

BD-STEPS Birth Defects Case Definitions

Table of Contents

- 1. Explanation/Glossary 2
- 2. Spina Bifida..... 4
- 3. Anophthalmia/microphthalmia 6
- 4. Anotia/Microtia..... 7
- 5-8. Conotruncal Heart Defects 8
- 9-11. Obstructive Heart Defects 10
- 12. Total Anomalous Pulmonary Venous Connection 12
- 13. Cleft Lip +/- Palate 13
- 14. Cleft Palate 14
- 15. Esophageal Atresia +/- TE Fistula 15
- 16. Limb Deficiency, Transverse..... 16
- 17. Diaphragmatic Hernia 18
- 18. Gastroschisis..... 19

1. Explanation/Glossary

BIRTH DEFECT & DEFINITION

- The specific birth defect and its definition are listed
- Other names for the defect are given as well as the defect group if applicable

TYPES & DEFINITIONS

- The types of the specific birth defect (if they exist) and their definitions are listed

INCLUSIONS

The standard inclusion criteria for the defect are listed; any exceptions to the standard inclusions are listed for that specific defect(s)

The standard inclusions are as follows:

- In general, case infants must have one or more of the specific birth defects listed in the abstraction instructions
- The etiology of the defect(s) must be unknown or uncertain
- Cases can be live born or stillborn infants ≥ 20 weeks' gestation or 500 grams, or prenatally diagnosed and terminated fetuses at any gestational age
- Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect will only be accepted as cases for a small number of defects; this information is listed in each defect category under the inclusion/exclusion boxes
- Cardiac defects must be confirmed by echocardiography (echo), catheterization (cath), surgery or autopsy; include cases diagnosed by prenatal ultrasound/echo only if done by a pediatric cardiologist or in a prenatal diagnosis center with expertise in this area
- Fetuses/infants with "apparently balanced" chromosomal translocations and inversions are included in the study

EXCLUSIONS

The standard exclusion criteria for the defect are listed. Any exceptions to the standard exclusions are listed in the abstraction instructions for that specific defect(s).

The standard exclusions are as follows:

- In general, fetuses/infants with chromosomal/microdeletion disorders or single gene disorders are excluded
- Cardiac defects that are clinically-diagnosed, i.e., using only physical exam, chest radiography, ECG are excluded
- Autopsy cases in which maceration or disruption (from termination techniques) preclude definitive diagnosis are excluded; this does not apply to defects for which prenatal diagnosis is accepted under certain circumstances (see INCLUSIONS)
- Conjoined and acardiac twins are excluded
- Selective CHARGE syndrome/association cases according to the following criteria:

- An effort should be made to determine if *CHD7* mutation testing has been performed
- All cases of CHARGE phenotype that are *CHD7* mutation positive are excluded from the Study
- All cases of CHARGE phenotype that are *CHD7* mutation negative or have had no mutation testing are included in the Study, even if the phenotype strongly fulfills the criteria of CHARGE syndrome
- Fetuses/infants with a microdeletion or microduplication detected by a chromosome microarray (aCGH) are excluded from the Study if parental studies did not show the same microdeletion/ microduplication, indicating that the event was most likely *de novo*. When parental studies were either not done or the results are not available, the local reviewer will exclude the case if at least one of the following criteria is met:
 - the microdeletion/microduplication was previously characterized as being associated with a clinical phenotype
 - the microdeletion/microduplication contains at least one gene that is known or strongly suspected to be dosage sensitive
 - the microdeletion/microduplication is of sufficient size to be likely pathogenic (generally >400 kb)

ICD-9-CM CODES

- The ICD-9-CM codes for the specific birth defect are listed
- The ICD-9 codes are used for the discharge diagnoses and are generally less specific than a surveillance system's BPA or modified codes

CBDRP CODES

The CBDRP codes for the eligible defect(s) are listed

2. Spina Bifida

BIRTH DEFECT & DEFINITION

- SPINA BIFIDA--herniation of the meninges and/or spinal cord tissue through a bony defect of spine closure

OTHER NAMES: spina bifida cystica, spina bifida aperta, myeloschisis, myelodysplasia, etc.

TYPES & DEFINITIONS

- MENINGOMYELOCELE/MYELOMENINGOCELE--90% of lesions, herniation of meninges and spinal cord tissue
- MENINGOCELE--herniation of meninges without spinal cord tissue
- RACHISCHISIS--spine defect without meninges covering the neural tissue
- LIPOMENINGOMYELOCELE/LIPOMENINGOCELE--lipomatous (fatty) tissue associated with a bony defect of the spine and herniation of meninges or spinal cord tissue, usually closed and located in the lumbosacral region
- MYELOCYSTOCELE--cystic lesion of the spinal cord central canal and herniation through a spinal defect
- OPEN LESION--neural tissue open to environment or covered by membrane only (90% of lesions)
- CLOSED LESION--neural tissue covered by normal skin
- LEVEL OF LESION--highest and lowest vertebrae--cervical (C), thoracic (T), lumbar (L), sacral (S)

INCLUSIONS

- All cases including those cases prenatally diagnosed that do not have a postnatal examination to confirm the defect

EXCLUSIONS

- Spina bifida occulta
- Primary tethered cord
- Syringomyelia (hydromyelia)
- Diastematomyelia
- Diplomyelia
- Caudal lipomatous lesions not documented to involve neural tissue
- Iniencephaly--a rare neural tube defect involving the occiput and inion, resulting in extreme retroflexion of the head variably combined with occipital encephalocele or rachischisis of the cervical and thoracic spine
- Craniorachischisis (anencephaly with contiguous rachischisis)
- Any type of spina bifida with coexisting anencephaly

ICD-9-CM CODES

- SPINA BIFIDA WITH HYDROCEPHALUS--741.0
- SPINA BIFIDA WITHOUT MENTION OF HYDROCEPHALUS--741.9

CBDRP CODES

- 741x0x: Meningomyelocele/myelomeningocele
- 741x1x: Meningocele
- 741x2x: Myelocele
- 741x3x: Myelocystocele

- 741x4x: Lipomeningomyelocele
- 741x5x: Lipomeningocele
- 741x6x: Rachischisis
- 741x8x: Other specified spina bifida
- 741x9x: Unspecified spina bifida
- 7410xx: Arnold Chiari malformation ± hydrocephalus, open lesion
- 7411xx: Arnold Chiari malformation ± hydrocephalus, closed lesion
- 7412xx: Arnold Chiari malformation ± hydrocephalus, unspecified open/closed lesion
- 7413xx: Hydrocephalus, other (aqueduct of Sylvius) or NOS, open lesion
- 7414xx: Hydrocephalus, other (aqueduct of Sylvius) or NOS, closed lesion
- 7415xx: Hydrocephalus, other (aqueduct of Sylvius) or NOS, unspecified open/closed lesion
- 7417xx: No mention hydrocephalus, open lesion
- 7418xx: No mention hydrocephalus, closed lesion
- 7419xx: No mention hydrocephalus, unspecified open/closed lesion
- 741xx1: Highest level, cervical
- 741xx2: Highest level, thoracic
- 741xx3: Highest level, lumbar
- 741xx4: Highest level, sacral
- 741xx9: Highest level, unspecified

3. Anophthalmia/microphthalmia

BIRTH DEFECT & DEFINITION

- ANOPHTHALMIA--total absence of the eye tissue or apparent absence of the globe in an orbit that otherwise contains normal adnexal structures
- MICROPHTHALMIA--reduction in the volume of the eye, usually characterized by corneal diameter less than 10 mm or anteroposterior globe diameter less than 20 mm

NOTE: these conditions may be seen with the ending "ia", "os" or "us"

TYPES & DEFINITIONS

- TRUE OR PRIMARY ANOPHTHALMIA--as above; occurs when there is complete failure of formation of the primary optic vesicle, usually bilateral; when unilateral, may have contralateral microphthalmia; verified only when histologic/microscopic exam shows that all ocular tissue is absent
- MICROPHTHALMIA--categories: colobomatous (uveal, iris, choroid and/or optic nerve) or noncolobomatous

OTHER NAMES: nanophthalmia = microphthalmic eye with normal intraocular structures and is a distinct genetic malformation

INCLUSIONS

- All cases must include diagnosis by an ophthalmologist or confirmation by surgical pathology or autopsy

EXCLUSIONS

- "Small eyes" or "small palpebral fissures" unless there is confirmation of anophthalmia or microphthalmia
- Isolated microcornea with normal ocular size
- Ocular colobomas without anophthalmia or microphthalmia

ICD-9-CM CODES

- ANOPHTHALMIA--743.00
- MICROPHTHALMIA--743.10-743.12

CBDRP CODES

- 743000-4: Anophthalmos
- 743100-4: Microphthalmos

4. Anotia/Microtia

BIRTH DEFECT & DEFINITION

- ANOTIA--total absence of the external ear and canal
- MICROTIA--malformation or hypoplasia of the auricle, ranging from measurably small external ear with minimal structural abnormality, to an ear with major structural alteration with absent or blind-ending canal

TYPES & DEFINITIONS

Microtia Classification System of Meurman (modified from Marks):

- TYPE I--generally small ear that retains most of the overall structure of the normal auricle--similar to lop/ cup defect, auditory meatus is usually patent and defects of the ossicular chain are infrequent
- TYPE II--moderately severe anomaly with longitudinal mass of cartilage with some resemblance to pinna (rudimentary auricle will be hook-shaped, have an S-shape or question mark appearance)
- TYPE III--ear is a rudiment of soft tissue and the auricle has no resemblance to the normal pinna
- TYPE IV--complete absence of all external ear structures, anotia

NOTE: types I - III will occasionally be accompanied by a preauricular tag(s)

INCLUSIONS

Standard

EXCLUSIONS

- Small ears NOS or small ears that retain most of the normal structure
- Type I microtia with or without abnormality of the external auditory canal
- Isolated atresia or stenosis of the external auditory canal
- Normal ears that are misplaced: low set, posteriorly rotated, etc.
- "Decreased cartilage" reported as part of the estimate of gestational age

ICD-9-CM CODES

- ANOTIA--744.01
- MICROTIA--744.23

NOTE: absence of the ear, congenital is included in the "other" code--744.09

CBDRP CODES

- 744010-4: Anotia
- 744210-4: Microtia

5-8. Conotruncal Heart Defects

Double Outlet Right Ventricle (DORV) (any type) was an eligible defect in NBDPS but is NOT in BD-STEPS. This includes all variants of DORV – those with transposed/malposed great vessels and those with normally related great vessels but with anatomy similar to TOF.

- In NBDPS, cases of DORV with anatomy similar to TOF were included in the Fallot (TOF) classification group. In BD-STEPS the Fallot category will not include these cases since DORV is ineligible. Thus, the Fallot category in NBDPS will NOT be exactly comparable to Fallot category in BD-STEPS. To facilitate data combination between the two studies, files will be created excluding the DORV-TOF cases from the Fallot category in NBDPS so the data can be combined and compared.
- In NBDPS, cases of DORV with transposed/malposed great arteries are a separate category distinct from either DORV or TGA. Thus, data combination of the TGA category between NBDPS and BDSTEPS will be unaffected by the ineligibility of DORV.

BIRTH DEFECT & DEFINITION

- CONOTRUNCAL HEART DEFECTS (outflow tract anomalies)--anomalies of the outflow tract of the heart

TYPES & DEFINITIONS

5. TRUNCUS ARTERIOSUS (TA)--single common arterial trunk instead of separate pulmonary artery and aorta, almost always associated with a malalignment-type VSD; there are subtypes 1, 2, 3 based on the pattern of truncal branching; no need to specify type
6. DEXTRO-TRANSPOSITION OF GREAT ARTERIES (DTGA, DTGV)--transposed great arteries such that the pulmonary artery arises from the left ventricle and the aorta arises from the right ventricle
 - May be isolated or with other congenital heart defects (e.g., VSD, pulmonic stenosis)
 - If occurs with a VSD, do not code the VSD separately; use the code dTGA-VSD (745110)
 - If no VSD, use code for dTGA with intact ventricular septum (745100)
7. TETRALOGY OF FALLOT (TOF, TET)--tetralogy = a malalignment-type VSD creates subvalvar pulmonic stenosis, overriding of the aorta, and right ventricular hypertrophy (= 4 defects in one code)
 - Do not code VSD and pulmonic stenosis separately
 - Absent and atretic pulmonary valve are distinctly different defects; thus, careful attention should be paid to the description and coding; use TOF code 745200 and PV insufficiency code 746020 for TOF with absent pulmonary valve
 - "Pentalogy of Fallot" (TOF + ASD2) is an archaic term. If noted in the medical record, code both defects separately (TOF and ASD).
8. PULMONARY ATRESIA--atresia of the pulmonary valve/artery; depending on subtype, is considered either in conotruncal defects (7a) or with obstructive defects (8b, 8c)
 - 8a. PULMONARY ATRESIA WITH VSD (PA/VSD, TETRALOGY WITH PULMONARY ATRESIA)--absent connection from the right ventricle to the pulmonary artery and the aorta, usually with malalignment-type VSD; CDRP code is 747310;

alternative archaic terms are Truncus, type 4 or pseudotruncus. This is included as a conotruncal defect.

INCLUSIONS

- Standard

EXCLUSIONS

- Standard

ICD-9-CM CODES

- TETRALOGY OF FALLOT--745.2
- PULMONARY ATRESIA WITH VSD, TETRALOGY OF FALLOT WITH PULMONARY ATRESIA--747.3 and 745.2
- TRUNCUS ARTERIOSUS--745.0
- DEXTRO-TRANSPOSITION OF GREAT ARTERIES--745.10

CBDRP CODES

- 745000: Truncus arteriosus (TA)
- 745100: Dextro-transposition of great arteries with intact ventricular septum (D-TGA/D-TGV w/ IVS)
- 745110: Dextro-transposition of great arteries with ventricular septal defect (D-TGA/D-TGV w/ VSD)
- 745200: Tetralogy of Fallot (TOF)
- 747310: Pulmonary atresia with VSD (tetralogy of Fallot with pulmonary atresia) (PA w/ VSD)

9-11. Obstructive Heart Defects

BIRTH DEFECT & DEFINITION

- OBSTRUCTIVE HEART DEFECTS--broad group of congenital heart defects in which there is obstruction to the flow of blood through either the left or right side of the heart or the great vessels

TYPES & DEFINITIONS

Right-Sided Obstructive Anomaly:

9. TRICUSPID ATRESIA (TriAtresia, TrA)--atretic connection between the right atrium and the right ventricle, due to the absence or non-patency of the valve
 - Be sure to code using the CBDRP tricuspid atresia code (746100) for atresia alone (not for stenosis)
 - Tricuspid stenosis is not a BD-STEPS-eligible defect; in the original ICD9-BPA system, one code (7461) lumped both atresia and stenosis, which was a cause for confusion; in the presence of other eligible codes, use 746880 ("CHD, OS") for tricuspid stenosis
8. PULMONARY ATRESIA --atresia of the pulmonary valve/artery; depending on subtype, is considered either in conotruncal defects (8a) or with obstructive defects (8b, 8c)
 - 8b. PULMONARY ATRESIA WITH VSD (NOT TOF VARIANT)--use this code (746030) if PA/VSD is present, but anatomic details of the VSD/aorta are **not** described as "membranous/malalignment-type," or if the VSD is "muscular". This is included as a right-sided obstructive defect.
 - 8c. PULMONARY ATRESIA WITH INTACT VENTRICULAR SEPTUM (PA/IVS)--this is a distinctly different defect; code as 746000. This is included as a right-sided obstructive defect.

Left-Sided Obstructive Anomaly:

10. COARCTATION OF THE AORTA (COA)--narrowing of the descending aorta, distal to the left subclavian; in most instances, the narrowing occurs close to the region where the ductus arteriosus inserts and is called juxtaductal coarctation
 - Code separately, even in the presence of hypoplastic left heart syndrome
 - There are no exclusions based on severity (even a 'mild' coarctation is included), although it is helpful to include information about the severity (gradient) or type (ledge vs. long segment coarctation) of the lesion
11. HYPOPLASTIC LEFT HEART SYNDROME (HLHS)--extreme smallness of the left-sided heart structures (mitral valve and left ventricle) and aorta (including the aortic valve, ascending aorta, arch, and sometimes descending aorta [coarctation]); implies normally related great arteries
 - Typical cases include mitral hypoplasia or atresia PLUS aortic hypoplasia or atresia, in the presence of a diminutive (non-apex forming) left ventricle
 - In the typical case of HLHS, coarctation should be coded separately when present; mitral and aortic atresia or hypoplasia do not need separate coding if HLHS is coded

- In the presence of an unbalanced AV canal with right dominance, in which the left ventricle and aorta may be small, code the individual anomalies, but do **not** use the HLHS code
- A ventricular septal defect may be present and its size may influence the dimensions of the left ventricle (mitral atresia and intact septum are often associated with very small ventricle)

INCLUSIONS

- Standard

EXCLUSIONS

- Coarctation of the aorta cases that are prenatally diagnosed but lack postnatal confirmation are excluded

ICD-9-CM CODES

- COARCTATION OF THE AORTA--747.10
- HYPOPLASTIC LEFT HEART SYNDROME--746.7
- PULMONARY VALVE ATRESIA WITH INTACT VENTRICULAR SEPTUM--746.01
- TRICUSPID ATRESIA--746.1

CBDRP CODES

- 747100: Coarctation of the aorta, preductal (proximal)
- 747110: Coarctation of the aorta, postductal (distal)
- 747120: Coarctation of the aorta, juxtaductal
- 747190: Coarctation of the aorta, NOS
- 746700: Hypoplastic left heart syndrome (HLHS)
- 746000: Pulmonary valve atresia/intact ventricular septum (PA/IVS)
- 746030: Pulmonary valve atresia with VSD (not tetralogy of Fallot variant) (PA w/ VSD, not TOF)
- 746100: Tricuspid atresia (TrA)

12. Total Anomalous Pulmonary Venous Connection

BIRTH DEFECT & DEFINITION

- TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION (RETURN/DRAINAGE)--a condition in which a pulmonary vein or combination of pulmonary veins drains anomalously into the systemic venous circulation to the right heart or the body instead of into the left heart; often occurs with other cardiac defects

TYPES & DEFINITIONS

- TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION (RETURN/DRAINAGE) (TAPVC/TAPVR/TAPVD)--failure of all pulmonary veins to connect to the left atrium

NOTE: pulmonary blood returns to the heart via supra-diaphragmatic or infra-diaphragmatic routes; these details are not needed for coding purposes

INCLUSIONS

Standard

EXCLUSIONS

- Cases prenatally diagnosed that do not have a postnatal diagnostic examination to confirm the diagnosis
- Partial anomalous pulmonary venous return/connection/drainage

ICD-9-CM CODES

- TOTAL ANOMALOUS PULMONARY VENOUS RETURN--747.41

CBDRP CODES

- 747420: Total anomalous pulmonary venous return/connection/drainage (TAPVR)

13. Cleft Lip +/- Palate

BIRTH DEFECT & DEFINITION

- CLEFT LIP +/- PALATE--incomplete closure of the lip; often accompanied by a maxillary alveolar (gum) defect and/or cleft palate ; maxillary alveolar defect may be a complete cleft that is continuous with the cleft palate, or it may be limited to a notch on the gum; cleft lip may be unilateral, bilateral, or median (distinguished from bilateral cleft lip by agenesis of premaxilla)

TYPES & DEFINITIONS

- COMPLETE CLEFT LIP--defect extends through the entirety of the lip and the nasal floor; may be unilateral or bilateral; usually associated with a more severe nasal deformation
- INCOMPLETE CLEFT LIP--defect of lip that does not extend into the nasal floor; may be unilateral or bilateral; there may be an incomplete cleft lip on one side and a complete cleft lip on the other side
- PSEUDOCLEFT LIP (excluded from BD-STEPS)--abnormal linear thickening or depressed groove of skin, or subtle scar-like pigmentary difference paralleling the philtral ridge on the affected side; may be associated with slight notch of the vermilion or a mild slouching of the alar cartilage

INCLUSIONS

- Standard
- If cleft palate is associated with any type of cleft lip, it is coded as a cleft lip and palate, not cleft palate

EXCLUSIONS

- Pseudocleft lip; microform cleft lip; forme fruste cleft lip
- Tessier type facial clefts
- Oblique facial clefts
- Prenatal diagnosis without postnatal confirmation of the defect(s)
- Any orofacial cleft with coexisting holoprosencephaly
- Any orofacial cleft with coexisting anencephaly

ICD-9-CM CODES

- CLEFT LIP--749.10-749.14
- CLEFT LIP WITH PALATE--749.20-749.25

CBDRP CODES

- 749101-3: Cleft lip, unilateral
- 749110: Cleft lip, bilateral
- 749120: Cleft lip, central
- 749495: Cleft lip, NOS
- 749201-3: Cleft lip and palate, unilateral
- 749210: Cleft lip and palate, bilateral cleft lip
- 749220: Cleft lip and palate, central cleft lip
- 749290: Cleft lip and palate, NOS

14. Cleft Palate

BIRTH DEFECT & DEFINITION

- CLEFT PALATE--hole in roof of the mouth; incomplete fusion of the palatal shelves; may be limited to soft palate or also extend onto hard palate; if cleft palate is associated with cleft lip, it is coded as a cleft lip and palate

TYPES & DEFINITIONS

- PIERRE ROBIN ANOMALY (SEQUENCE)--combination of micrognathia, cleft palate, glossoptosis (tongue falls back into pharynx)
- SUBMUCOUS CLEFT PALATE (excluded from BD-STEPS)--defect of the soft palate with mucosa or a reduced, thin muscle layer bridging the midline; difficult to diagnose clinically in 1st year; often associated with a bifid uvula

INCLUSIONS

Standard

EXCLUSIONS

- Submucous cleft palate
- Bifid or cleft uvula without overt cleft palate
- Any orofacial cleft with coexisting holoprosencephaly
- Any orofacial cleft with coexisting anencephaly

ICD-9-CM CODES

- CLEFT PALATE--749.00-749.04

CBDRP CODES

- 749001-3: Cleft hard palate, unilateral
- 749010: Cleft hard palate, bilateral
- 749020: Cleft hard palate, central
- 749030: Cleft hard palate, NOS
- 749041-3: Cleft soft palate, unilateral
- 749050: Cleft soft palate, bilateral
- 749060: Cleft soft palate, central
- 749070: Cleft soft palate, NOS
- 749090: Cleft palate, NOS

15. Esophageal Atresia +/- TE Fistula

BIRTH DEFECT & DEFINITION

- ESOPHAGEAL ATRESIA +/- TRACHEOESOPHAGEAL FISTULA (T-E FISTULA, TEF)--congenital complete discontinuity of the lumen of the esophagus resulting in a blind esophageal pouch occurring with or without an abnormal communication between the esophagus and trachea

TYPES & DEFINITIONS

- There are several classification schemas
- In 90% of cases the upper esophagus ends in a blind pouch and the lower segment forms a fistula with the trachea

INCLUSIONS

Standard

EXCLUSIONS

- TE fistula without esophageal atresia
- Esophageal stenosis
- Trachea atresia
- Tracheoesophageal cleft

ICD-9-CM CODES

- ESOPHAGEAL ATRESIA, TRACHEOESOPHAGEAL FISTULA--750.3

CBDRP CODES

- 750300: Esophageal atresia without TE fistula
- 750310: Esophageal atresia with TE fistula

16. Limb Deficiency, Transverse

BIRTH DEFECT & DEFINITION

- TRANSVERSE LIMB DEFICIENCY--complete or partial absence of distal structures of a limb in a transverse plane at the point where the deficiency begins, with proximal structures essentially intact

OTHER NAMES: transverse limb reduction defect, congenital amputation

TYPES & DEFINITIONS

- AMELIA--complete absence of a limb
- HEMI- OR MEROMELIA--partial absence of a limb (rather nonspecific; can also be used for longitudinal defects)
- TRANSVERSE TERMINAL DEFICIENCY--absence of distal structures with proximal structures essentially intact (used for deficiencies below the elbow)
- APHALANGIA--absence of phalanges
- ADACTYLY--absence of digits
- OLIGODACTYLY--fewer than 5 digits
- ACHEIRIA--absence of a hand
- AMNION RUPTURE SEQUENCE--limb deficiencies and constrictions associated with tears or rupture in the amnion

INCLUSIONS

- Standard
- Isolated missing digits, except isolated missing thumb (would be longitudinal limb deficiency)

EXCLUSIONS

- Unspecified type of limb deficiency
- Generalized limb shortening including chondrodysplasias
- Nail hypoplasia
- Brachydactylies type A-E
- Lower extremity deficiencies with sirenomelia sequence
- Absent digits with split-hand or split-foot
- Absent digits with longitudinal deficiencies (absent digits with corresponding metacarpal/metatarsal +/- long bone deficiencies, e.g., radial, ulnar, tibial, or fibular ray defects)
- LIMB-BODY WALL COMPLEX (excluded from BD-STEPS)--disruption complex involving body wall defects and limb deficiencies, as well as neural tube defects, heart, and other anomalies

ICD-9-CM CODES

- UPPER LIMB TRANSVERSE DEFICIENCY--755.21
- UPPER LIMB LONGITUDINAL DEFICIENCY OF PHALANGE(S)--755.29
- LOWER LIMB TRANSVERSE DEFICIENCY--755.31
- LOWER LIMB LONGITUDINAL DEFICIENCY OF PHALANGE(S)--755.39

CBDRP CODES

- 755200-4: Transverse deficiency or amputation of the arm, NOS
- 755205-9: Total absence of the arm

- 755240-4: Absence of the forearm and hand
- 755245-9: Absence of hand or fingers
- 755300-4: Transverse deficiency or amputation of the leg, NOS
- 755305-9: Total absence of the lower limb
- 755340-4: Absence of the lower leg and foot
- 755345-9: Absence of foot or toes

17. Diaphragmatic Hernia

BIRTH DEFECT & DEFINITION

- DIAPHRAGMATIC DEFECTS (HERNIA)--incomplete formation of the diaphragm through which some portion of the abdominal contents herniates into the thoracic cavity

OTHER NAMES: congenital diaphragmatic hernia (CDH), absence, agenesis, or aplasia of diaphragm, hemidiaphragm

TYPES & DEFINITIONS

- POSTEROLATERAL HERNIA = BOCHDALEK HERNIA--defect involving the posterior and/or lateral portions of the diaphragm
- AGENESIS--apparent absence of an entire side of diaphragm; represents a large Bochdalek hernia
- ANTERIOR HERNIA = MORGAGNI HERNIA (aka Retrosternal, Parasternal, Morgagni-Larrey hernia)
- LARGE ANTERIOR HERNIA = SEPTUM TRANSVERSUM HERNIA--type of defect found in Pentalogy of Cantrell
- PARAESOPHAGEAL HERNIA--defect in the diaphragm surrounding the esophagus
- OTHER--includes, for example, central diaphragm defects, anterolateral defects, and unusual/atypical defects
- HERNIA SAC--approximately 15% of CDH have a sac, which is a localized thinning or out-pouching of the diaphragm; a sac is **not** a type of hernia

INCLUSIONS

- Standard
- Prenatally diagnosed cases should be included only if bowel was documented in the chest by prenatal ultrasound
- Diaphragmatic hernia with Pentalogy of Cantrell

EXCLUSIONS

- Eventration of the diaphragm--not a true herniation, but an upward displacement of abdominal contents into an out-pouched diaphragm resulting from weakness or absence of diaphragmatic musculature
- Hiatal hernia
- CCAM (cystic adenomatoid malformation of the lung)

ICD-9-CM CODES

- DIAPHRAGMATIC HERNIA--756.6

CBDRP CODES

- 756600-4: Diaphragmatic hernia, NOS
- 756605: Diaphragmatic hernia, esophageal
- 756610-4: Diaphragmatic hernia, Bochdalek
- 756615-9: Diaphragmatic hernia, Morgagni

18. Gastroschisis

BIRTH DEFECT & DEFINITION

- GASTROSCHISIS--congenital fissure of the anterior abdominal wall, lateral to the umbilicus, usually to the right, with a small bridge of skin separating the defect from the umbilicus; accompanied by herniation of the small, and part of the large, intestines, and occasionally other abdominal organs, into the amniotic cavity, and lacking a protective membrane

TYPES & DEFINITIONS

- LIMB-BODY WALL COMPLEX (excluded from BD-STEPS)--disruption complex involving body wall defects and limb deficiencies, as well as neural tube defects, heart, and other anomalies

INCLUSIONS

- Standard
- Prenatally diagnosed cases if high resolution ultrasound was done and the umbilicus was visualized

EXCLUSIONS

- Limb-body wall complex

ICD-9-CM CODES

- GASTROSCHISIS--756.73

CBDRP CODES

- 756710: Gastroschisis