BD STEPS Birth Defects Case Definitions

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1. 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; iniencephaly always has a closed cranium;

ICD-9-CM CODES

- SPINA BIFIDA WITH HYDROCEPHALUS--741.0

- SPINA BIFIDA WITHOUT MENTION OF HYDROCEPHALUS--741.9

BDSTEPS 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

2. 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
- Cryptophthalmos

ICD-9-CM CODES

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

BDSTEPS CODES

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

3. 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

BDSTEPS CODES

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

4-7. Conotruncal Heart Defects

The BDSTEPS definitions of dTGA and TOF is NOT exactly comparable to the NBDPS definitions of dTGA and TOF. DORV, which can be a sub-type of dTGA or TOF, is excluded from BD-STEPS altogether. However, the NBDPS cases of DORV-dTGA and DORV-TOF could be identified and excluded from the NBDPS dTGA and TOF case groups if analysts wanted to combine dTGA or TOF data from both studies.

BIRTH DEFECT & DEFINITION

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

TYPES & DEFINITIONS

4. 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
5. 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
    - o May be isolated or with other congenital heart defects (e.g., VSD, pulmonic stenosis)
    - o If occurs with a VSD, do not code the VSD separately; use the code dTGA-VSD (745110)
    - o If no VSD, use code for dTGA with intact ventricular septum (745100)
6. 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)
    - o Do not code VSD and pulmonic stenosis separately
    - o 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
    - o "Pentalogy of Fallot" (TOF + ASD2) is an archaic term. If noted in the medical record, code both defects separately (TOF and ASD).
7. PULMONARY ATRESIA--atresia of the pulmonary valve/artery; depending on subtype, is considered either in conotruncal defects (7a) or with obstructive defects (7b, 7c)

    7a. 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; BDSTEPS code is 747310; alternative archaic terms are Truncus, type 4 or pseudotruncus. This is included as a conotruncal defect.

INCLUSIONS

- Standard
- Include infants who are NEGATIVE or NOT TESTED for 22q11.2 deletion

EXCLUSIONS

- Exclude infants who are POSITIVE for 22q11.2 deletion

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

BDSTEPS 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)

8-10. 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:

8. 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 BDSTEPS tricuspid atresia code (746100) for atresia alone (not for stenosis)
    - Tricuspid stenosis is not a BDSTEPS-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
7. PULMONARY ATRESIA --atresia of the pulmonary valve/artery; depending on subtype, is considered either in conotruncal defects (7a) or with obstructive defects (7b, 7c)
    7b. 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.
    7c. 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:

9. 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
10. 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--746.00
- TRICUSPID ATRESIA--746.1

BDSTEPS 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)

11. Anomalous Pulmonary Venous Return

BIRTH DEFECT & DEFINITION

- ANOMALOUS PULMONARY VENOUS RETURN (CONNECTION/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 RETURN (CONNECTION/DRAINAGE) (TAPVR/TAPVC/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

- PARTIAL ANOMALOUS PULMONARY VENOUS RETURN (CONNECTION/DRAINAGE) (PAPVR/PAPVC/PAPVD)--failure of 1,2,or 3 of the 4 pulmonary veins to connect to the left atrium; often associated with a sinus venosus type ASD

INCLUSIONS

Standard

EXCLUSIONS

- Cases prenatally diagnosed that do not have a postnatal diagnostic examination to confirm the diagnosis

ICD-9-CM CODES

- TOTAL ANOMALOUS PULMONARY VENOUS RETURN--747.41
- PARTIAL ANOMALOUS PULMONARY VENOUS RETURN--747.42

BDSTEPS CODES

- 747420: Total anomalous pulmonary venous return/connection/drainage (TAPVR)
- 747430: Partial anomalous pulmonary venous return/connection/drainage (PAPVR)

12. 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 BDSTEPS)--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 vermillion 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)

ICD-9-CM CODES

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

BDSTEPS 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

13. 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 BDSTEPS)--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

ICD-9-CM CODES

- CLEFT PALATE--749.00-749.04

BDSTEPS 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

14. 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

BDSTEPS CODES

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

15. 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: 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

INCLUSIONS

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

EXCLUSIONS

- Unspecified limb deficiency
- Generalized limb shortening including chondrodysplasias
- Nail hypoplasia
- Brachydactylies type A-E
- Lower extremity deficiencies with sirenomelia sequence

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

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

16. 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

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

BDSTEPS CODES

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

17. 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--disruption complex involving lateral body wall defect, limb reduction defect, neural tube defects, heart and other anomalies

INCLUSIONS

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

EXCLUSIONS

Standard

ICD-9-CM CODES

- GASTROSCHISIS--756.79

BDSTEPS CODES

- 756710: Gastroschisis