 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 may be a component of Goldenhar and other syndromes.

Common Truncus (Truncus Arteriosus or TA)

Description	Failure of separation of the aorta and the pulmonary artery, resulting in a single common arterial trunk carrying blood from the heart to both the body and lungs.
Inclusions	Common truncus Truncus arteriosus (TA)
Exclusions	Aorto-pulmonary window
ICD-9-CM Codes	745.0
CDC/BPA Codes	745.00 – 745.01
Diagnostic Methods	While truncus defects may be suspected by clinical presentation, they may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.

Additional Information:
A ventricular septal defect is often 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. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

Transposition of the Great Arteries (TGA)

Description	Transposition of the aorta and the pulmonary artery 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).
Inclusions	Complete transposition (d-TGA without a VSD) Corrected transposition (l-TGA) Incomplete transposition (d-TGA with a VSD) Transposition of the Great Arteries (TGA), not otherwise specified Transposition of the Great Vessels (TGV)
Exclusions	NA
ICD-9-CM Codes	745.10, 745.11, 745.12, 745.19
CDC/BPA Codes	745.10 – 745.19
Diagnostic Methods	While transposition defects may be suspected by clinical presentation, they may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.

Additional Information:
In order for a child with d-TGA to survive, a communication must be present between the pulmonary and systemic circulations to allow oxygenated blood from the lungs to reach the right ventricle for distribution to the rest of the body through the abnormally placed aorta. In most instances, this communication is through a ventricular septal defect (incomplete TGA). If a VSD is not present, oxygenated blood from the lungs is returned directly to the lungs without being distributed to the rest of the body (complete TGA).

If the defect coding system does not include unique codes to differentiate TGA with and without a VSD (complete vs. incomplete), the VSD should be coded separately when present.

l-TGA (corrected transposition) is a defect in which the ventricle on the right side of the heart has the anatomic appearance of the left ventricle, and the ventricle on the left side of the heart has the anatomic appearance of the right ventricle (ventricular inversion). The pulmonary artery arises from the anatomic left ventricle and the aorta arises from the anatomic right ventricle (hence the designation of transposition). Because blood from the ventricle on the right flows through the pulmonary artery, and that from the ventricle on the left flows through the aorta, circulation is

normal as long as there are no other defects.

Transposition of the great arteries is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

Tetralogy of Fallot

Description	The simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis, a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy.
Inclusions	Pentalogy of Fallot – Tetralogy of Fallot with an associated inter-atrial communication, either a patent foramen ovale (PFO) or an atrial septal defect (ASD). Tetralogy of Fallot Tet TOF Some coding systems may also include Trilogy of Fallot, or Fallot's Triad – the simultaneous presence of an atrial septal defect, pulmonic stenosis, and right ventricular hypertrophy.
Exclusions	Simultaneous occurrence of a VSD and pulmonary stenosis that has TOF physiology but has not been diagnosed as Tetralogy of Fallot.
ICD-9-CM Codes	745.2
CDC/BPA Codes	745.20 – 745.21, 746.84
Diagnostic Methods	While Tetralogy of Fallot may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally.

Additional Information:
Children with Tetralogy of Fallot 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 Tetralogy of Fallot, may experience similar episodes. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean a child has been diagnosed with Tetralogy of Fallot.

Tetralogy of Fallot is one of several abnormalities of the outflow tract of the heart known as conotruncal defects. Some infants with these defects have a deletion on the short arm of chromosome 22 (22q11 deletion). This deletion is diagnosed using fluorescent *in situ* hybridization (FISH) and will not necessarily be detected on a routine karyotype analysis.

Ventricular Septal Defect (VSD)

Description	An opening in the septum that separates the left and right ventricles of the heart.
Inclusions	Ventricular septal defect VSD
Exclusions	Ventricular septal defects that occur as part of Tetralogy of Fallot or an endocardial cushion defect. Inflow-type, subtricuspid, and canal-type VSDs are assumed to be part of an endocardial cushion defect and should not be coded separately .
ICD-9-CM Codes	745.4
CDC/BPA Codes	745.40 – 745.59, excluding 745.498
Diagnostic Methods	Some isolated VSDs may be diagnosed on physical examination and/or EKG without direct imaging of the heart. However, many VSDs may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While VSDs may be identified by prenatal ultrasound, many close spontaneously before delivery. For this reason, VSDs that are diagnosed prenatally should not be included unless they have been confirmed postnatally. In addition, the absence of a VSD on prenatal ultrasound does not necessarily mean that a VSD will not be diagnosed after delivery, as it is not always possible to accurately visualize the entire ventricular septum by prenatal ultrasound.

Additional Information:

VSDs may be of several types, depending on the location of the opening along the ventricular septum. The most common are:

- Muscular
- Membranous
- Perimembranous

However, in many instances the type of VSD may not be specified in the medical record.

Many muscular, membranous and perimembranous VSDs may close spontaneously in the first weeks or months of life without treatment.

An aneurysm of the ventricular septum indicates a membranous or perimembranous VSD that is in the process of closing.

Atrial Septal Defect (ASD)

Description	An opening in the septum that separates the left and right atria of the heart.
Inclusions	Atrial septal defect, type not specified ASD Secundum ASD (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.
Exclusions	Atrioventricular septal defects (AVSD) – These are included under endocardial cushion defects (see below). Patent foramen ovale (PFO) – A PFO is normal *in utero* and frequently does not close until 24 to 48 hours after birth. Primum ASD (1° ASD) – These are included under endocardial cushion defects (see below).
ICD-9-CM Codes	745.5
CDC/BPA Codes	745.50 – 745.59, excluding 745.50
Diagnostic Methods	Some isolated ASDs may be diagnosed based on physical examination and/or EKG without direct imaging of the heart. However, many ASDs may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While ASDs may be identified by prenatal ultrasound, they may close spontaneously before delivery. For this reason, ASDs that are diagnosed prenatally should not be included unless they have been confirmed postnatally. In addition, the absence of an ASD on prenatal ultrasound does not necessarily mean that an ASD will not be diagnosed after delivery, as it is not always possible to accurately visualize the entire atrial septum by prenatal ultrasound.

Additional Information:
Secundum ASDs are usually located toward the middle of the atrial septum. Some close spontaneously without treatment.

Primum ASDs are located in the lower portion of the atrial septum, are etiologically related to endocardial cushion (AV canal) defects, and never close spontaneously.

Endocardial Cushion Defect

Description	A defect in both the lower portion of the atrial septum and the upper portion of the ventricular septum, producing a large opening (canal) in the central part of the heart. The adjacent parts of the mitral and tricuspid valves may also be abnormal, resulting in a single common atrioventricular valve. In extreme cases, virtually the entire atrial and ventricular septae may be missing.
Inclusions	Atrioventricular septal defect (AVSD) Common or complete atrioventricular (AV) canal Common atrioventricular (AV) orifices Endocardial cushion defect Primum atrial septal defect (1° ASD) – A defect only in the lower portion of the atrial septum. While this does not also involve a defect in the upper portion of the ventricular septum, it is etiologically related to the more complete form. A cleft mitral valve is often present. Common atrium – A very large primum ASD. Incomplete AV canal (incomplete endocardial cushion defect) – Same as a primum ASD. Inflow-type, subtricuspid, or canal-type ventricular septal defect (VSDAVC) – 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.
Exclusions	Secundum ASDs that coexist with a VSD. In this instance, both the ASD and the VSD should be coded.
ICD-9-CM Codes	745.60, 745.61, 745.69
CDC/BPA Codes	745.60 – 745.69
Diagnostic Methods	While endocardial cushion defects may be suspected by clinical presentation, examination, and EKG changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to

distinguish this condition from other abnormalities of the cardiac septae prenatally. Live-born children who survive should always have confirmation of the defect postnatally.

Additional Information:
Endocardial cushion defects are known to be associated with Down syndrome. Approximately 20% of children with Down syndrome have some type of endocardial cushion defect. Conversely, approximately 70% of children with an endocardial cushion defect have Down syndrome.

Pulmonary Valve Atresia and Stenosis

Description	Pulmonary valve atresia – Lack of patency, or failure of formation altogether, of the pulmonary valve, resulting in obstruction of blood flow from the right ventricle to the pulmonary artery. Pulmonary valve stenosis – Obstruction or narrowing of the pulmonary valve, which may impair blood flow from the right ventricle to the pulmonary artery.
Inclusions	Pulmonary valve atresia Pulmonary valve stenosis Pulmonic stenosis (PS)
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.
ICD-9-CM Codes	746.01, 746.02
CDC/BPA Codes	746.00 – 746.01
Diagnostic Methods	While pulmonary valve atresia or stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of pulmonary valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Pulmonary valve atresia or stenosis may occur with or without a coexisting ventricular septal defect. When it occurs with a VSD, the child may experience episodes of cyanosis or hypoxia similar to those seen in children with Tetralogy of Fallot. This results from shunting of unoxygenated blood across the VSD from the right to the left ventricle. Thus, the occurrence of cyanosis or hypoxia does not necessarily mean that the child has Tetralogy of Fallot.

Tricuspid Valve Atresia and Stenosis

Description	Tricuspid valve atresia – Lack of patency, or failure of formation altogether, of the tricuspid valve, resulting in obstruction of blood flow from the right atrium to the right ventricle. Tricuspid valve stenosis – Obstruction or narrowing of the tricuspid valve, which may impair blood flow from the right atrium to the right ventricle.
Inclusions	Tricuspid atresia Tricuspid stenosis
Exclusions	Tricuspid regurgitation without specific mention of tricuspid atresia or stenosis.
ICD-9-CM Codes	746.1
CDC/BPA Codes	746.10 (excluding 746.105)
Diagnostic Methods	While tricuspid valve atresia or stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of tricuspid valve atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.
Additional Information	NA

Ebstein's Anomaly

Description	Downward displacement if the tricuspid valve into the right ventricle. The tricuspid valve is usually hypoplastic and regurgitant.
Inclusions	Ebstein's anomaly Ebstein malformation
Exclusions	NA
ICD-9-CM Codes	746.2
CDC/BPA Codes	746.20
Diagnostic Methods	While Ebstein's anomaly may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of Ebstein's anomaly on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Ebstein's anomaly has been associated with lithium exposure during gestation. However, the magnitude of this association is probably very small.

Aortic Valve Stenosis

Description	Obstruction or narrowing of the aortic valve, which may impair blood flow from the left ventricle to the aorta.
Inclusions	Stenosis of the aortic valve
Exclusions	Stenosis of the aorta without mention of the aortic valve. Supra-valvular or sub-valvular aortic stenosis.
ICD-9-CM Codes	746.3
CDC/BPA Codes	746.30
Diagnostic Methods	While aortic valve stenosis may be suspected by clinical presentation, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of aortic valve stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.
Additional Information	NA

Hypoplastic Left Heart Syndrome (HLHS)

Description	A condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and aortic valves, and hypoplasia and coarctation of the aorta.
Inclusions	Any diagnosis of hypoplastic left heart syndrome, regardless of whether all conditions in the classical definition are present.
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. Hypoplastic left heart or small left ventricle that occurs as part of another complex heart defect, such as an endocardial cushion defect (AV canal).
ICD-9-CM Codes	746.7
CDC/BPA Codes	746.70
Diagnostic Methods	While hypoplastic left heart may be suspected by clinical presentation, examination, and EKG changes, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish this condition from other abnormalities of the left ventricle prenatally. Live-born children who survive should always have confirmation of the defect postnatally before being included.
Additional Information	NA

Patent Ductus Arteriosus (PDA)

Description	Abnormally persistent blood flow through the ductus arteriosus beyond the first few days of life.
Inclusions	Patent ductus arteriosus in infants with birth weight ≥ 2,500 grams who have not been given prostaglandin (see below).
Exclusions	Patent ductus arteriosus in infants with birth weight < 2,500 grams. Patent ductus arteriosus in infants ≥ 2,500 grams who have been given prostaglandin (see below).
ICD-9-CM Codes	747.0
CDC/BPA Codes	747.00
Diagnostic Methods	Some instances of patent ductus arteriosus may be diagnosed on physical examination. However, many PDAs may be diagnosed conclusively only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	Because a patent ductus arteriosus is normal and necessary during fetal life, this condition should not be included in surveillance data unless diagnosed postnatally at an appropriate age.

Additional Information:
In the normal fetal circulation, blood flows from the right ventricle to the pulmonary artery, then crosses the ductus arteriosus to the aorta for distribution to the body and the placenta. This bypasses much of the pulmonary circulation, since fetal blood is oxygenated by the placenta and not the lungs. Over the first hours after a normal full-term birth, smooth muscle in the wall of the ductus contracts and thickens to prevent blood flow through the ductus. Over the subsequent 2 to 3 weeks of life, the ductus is replaced by fibrous tissue and the communication is permanently sealed. Persistence of a patent ductus through which blood may flow beyond that time is abnormal.

In preterm infants, the ability of the ductus to constrict and close after delivery is not fully developed. Patent ductus arteriosus in a preterm infant is more likely to be a consequence of prematurity rather than an inherent abnormality. In these infants, it should not be coded as a defect.

The length of time required for the ductus to close is somewhat variable among term infants, and there is disagreement among specialists about the length of time after which patency is abnormal. Some birth defects surveillance programs only include PDAs that have been present for at least 6 weeks after birth.

Term infants who have additional heart defects may have abnormal patterns of blood flow or abnormal pressures in the pulmonary artery and aorta which prevent the ductus from closing. In these instances, the PDA is not an inherent abnormality but secondary to the additional defects.

In some severe heart defects, such as pulmonary atresia or d-TGA without a VSD, the infant's initial survival may depend on the presence of a patent ductus arteriosus in order for blood to reach the lungs for oxygenation. Prostaglandin (PGE) may be administered intravenously to maintain the patency of the ductus. In these instances, the PDA is an artifact of treatment of the underlying condition and should not be coded as a defect.

Patent ductus arteriosus may be a component of persistent transitional (fetal) circulation, in which the fetal pattern of blood flow through the ductus and bypassing the lungs, persists after birth. This is often a physiologic response to hypoxia from respiratory suppression, as may be seen with meconium aspiration.

Coarctation of the Aorta

Description	Narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body. The most common site of coarctation occurs distal to the origin of the left subclavian artery in the region of the ductus arteriosus.
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.
Exclusions	NA
ICD-9-CM Codes	747.10
CDC/BPA Codes	747.10 – 747.19
Diagnostic Methods	While coarctation of the aorta may be suspected by clinical presentation and examination, it may be conclusively diagnosed only through direct visualization of the heart by cardiac echo (echocardiography), catheterization, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of coarctation of the aorta on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Left-sided obstructive lesions of the heart, such as coarctation, have been associated with Turner syndrome (karyotype 45,X and other variants).

Cleft Palate without Cleft Lip

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.
Inclusions	Bifid or cleft uvula 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.
Exclusions	Cleft palate that coexists with a cleft lip. These should be coded as cleft lip only (see below).
ICD-9-CM Codes	749.0
CDC/BPA Codes	749.00 – 749.09
Diagnostic Methods	Cleft palate is usually easily recognized on physical examination by direct visualization of the pharynx after delivery. It may also be seen on CT or MRI scan, at surgery or autopsy. However, submucous cleft palate may be difficult to diagnose by physical examination during the first year of life.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of cleft palate on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Cleft palate may be unilateral, bilateral, or central in location. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded.

Cleft palate sometimes may be described as U-shaped or V-shaped. This distinction is not clinically meaningful and these conditions should not be coded differently.

Bifid uvula is often seen in association with a submucous cleft palate. However, bifid uvula also may occur alone. The presence of submucous cleft palate does not necessarily mean that a bifid uvula is present.

Cleft palate is one component of the Pierre Robin sequence, which also includes micrognathia and glossoptosis (when the tongue falls backward into the posterior pharynx). When diagnosed, Pierre Robin sequence should be coded separately.

Cleft Lip with and without Cleft Palate

Description	A defect in the upper lip resulting from incomplete fusion of the parts of the lip.
Inclusions	Complete cleft lip – The defect extends through the entire lip into the floor of the nose. Incomplete cleft lip – The defect extends through part of the lip but not into the floor of the nose.
Exclusions	Pseudocleft lip – An abnormal linear thickening, depressed grove, or scar-like pigmentary change on the skin of the lip without an actual cleft. Oblique facial clefts Cleft palate without an associated cleft lip
ICD-9-CM Codes	749.1, 749.2
CDC/BPA Codes	749.10 – 749.29
Diagnostic Methods	Cleft lip is usually easily recognized on physical examination after delivery. It may also be seen on CT or MRI scan, at surgery or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of cleft lip on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Cleft lip may be unilateral, bilateral, or central in location. If the defect coding system includes unique codes for these different types, the location of the cleft should be coded.

Cleft lip may also be seen in association with amniotic bands. In this instance, the amniotic bands should also be coded.

Choanal Atresia

Description	Congenital obstruction of the opening of the nasal cavity into the nasopharynx on either side. This prevents communication of the nasal cavity with the pharynx.
Inclusions	Choanal atresia, type not specified Choanal stenosis Membranous choanal atresia, with or without a bony rim Completely bony choanal atresia
Exclusions	NA
ICD-9-CM Codes	748.0
CDC/BPA Codes	748.00
Diagnostic Methods	Bilateral choanal atresia is usually easily recognized at birth from the clinical presentation of obligate mouth-breathing. Unilateral choanal atresia may be suspected by clinical examination. Both conditions may be diagnosed by the inability to pass a feeding tube from the nasal passage(s) into the posterior pharynx. Both conditions may also be seen on CT or MRI scan, at surgery or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of choanal atresia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Choanal atresia or stenosis may be unilateral or bilateral. If the defect coding system includes unique codes for these different types, the location should be coded.

Choanal atresia is one of the defects reported as part of the CHARGE association, which may also include colobomas, heart defects, retarded growth and development, genital hypoplasia, and ear anomalies and/or deafness.

Esophageal Atresia/Tracheoesophageal Fistula

Description	Esophageal atresia – A condition in which the esophagus ends in a blind pouch and fails to connect with the stomach. Tracheoesophageal fistula – An abnormal communication between the esophagus and the trachea. This is almost always associated with some form of esophageal atresia.
Inclusions	Esophageal atresia alone Esophageal atresia with tracheoesohpageal (TE) fistula Esophageal stenosis, stricture, ring, or web TE fistula Tracheoesophageal fistula, all types
Exclusions	Tracheal atresia Tracheoesophageal cleft
ICD-9-CM Codes	750.3
CDC/BPA Codes	750.30 – 750.35
Diagnostic Methods	The diagnosis may be suspected by the clinical presentation of polyhydramnios, vomiting, or respiratory distress. Esophageal atresia may be diagnosed by x-ray documentation of failure of a feeding tube to pass from the pharynx into the stomach. Tracheoesophageal atresia may be conclusively diagnosed only by CT or MRI scan, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included.

Additional Information:
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 stomach remains patent.

TE fistula is one of the defects reported as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, anal atresia, renal defects, and limb anomalies.

Rectal and Large Intestinal Atresia/Stenosis

Description	Complete or partial occlusion of the lumen of one or more segments of the large intestine and/or rectum.
Inclusions	Anal atresia or stenosis Colonic atresia or stenosis Imperforate anus Large intestinal atresia or stenosis Rectal atresia or stenosis
Exclusions	Apple peel intestinal atresia Duodenal atresia or stenosis Ileal atresia or stenosis Jejunal atresia or stenosis Small intestinal atresia or stenosis
ICD-9-CM Codes	751.2
CDC/BPA Codes	751.20 – 751.24
Diagnostic Methods	Anal atresia (imperforate anus) is usually easily recognized at birth by physical examination. While large intestinal and rectal atresia or stenosis may be suspected by the clinical presentation of failure to pass meconium or stool, they may be conclusively diagnosed only through direct imaging of the bowel by x-ray, barium enema, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of intestinal, rectal or anal atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
These conditions may occur with or without a fistula.

Anal atresia is one of the defects reported as part of the VATER, or VACTERL, association, which may also include vertebral and cardiac defects, TE fistula, renal defects, and limb anomalies.

Pyloric Stenosis

Description	Hypertrophy (thickening) of the muscles of the pylorus connecting the stomach to the duodenum, resulting in complete or partial obstruction of the passage of food and gastric contents.
Inclusions	Infantile (congenital) hypertrophic pyloric stenosis Pyloric stenosis
Exclusions	Pylorospasm (intermittent spasm of the pyloric muscles) without permanent narrowing of the lumen.
ICD-9-CM Codes	750.5
CDC/BPA Codes	750.51
Diagnostic Methods	Many instances of pyloric stenosis may be diagnosed by the clinical presentation and physical examination. However, other cases may be diagnosed conclusively only by abdominal ultrasound or contrast x-ray of the stomach.
Prenatal Diagnoses Not Confirmed Postnatally	In rare cases, pyloric stenosis may develop prenatally and may be identified on prenatal ultrasound. However, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of pyloric stenosis on prenatal ultrasound does not mean that it will not develop after delivery (see below).

Additional Information:
Pyloric stenosis most typically presents with intractable vomiting in a 3- to 4-week-old infant. While it may appear late in gestation, it develops more commonly in the first month or two after birth. As such, it may not be a truly congenital defect.

The etiology of pyloric stenosis remains unclear, but is probably multifactorial with both genetic and environmental influences. Pyloric stenosis has been associated with erythromycin use in newborn infants.

Hirschsprung Disease (Congenital Megacolon)

Description	Hirschsprung disease – Absence of the parasympathetic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum, which may result in congenital megacolon. Megacolon – Enlargement of the diameter of part or all of the colon.
Inclusions	Aganglionic megacolon Congenital megacolon Hirschsprung disease, type not specified Long-segment Hirschsprung disease (Type II) Short-segment Hirschsprung disease (Type I) Total colon (intestinal) aganglionosis
Exclusions	Psychogenic megacolon
ICD-9-CM Codes	751.3
CDC/BPA Codes	751.30 – 751.34
Diagnostic Methods	Hirschsprung disease (congenital megacolon) may be suspected by contrast x-ray (barium enema). However, it may be diagnosed conclusively only through direct assessment of the presence or absence of ganglion cells in rectal tissue at biopsy, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of congenital megacolon on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:
Megacolon may result from any condition that inhibits normal passage of the intestinal contents. Primary underlying conditions include Hirschsprung disease, rectal and large intestinal atresia/stenosis, imperforate anus, and voluntary stool retention (psychogenic megacolon). In Hirschsprung disease, the aganglionic segment of intestine is small and empty, while the normally enervated segment proximal to the affected area is enlarged and filled with fecal matter.

Hirschsprung disease is classified according to the extent of aganglionosis. In 80% of cases, aganglionosis extends from the anal sphincter and rectum to the middle of the sigmoid colon; in 10% to 20% of cases, it extends further to the transverse or right colon; in 3% of cases, aganglionosis involves the entire colon.

Possible complications of Hirschsprung disease/congenital megacolon include bowel perforation, enterocolitis (intestinal inflammation), peritonitis (inflammation of the lining of the abdomen), and septicemia (bloodstream infection).

Approximately 3% of infants with Down syndrome have aganglionosis of the colon. When Down syndrome and Hirschsprung disease/congenital megacolon occur in the same infant, both conditions should be coded.

Biliary Atresia

Description	Congenital absence of the lumen of the extrahepatic bile ducts.
Inclusions	Agenesis, absence, hypoplasia, obstruction or stricture of the bile duct(s)
Exclusions	Congenital or neonatal hepatitis Intrahepatic biliary atresia (absence or paucity of bile ducts within the liver) not associated with extrahepatic biliary atresia
ICD-9-CM Codes	751.61
CDC/BPA Codes	751.65
Diagnostic Methods	Biliary atresia may be suspected by the clinical presentation and the presence of elevated direct bilirubin and liver function tests. However, it may be conclusively diagnosed only through direct assessment of the bile ducts by abdominal ultrasound, CT or MRI scan, biliary excretion study (HIDA scan), surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	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.

Additional Information:
The liver contains within its substance intrahepatic bile ducts and passages that join and coalesce to form two main ducts that carry bile out of the liver.

The extrahepatic bile ducts include the hepatic duct (formed by the two main ducts that carry bile out of the liver), the cystic duct (which carries bile out of the gallbladder where it is stored), and the common bile duct (formed by the junction of the hepatic duct and the cystic duct), which carries bile into the duodenum for excretion.

When extrahepatic biliary atresia is present, the intrahepatic bile ducts may also be abnormal or atretic.

Patients with biliary atresia may have jaundice due to direct hyperbilirubinemia, which is not treated with phototherapy. The more common type of neonatal jaundice due to indirect hyperbilirubinemia may be treated with phototherapy and does not indicate the presence of biliary atresia.

Renal Agenesis/Hypoplasia

Description	Renal agenesis – Complete absence of the kidney Renal hypoplasia – Incomplete development of the kidney
Inclusions	Renal agenesis, dysgenesis, aplasia, or hypoplasia Potter syndrome secondary to renal agenesis/hypoplasia
Exclusions	Cystic renal dysplasia Cystic kidney disease Multicystic kidney Multicystic dysplastic kidney Polycystic kidney Renal cysts Renal dysplasia Small kidney
ICD-9-CM Codes	753.0
CDC/BPA Codes	753.00 – 753.01
Diagnostic Methods	Bilateral renal agenesis is usually easily recognized on physical examination after delivery. 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. Each of these diagnoses may be conclusively diagnosed only through direct assessment by abdominal ultrasound, CT or MRI scan, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	Bilateral renal agenesis may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. While bilateral renal hypoplasia and unilateral renal agenesis/hypoplasia may be suspected by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. Lack of visualization of a kidney on prenatal ultrasound does not always indicate that the kidney is truly absent.

Additional Information:
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.

Bladder Exstrophy

Description	A defect in the lower abdominal wall and anterior wall of the bladder through which the lining of the bladder is exposed to the outside.
Inclusions	Classic bladder exstrophy Ectopia vesicae Epispadias-exstrophy complex Extroversion of the bladder Variants of bladder exstrophy Vesical exstrophy
Exclusions	Ambiguous genitalia without mention of bladder exstrophy Cloacal exstrophy Isolated epispadias
ICD-9-CM Codes	753.5
CDC/BPA Codes	753.50
Diagnostic Methods	Bladder exstrophy is easily recognized on physical examination at delivery. However, the exact nature of the defect and associated anomalies may only be distinguished by abdominal ultrasound, contrast x-ray studies, CT or MRI scan, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish bladder exstrophy from cloacal exstrophy. Live-born children who survive should always have confirmation of the defect postnatally before being included.

Additional Information:
In the classic form of bladder exstrophy, the entire urinary tract is open anteriorly from the urethral meatus to the umbilicus. The pubic bones are widely separated, as are the abdominal muscles and fascia. There is eversion/exposure of the posterior bladder wall. The genitalia of either gender may be involved and may be bifid or duplicated. The classic form of bladder exstrophy occurs more frequently in males.

Variants of bladder exstrophy occur more rarely and affect females more often then males. Included among these variants are superior vesical fistula, closed exstrophy, duplicate exstrophy, pseudoexstrophy, inferior vesicle. Epispadias is almost uniformly present, but should not be coded separately.

Ambiguous genitalia may be noted in patients with bladder exstrophy if an obvious scrotum and

testes are not present. However, ambiguous genitalia should not be coded as a separate defect in these instances.

Bladder exstrophy should be distinguished from cloacal exstrophy, in which the urinary, intestinal, and genital structures open into a common cavity (the cloaca). The distinction may only be possible with detailed diagnostic studies, surgery, or at autopsy. In cloacal exstrophy, bladder exstrophy and imperforate anus are also present. In bladder exstrophy without cloacal exstrophy, the anus is patent. When both bladder and cloacal exstrophy are present, only cloacal exstrophy should be coded.

Obstructive Genitourinary Defect

Description	Partial or complete obstruction of the flow of urine at any level of the genitourinary tract from the kidney to the urethra.
Inclusions	Atresia, stenosis, stricture or occlusion of one or both ureters, the bladder neck, the urethra or urethral meatus Dilatation of one or both ureters Hydronephrosis Hydroureter Hypoplastic ureter Megaloureter Posterior urethral valves Obstruction of the ureteropelvic junction (UPJ), the ureterovesical (UV) junction, or the vesicourethral (VU) junction Urethral valves, type not specified
Exclusions	Inhibition of urinary flow at any of the above sites resulting solely from neurologic impairment.
ICD-9-CM Codes	753.2, 753.6
CDC/BPA Codes	753.20 – 753.29, 753.60 – 753.69
Diagnostic Methods	Genitourinary tract obstruction may be suspected by the clinical presentation. However, the exact nature of the defect and the level of obstruction may only be distinguished by direct visualization. The upper urinary tract (kidneys and ureters) is usually visualized with renal ultrasound, radionuclide scan, or a contrast study such as intravenous pyelography (IVP). The lower urinary tract (bladder and urethra) is usually visualized directly with cystoscopy or urethral endoscopy, or with contrast studies such as voiding cystourethrogram (VCUG) and sometimes IVP. Obstructions also may be diagnosed at surgery or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While obstructive genitourinary defects may be identified by prenatal ultrasound, many lesions diminish or resolve spontaneously prior to birth. For this reason, they should not be included in surveillance data without postnatal confirmation (see below). In addition, the absence of genitourinary obstruction on prenatal ultrasound does not necessarily mean that an obstructive defect will not be diagnosed after delivery.

Additional Information:
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.

Increased use of ultrasound screening has led to the recognition of asymptomatic genitourinary tract obstructions in the fetus and newborn, many of which resolve without treatment and would not otherwise have been diagnosed. Inclusion of these lesions in birth defects surveillance data may inflate the apparent frequency of significant obstructive genitourinary defects. If it is possible to correlate the findings on prenatal and/or newborn ultrasound with the clinical course of symptoms and treatment, this should factor into the decision as to which obstructive lesions to include in the surveillance data.

Hypospadias and Epispadias

Description	Hypospadias – 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. Epispadias – Displacement of the opening of the urethra (urethral meatus) dorsally and proximally (on the top and closer to the body) in relation to the tip of the glans of the penis.
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°, or penile 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 Epispadias
Exclusions	Chordee alone without associated hypospadias Ambiguous genitalia
ICD-9-CM Codes	Hypospadias 752.61 Epispadias 752.62
CDC/BPA Codes	Hypospadias 752.600 – 752.607, 752.620, 752.605 – 752.607 Epispadias 752.621
Diagnostic Methods	Both hypospadias and epispadias are usually easily recognized on physical examination at delivery. They may also be seen on contrast x-rays of the urinary tract, at surgery or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be diagnosed by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of hypospadias or epispadias on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery.

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.

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.

Epispadias is almost uniformly present with bladder exstrophy. In these cases, only the bladder exstrophy should be coded.

Reduction Deformity, Upper Limbs

Description

Complete or partial absence of the upper arm (humerus), lower arm (radius and/or ulna), wrist (carpals), hand (metacarpals), or fingers (phalanges).

Inclusions

Transverse limb reduction – Complete or partial absence of the distal (furthest from the body) structures of the arm 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. Types of transverse limb reductions include:

Acheiria – Absence of a hand
Adactyly – Absence of digits (fingers), excluding isolated missing thumb (see below)
Aphalangia – Absence of phalanges. Fingers contain 3 phalanges each. The thumb contains 2 phalanges.

Amelia – Complete absence of the upper limb (humers, radius, ulna, wrist, hand and fingers).

Hemimelia, Meromelia – Partial absence of a limb. This may refer to either transverse or longitudinal reductions.

Oligodactyly – Fewer than 5 digits.

Transverse terminal deficiency – Complete absence of the distal structures of the arm with the proximal structures intact. This term usually refers to reduction defects below the elbow.

Congenital amputation, type not specified.

Longitudinal limb reduction – Partial absence of the arm in parallel with the long axis of the arm. These may involve preaxial (on the thumb side), postaxial (on the fifth finger side), or central parts of the arm. Types of longitudinal limb reductions include:

Ectrodactyly
Ectromelia
Isolated missing thumb
Lobster claw hand
Radial aplasia or hypoplasia

Split-hand malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb

reduction in which there is complete or partial absence of one or more of the central rays (second through fourth fingers and their associated metacarpal bones) of the hand.

Ulnar aplasia or hypoplasia

Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the arm with all or part of the distal segment present.

Phocomelia is a general term for any type of intercalary limb reduction.

Reduction deformities of the upper limb not elsewhere coded or of unspecified type – Complete or partial absence of the arm that does not fall within the above categories or for which there is no specific description.

Exclusions

Shortened arms, forearms, hands, or fingers that have all of their component parts, including those that are part of a generalized chondodystrophy, osteodystrophy, or dwarfism.

Hypoplastic nails

ICD-9-CM Codes

755.20 – 755.29

CDC/BPA Codes

755.20 – 755.29

Diagnostic Methods

Limb reductions are usually easily recognized on physical examination at delivery. However, the exact nature of the defect may only be distinguished by x-ray, surgery, or autopsy.

Prenatal Diagnoses Not Confirmed Postnatally

While these conditions may be identified by prenatal ultrasound, they generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present. Live-born children who survive should always have confirmation of the defect postnatally before being included.

Additional Information:

The terminology for limb reduction deformities is often confusing. Some terms (such as "phocomelia"), have been misused and others (such as "ectrodactyly"), have been used for both longitudinal and transverse defects. If medical record review is available, it is important to look for a complete description of all structures that are present and absent in order to verify the diagnosis.

Preaxial refers to the side of the arm on which the thumb and radius are located.
Postaxial refers to the side of the arm on which the fifth finger and ulna are located.

Transverse limb reductions may be seen in association with amniotic bands. When both are present, both conditions should be coded.

Rudimentary or nubbin fingers may be present at the distal end of a transverse limb reduction. Their presence alone does not change the classification of the defect as transverse.

Joint contractures are commonly seen in association with longitudinal limb deficiencies.

Intercalary reduction deformities (phocomelia) have been associated with the use of thalidomide during early pregnancy. However, thalidomide use may result in a number of other defects, including longitudinal reduction deformities. Intercalary defects also may occur without exposure to thalidomide.

Reduction deformities are one of the defects that may be reported as part of:

The VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia.

Poland anomaly, which also includes deficiency of the pectoralis muscle on the same side.

Moebius anomaly (Oromandibular-Limb Hypogenesis Spectrum), which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), sixth and seventh cranial nerve palsies.

Reduction Deformity, Lower Limbs

Description

Complete or partial absence of the upper leg (femur), lower leg (tibia and/or fibula), ankle (tarsals), foot (metatarsals), or toes (phalanges).

Inclusions

Transverse limb reduction – Complete or partial absence of the distal (furthest from the body) structures of the 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. Types of transverse limb reductions include:

Adactyly – Absence of digits (toes)
Aphalangia – Absence of phalanges. The smaller toes contain 3 phalanges each. The big toe contains 2 phalanges.

Amelia – Complete absence of the lower limb (femur, tibia, fibula, ankle, foot, and toes).

Hemimelia, Meromelia – Partial absence of a limb. This may refer to either transverse or longitudinal reductions.

Oligodactyly – Fewer than 5 digits.

Transverse terminal deficiency – Complete absence of the distal structures of the leg with the proximal structures intact.

Congenital amputation, type not specified

Longitudinal limb reduction – Partial absence of the leg in parallel with the long axis of the leg. These may involve preaxial (on the big toe side), postaxial (on the fifth toe side), or central parts of the leg. Types of longitudinal limb reductions include:

Ectrodactyly
Ectromelia
Fibular aplasia or hypoplasia

Split-foot malformation (split hand/split foot malformation, SHSF) – A central longitudinal limb reduction in which there is complete or partial absence of one or more of the central rays (second through fourth toes and their associated metatarsal bones) of the foot.

	Tibial aplasia or hypoplasia
	Intercalary limb reduction – Complete or partial absence of the proximal (closest to the body) or middle segments of the leg with all or part of the distal segment present.
	Phocomelia – A general term for any type of intercalary limb reduction.
	Reduction deformities of the lower limb not elsewhere coded or of unspecified type – Complete or partial absence of the leg that does not fall within the above categories or for which there is no specific description.
Exclusions	Shortened upper and/or lower legs, feet, or toes that have all of their component parts, including those that are part of a generalized chondodystrophy, osteodystrophy, or dwarfism.
	Hypoplastic nails.
ICD-9-CM Codes	755.30 – 755.39
CDC/BPA Codes	755.30 – 755.39
Diagnostic Methods	Limb reductions are usually easily recognized on physical examination at delivery. However, the exact nature of the defect may only be distinguished by x-ray, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	While these conditions may be identified by prenatal ultrasound, they generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present. Live-born children who survive should always have confirmation of the defect postnatally before being included.

Additional Information:
The terminology for limb reduction deformities is often confusing. Some terms (such as "phocomelia") have been misused and others (such as "ectrodactyly") have been used for both longitudinal and transverse defects. If medical record review is available, it is important to look for a complete description of all structures that are present and absent in order to verify the diagnosis.

Preaxial refers to the side of the leg on which the big toe and tibia are located.
Postaxial refers to the side of the leg on which the fifth toe and fibula are located.

Transverse limb reductions may be seen in association with amniotic bands. When both are

present, both conditions should be coded.

Rudimentary or nubbin toes may be present at the distal end of a transverse limb reduction. Their presence alone does not change the classification of the defect as transverse.

Joint contractures are commonly seen in association with longitudinal limb deficiencies.

Intercalary reduction deformities (phocomelia) have been associated with the use of thalidomide during early pregnancy. However, thalidomide use may result in a number of other defects, including longitudinal reduction deformities. Intercalary defects also may occur without exposure to thalidomide.

Reduction deformities are one of the defects that may be reported as part of:

The VATER or VACTERL association, which also may include vertebral, cardiac and renal defects, TE fistula, and anal atresia.

Moebius anomaly (Oromandibular-Limb Hypogenesis Spectrum), which also may include a small mouth, small chin (micrognathia), small tongue (hypoglossia), sixth and seventh cranial nerve palsies.

Gastroschisis

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.
Inclusions	Gastroschisis
Exclusions	Omphalocele
ICD-9-CM Codes	756.79
CDC/BPA Codes	756.71
Diagnostic Methods	Gastroschisis is usually easily recognized on physical examination after delivery. However, in some instances, it may be conclusively distinguished from omphalocele only at surgery or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	Gastroschisis may be included when only diagnosed prenatally. However, it may be difficult to distinguish gastroschisis from omphalocele on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of gastroschisis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

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 alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (AFAFP) may be elevated with gastroschisis. However, these screening tests alone are not sufficient to diagnose the condition.

Omphalocele

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.
Inclusions	Omphalocele
Exclusions	Gastroschisis Umbilical hernia
ICD-9-CM Codes	756.79
CDC/BPA Codes	756.70
Diagnostic Methods	Omphalocele is usually easily recognized on physical examination after delivery. However, in some instances, it may be conclusively distinguished from gastroschisis only at surgery or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	Omphalocele may be included when only diagnosed prenatally. However, it may be difficult to distinguish omphalocele from gastroschisis on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of omphalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

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 alphafetoprotein (MSAFP) and/or amniotic fluid alphafetoprotein (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.

Congenital Hip Dislocation

Description	Location of the head of the femur (bone of the upper leg) outside its normal location in the cup-shaped cavity formed by the hip bones (acetabulum).
Inclusions	Congenital hip dislocation, unilateral or bilateral Developmental dysplasia of the hip Teratologic hip dislocation
Exclusions	Flexion deformity/contracture of the hip Hip click Predislocation of the hip Preluxation of the hip Subluxation of the hip Unstable hip
ICD-9-CM Codes	754.30, 754.31, 754.35
CDC/BPA Codes	754.30
Diagnostic Methods	Hip dislocation may be suspected, and sometimes diagnosed, by physical examination. However, ultrasound or x-ray are the definitive diagnostic tests.
Prenatal Diagnoses Not Confirmed Postnatally	While this condition may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of hip dislocation on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery.

Additional Information:

The terminology describing congenital hip dislocation is often confusing. An unstable hip, in which the femoral head may be moved in and out of the acetabulum on physical examination, often resolves spontaneously in young infants. A truly dislocated hip, in which the femoral head remains out of the acetabulum for a prolonged period, may result in acetabular deformity unless treated. Hence, the designation developmental dysplasia of the hip.

The stability of the hip joint may be evaluated on physical examination. In the Barlow test, lateral pressure is applied to the hip with the knees flexed in an attempt to move the head of the femur out of the hip joint (acetabulum) into a dislocated position. In the Ortolani maneuver, a laterally dislocated femoral head is moved back into normal position in the acetabulum by applying pressure medially. The presence of either sign indicates a hip dislocation is present. However, their absence does not always mean that a dislocation is not present. In some instances, the femoral head may be fixed in a dislocated position and it may not be possible to move it in and out of the joint.

Congenital hip dislocation occurs more frequently after footling or breech deliveries and is more common in females than males. It is most often an isolated condition, although hip dysplasia may occur with generalized skeletal abnormalities and in some genetic syndromes. Some instances of congenital hip dislocation are probably familial.

Diaphragmatic Hernia

Description	Incomplete formation of the diaphragm through which a portion of the abdominal contents herniate into the thoracic cavity.
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.
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.
ICD-9-CM Codes	756.6
CDC/BPA Codes	756.610 – 756.617
Diagnostic Methods	While diaphragmatic hernia may be suspected by the clinical presentation of respiratory distress, feeding intolerance, and/or cardiac compromise, it may be conclusively diagnosed only through x-ray, contrast study of the bowel, CT or MRI scan, surgery, or autopsy.
Prenatal Diagnoses Not Confirmed Postnatally	Diaphragmatic hernia may be included in surveillance data when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included.

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.

Trisomy 13

Description	The presence of three copies of all or a large part of chromosome 13.
Inclusions	Patau syndrome Mosaic Patau syndrome Mosaic trisomy 13 Translocation Patau syndrome Translocation trisomy 13 Trisomy 13, not otherwise specified Trisomy D_1, not otherwise specified
Exclusions	Balanced translocations involving chromosome 13
ICD-9-CM Codes	758.1
CDC/BPA Codes	758.10 – 758.19
Diagnostic Methods	Trisomy 13 may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.
Prenatal Diagnoses Not Confirmed Postnatally	Trisomy 13 may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below).

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, PUBS, 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 may varies among affected individuals.

Down Syndrome (Trisomy 21)

Description	The presence of three copies of all or a large part of chromosome 21.
Inclusions	Down syndrome Mosaic Down syndrome Mosaic trisomy 21 Translocation Down syndrome Translocation trisomy 21 Trisomy 21, not otherwise specified
Exclusions	Balanced translocations involving chromosome 21 "Downs facies" without associated trisomy 21.
ICD-9-CM Codes	758.0
CDC/BPA Codes	758.00 – 758.09
Diagnostic Methods	Down syndrome may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.
Prenatal Diagnoses Not Confirmed Postnatally	Down syndrome may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 21 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below).

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, PUBS, 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, upslanting 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 endocardial cushion 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.

Trisomy 18

Description	The presence of three copies of all or a large part of chromosome 18.
Inclusions	Edwards syndrome Mosaic Edwards syndrome Mosaic trisomy 18 Translocation Edwards syndrome Translocation trisomy 18 Trisomy 18, not otherwise specified
Exclusions	Balanced translocations involving chromosome 18
ICD-9-CM Codes	758.2
CDC/BPA Codes	758.20 – 758.290
Diagnostic Methods	Trisomy 18 may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.
Prenatal Diagnoses Not Confirmed Postnatally	Trisomy 18 may be included when only diagnosed through direct analysis of fetal chromsomes obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below).

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, PUBS, or preferably postnatal blood or tissue samples.

Most pregnancies affected with trisomy 18 result in spontaneous abortion. Approximately 50% of

live-born 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.

Fetal Aclohol Syndrome (FAS)

Description	A spectrum of abnormalities resulting from exposure to alcohol *in utero*. While the specific abnormalities vary among individuals, the hallmarks include growth deficiency, microcephaly, facial dysmorphisms, and neurodevelopmental abnormalities.
Inclusions	Fetal alcohol syndrome (FAS)
Exclusions	Fetal alcohol effects/facies, without diagnosis of FAS
ICD-9-CM Codes	760.71
CDC/BPA Codes	760.71
Diagnostic Methods	While fetal alcohol syndrome may be suspected from a history of maternal alcohol use during pregnancy, the condition may be conclusively diagnosed only through direct examination of the infant by a physician (usually a dysmorphologist or developmental specialist) familiar with the spectrum of FAS abnormalities.
Prenatal Diagnoses Not Confirmed Postnatally	While fetal alcohol syndrome may be suspected from a history of maternal alcohol use during pregnancy, the condition should not be included in surveillance data without postnatal confirmation.

Additional Information:
A number of minor malformations may be present with fetal alcohol syndrome, most notably hypoplasia of the maxillary bone (middle) of the face, and a thin upper lip with smooth philtrum (crease). However, these are often subtle in the newborn and may not be recognized until later in childhood. Older children with FAS may manifest poor coordination, irritability, hyperactivity, and neurodevelopmental abnormalities.

Fetal alcohol syndrome is the extreme of a spectrum of effects on growth and development resulting from alcohol exposure *in utero*. At low levels of exposure, the only apparent effect may be a reduction in birth weight. The clinical features and neurodevelopmental abnormalities become increasingly prominent with increasing levels of exposure.

Amniotic Bands

Description	Strands of tissue that float in the amniotic fluid as a consequence of tears or ruptures in the amniotic membrane which surrounds the fetus during development.
Inclusions	Amniotic bands Amniotic band sequence, syndrome, or disruption complex Amniotic rupture sequence Streeter bands Constriction rings – Soft tissue depressions or grooves encircling part of the body, usually a limb.
Exclusions	NA
ICD-9-CM Codes	NA
CDC/BPA Codes	658.80
Diagnostic Methods	Structural defects resulting from amniotic bands usually are readily apparent on physical examination after delivery. However, the fact that they are a consequence of amniotic bands may not be apparent unless a remnant of an amniotic strand is present or amniotic bands were noted on prenatal ultrasound.
Prenatal Diagnoses Not Confirmed Postnatally	When amniotic bands are seen on prenatal ultrasound, their presence should be correlated with any structural defects noted, and the guidelines for including those defects when only diagnosed prenatally should be followed. Live-born children who survive should always be examined for evidence of amniotic bands postnatally. In addition, the absence of amniotic bands on prenatal ultrasound does not necessarily mean that they are not truly present.

Additional Information:
Amniotic bands may be present in the amniotic sac without impacting the fetus. When noted as an isolated condition without associated structural defects, they should not be coded.

Structural defects that may occur as a result of amniotic bands include:

- Pseudosyndactyly (digits compressed together by an encircling band)
- Distal limb amputation, hypoplasia, lymphedema, or deformation
- Oral clefts
- Encephalocele
- Anencephaly

Other disruptive defects of the skull