Congenital Malformations and Deformations

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AGENDA

• According to WHO, congenital anomalies are defined as:
  – Chromosomal abnormalities;
  – Inborn errors of metabolism; and
  – Hereditary diseases
• Present at birth
Congenital heart defects

- Congenital heart defects are the most common type of birth defect
- Present in 9 out of every 1,000 births
- May range from simple to severe
- Can increase risk of developing other cardiac conditions

Coding congenital conditions in ICD-10-CM

Atrial septal defect (ASD) – Q21.1, Q21.2
  - Among the most common heart defects
Aortic valvular stenosis – Q23.0
  - Causes obstruction to blood flow between the left ventricle and aorta
Ventricular septal defect (VSD) – Q21.0
  - Holes in the septum between the ventricles
  - May close on their own
Valvular Stenosis

- Aortic valve
  - Allows blood to flow from left ventricle into the aorta and out to the rest of the body
- Pulmonary valve
  - Separates the right ventricle from the pulmonary artery

Valvular Stenosis

- Mitral valve
  - Opens to allow blood to flow from the left atrium into the left ventricle
- Tricuspid valve
  - Allows blood to flow from the right atrium into the right ventricle

Valvular Stenosis

Congenital Conditions

- Aortic valve
  - Most common bicuspid aortic valve
- Pulmonic valve
  - Second most common congenital heart defect
Valvular Stenosis

Congenital Conditions

- Mitral valve
  - Seems to be an increase in patients with parents with LVOTOs
- Tricuspid valve
  - May have deformed leaflets, chordae, and displacement

Valvular Stenosis

- Symptoms
  - Shortness of breath
  - Weakness/dizziness
  - Chest discomfort
  - Heart palpitations
  - Edema of the lower extremities or abdomen
  - Rapid weight gain

Example

- One-year-old Jack is brought in by his parents for treatment options for his congenital aortic stenosis. First noted with a heart murmur, EKG, Echo, and MRI indicated aortic stenosis. Balloon valvuloplasty is discussed and Jack’s parents are agreeable to the procedure.
  - Q23.0 Congenital stenosis of aortic valve
Example

- An 2-year-old girl is referred for cyanosis and shortness of breath since birth. In her first year, her mother states she was a slow feeder, at times becoming very blue. She did not walk until just recently. On exam she is normally developed and exhibited central cyanosis and clubbing equal in in the upper an lower halves of her body. Thrill and murmur detected in left of the lower end of the sternum. Diagnostics included X-ray, ECG, and cardiac cath confirm congenital tricuspid stenosis.

Q22.4 Congenital tricuspid stenosis

Documentation

- Clinical documentation should include:
  - Valve(s) affected
  - Other associated conditions
    - Insufficiency
    - atresia
  - Acquired or congenital
    - Different codes

ICD-10-CM Coding

- Q22.1 Congenital pulmonary valve stenosis
- Q22.4 Congenital tricuspid stenosis
- Q23.0 Congenital stenosis of aortic valve
- Q23.2 Congenital mitral stenosis
Example

An 11-year old boy presented for closure of an atrial septal defect after a transthoracic echocardiogram showed an ostium secundum atrial septal defect. The defect measured 13 x 14 mm by TEE. A 14 mm AMPLATZER® Septal Occluder (ASO) was chosen and deployed across the defect.

Q21.1 Atrial septal defect

Example

During a routine physical exam for participation in interscholastic sports, the physician noted that a twelve-year-old boy had a long continuous heart murmur at the second intercostal space near the left sternal border. A systolic thrill was also noted in the same region. When questioned, the patient's mother recalled that the child had periods of cyanosis and breathlessness as an infant. The child mentioned that he tires easily during physical activity. Doppler ultrasound was ordered, which revealed a patent ductus arteriosus.

Q25.0 Patent ductus arteriosus

Congenital heart defects

- Transposition of great vessels (TGV) – Q20.3
  - Refers to the aorta and pulmonary artery
  - The vessels arise from the wrong ventricle (transposed from normal position)
- Tetralogy of Fallot (TOF) – Q21.3
  - Combination of 4 related heart defects
  - Pulmonary stenosis, ventricular septal defect, overriding aorta, and right ventricular hypertrophy
Example

- A four month old male born at 40 weeks gestation was brought in for a check-up. Shortly after birth, the patient appeared cyanotic which led to further examination. A cardiac catheterization was performed and indicated Tetralogy of Fallot. Parents bring the baby in today for examination and discussion on next surgical interventions needed.

Q21.3 Tetralogy of Fallot

Neural tube defects (NTDs)

Neural tube defects

- Abnormalities that occur in the brain, spinal cord, and spinal column of a developing embryo and present at birth
- Can occur in approximately one in 1,000 live births
- Two types:
  - Open and closed

Neural tube defects (NTDs)

- Anencephaly (Q00.0) is a severe, but less common type of NTD that occurs when a baby is born without parts of the brain and skull because the upper part of the neural tube does not completely close. These babies are oftentimes born without a forebrain and cerebrum. If the infant is not stillborn, it is usually blind, deaf, and unconscious and dies shortly after birth.
Neural tube defects (NTDs)

• Craniorachischisis (Q00.1) is a congenital fissure of the skull and vertebral column. If the defect is limited to the cranial vault, it is defined as anencephaly. If it extends beyond the cranium, then it is defined as cranioschischisis.

Neural tube defects (NTDs)

• Iniencephaly (Q00.2) is a severe and rare type of NTD that occurs when the infant's head is severely bent backwards and the spine is greatly distorted. A baby with this type of NTD is most times born without a neck, with the skin of the face directly connected to the chest and the scalp connected to the back. Babies born with this defect may also have other types of abnormalities (cleft lip/palate, heart irregularities, intestinal malformations, etc) and usually do not survive more than a few hours.

Neural tube defects (NTDs)

• The encephalocele codes are broken down by site of occurrence:
  – Frontal encephalocele – Q01.0
  – Nasofrontal encephalocele – Q01.1
  – Occipital encephalocele – Q01.2
  – Other sites – Q01.8
### Neural tube defects (NTDs)

- Spina bifida is the most common type of neural tube defect. The two most common types are:
- Meningocele – A sac of fluid protrudes through an opening in the infant’s back, but the spinal cord is not in the sac.
- Myelomeningocele – The most serious type of spina bifida. As with a meningocele, a sac of fluid protrudes through an opening in the infant's back. Unlike a meningocele, though, part of the spinal cord and nerves are in the sac and are damaged.

### Neural tube defects (NTDs)

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
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<tbody>
<tr>
<td>Q05.0</td>
<td>Cervical spina bifida with hydrocephalus</td>
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<tr>
<td>Q05.1</td>
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</tr>
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<tr>
<td>Q05.4</td>
<td>Unspecified spina bifida with hydrocephalus</td>
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<td>Q05.5</td>
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<td>Q05.6</td>
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<td>Q05.9</td>
<td>Spina bifida unspecified</td>
</tr>
</tbody>
</table>

### Example

- Lance is born by cesarean section. He has bulging fontanels, a head circumference of 40cm, and a chest circumference of 34cm. In the lumbar spine region a sac-like projection is noted. Lance is taken to the Pediatric ICU with a diagnosis of lumbar myelomeningocele and hydrocephalus.

Q05.2 Lumbar spina bifida with hydrocephalus
Neural tube defects (NTDs)
• Closed neural tube defects occur when the spinal defect is covered by skin. These are rarer than open NTDs and include:
  – Amyelia (Q06.0)
  – Hypoplasia and dysplasia of spinal cord (Q06.1)
  – Diastematomyelia (Q06.2)
  – Hydromyelia (Q06.4)
  – Lipomyelomeningocele/Lipomeningocele (Q06.8)
  – Tethered cord (Q06.8)

Neural tube defects (NTDs)
• Spina bifida occulta (SBO) (Q76.0) is not considered a true neural tube defect and is not located with the congenital malformations of the nervous system. There is a small gap in the spine, but the meninges do not herniate through the spine. It is located in category Q76, *Congenital malformations of the spine and bony thorax*.

Example
• Martin is born with what appears to be sacral dimple with a tuft of hair. An ultrasound was performed and spina bifida occulta is confirmed.

  Q76.0 Spina bifida occulta
Diseases and Disorders

- Spina Bifida
  - Spinal cord and meninges (spinal cord tissue) stick out of the child’s back
  - Many children also have a condition called hydrocephalus

ICD-10-CM Coding

- Code choice selections include:
  - Site
  - Presence of hydrocephalus, as necessary

ICD-10-CM Examples

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Down syndrome

**Down Syndrome**

- Genetic condition
- Person is born with an extra copy of chromosome 21
- WHO estimates the incidence of Down syndrome between 1 in 1,000 to 1 in 1,100 live births worldwide

**Example**

- 5½ year-old Lola is brought in for a check-up and referral. She has nonmosaic Down syndrome and Tetralogy of Fallot. She has had multiple surgeries since birth due to the congenital heart defect and she needs Cardiology evaluation for correction of her ventricular septal defect.

- **Q90.0 Trisomy 21, nonmosaicism**
- **Q21.3 Tetralogy of Fallot**
Other trisomies
- Trisomy 18, nonmosaicism Q91.0
- Trisomy 18, mosaicism Q91.1
- Trisomy 18, translocation Q91.2
- Trisomy 18, unspecified Q91.3
- Trisomy 13, nonmosaicism Q91.4
- Trisomy 13, mosaicism Q91.5
- Trisomy 13, translocation Q91.6
- Trisomy 13, unspecified Q91.7

Cleft lip and cleft palate
- Cleft Lip
  - Unilateral
  - Bilateral
  - Median
- Hard Palate
- Soft Palate

Cleft lip and cleft palate
- Q35.1 Cleft hard palate
- Q35.2 Cleft soft palate
- Q35.5 Cleft hard palate with cleft soft palate
- Q36.0 Cleft lip, bilateral
- Q36.1 Cleft lip, median
- Q36.9 Cleft lip, unilateral
- Q37.0 Cleft hard palate with bilateral cleft lip
- Q37.1 Cleft hard palate with unilateral cleft lip
- Q37.2 Cleft soft palate with bilateral cleft lip
- Q37.3 Cleft soft palate with unilateral cleft lip
- Q37.4 Cleft hard and soft palate with bilateral cleft lip
- Q37.5 Cleft hard and soft palate with unilateral cleft lip
Example

• Marcus is brought in for a check-up. He has a unilateral cleft lip with a cleft hard palate.

Q37.1 Cleft hard palate with unilateral cleft lip

Undescended and ectopic testes

• Cryptorchidism – undescended testicle(s)
  – About 3-5% of term male infants
  – About 30% of premature males
  – May spontaneously descend within first 3 months
• Ectopic testis – deviation from normal path of descent
  – Cannot be manipulated into the scrotum
  – Usually well developed and histologically normal

Undescended and ectopic testes

• Retractile testis (Q55.22) is not actually an undescended testicle, rather a testicle that descended normally, but that is connected to more active muscles that cause the testicle to move in and out of the base of the scrotum. They function normally and usually descend and relax in the scrotum permanently during puberty.
Undescended and ectopic testes

- Q53.01 Ectopic testis, unilateral
- Q53.02 Ectopic testes, bilateral
- Q53.11 Abdominal testis, unilateral (the testis is undescended and is in the abdominal cavity)
- Q53.12 Ectopic perineal testis, unilateral (the testis has descended, but is located in an abnormal position in the perineal area)
- Q53.21 Abdominal testis, bilateral
- Q53.22 Ectopic perineal testis, bilateral

Example

- Mason was born with a left undescended abdominal testis. His parents bring him in for a check on his condition as he now is three months old. Upon examination, the testis has still not descended, so referral is made to a Pediatric Urologist.

  Q53.11 Abdominal testis, unilateral

Polydactyly and syndactyly

- Polydactyly and syndactyly – most common malformations affecting the hands and feet
- Syndactyly – fusing together or webbing of fingers or toes
- Polydactyly – presence of extra fingers or toes
  - Postaxial – extra digit next to the little finger
  - Preaxial – extra digit near the thumb
Polydactyly and syndactyly

There are four codes for polydactyly:
- Q69.0 Accessory finger(s)
- Q69.1 Accessory thumb(s)
- Q69.2 Accessory toes(s)
- Q69.9 Polydactyly, unspecified

Polydactyly and syndactyly

• Syndactyly codes are located in category Q70:
  – Fused fingers and fused toes (Q70.0-, Q70.2-): also called complex syndactyly
  – Webbed fingers and webbed toes (Q70.1-, Q70.3-): also called simple syndactyly

Polydactyly and syndactyly

• There is a code for polysyndactyly (Q70.4), which is multiple webbed and/or fused digits.
  • Excludes 1 – specified syndactyly of hands and feet – code to specified conditions (Q70.0- - Q70.3-)
Example

- Julie is born via vaginal delivery without complications. At birth, it is noted that she has fusion of fingers on her left hand and webbed toes on her left foot.

  Q70.02 Fused fingers, left hand
  Q70.32 Webbed toes, left foot

Questions?

You Can Do It!