Birth defects surveillance training facilitator's guide, second edition

Module 4: Congenital anomalies and infectious syndromes (slides 2 – 94)

Module 5: Coding (slides 95 – 102)

Module 6: Three clinical scenarios: Baby Adaeze, Baby Esther, and Baby Matias (slides 103 – 220)

Module 7: Data quality exercises (slides 221 – 248)

Source for all illustrations (unless otherwise noted): CDC, NCBDDD
The findings and conclusions in this report are those of the authors and do not necessarily represent the official position of the United States Centers for Disease Control and Prevention.

World Health Organization

CDC

International Clearinghouse for Birth Defects Surveillance and Research

Facilitator's Guide

Module 4: Congenital anomalies and infectious syndromes

World Health Organization

CDC

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Objectives

By the end of this module, participants will be able to:

- describe basic congenital anomaly characteristics (internal vs. external findings);
- use the description checklist in the QRH to describe features and subtypes of selected congenital anomalies; and
- understand which codes should be used for each of the internal and external congenital anomalies.

Major congenital anomalies for monitoring

- When collecting public health surveillance data for congenital anomalies, the quality of the data is as important as the quantity.
- High-quality data on a smaller number of congenital anomalies will be more useful to public health congenital anomaly surveillance than poor-quality data on all congenital anomalies.
- [INSERT VIDEO link that best fits your audience, see Facilitator's Guide]
- For each anomaly, we will discuss how congenital anomalies are coded utilizing the International statistical classification of diseases and related health problems, 10th edition (ICD-10), and the Royal College of Paediatrics and Child Health (RCPCH) adaptation.

Congenital Anomalies

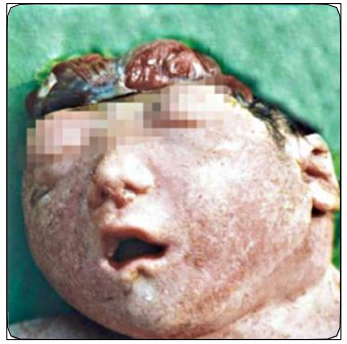
- Neural tube defects
  - Anencephaly, encephalocele, spina bifida
  - Craniorachischisis, iniencephaly
- Microcephaly
- Microtia/anotia
- Congenital heart defects
  - Hypoplastic left heart syndrome, interrupted aortic arch, common truncus
  - Pulmonary valve atresia, tricuspid atresia
  - Tetralogy of Fallot, transposition of great arteries
- Orofacial clefts
  - Cleft lip, cleft palate, cleft lip with cleft palate
- Digestive system anomalies
  - Oesophageal atresia/trachea-oesophageal fistula
  - Large intestinal atresia
  - Anorectal atresia
- Genital and urinary anomalies
  - Hypospadias
  - Renal agenesis and hypoplasia
- Musculoskeletal anomalies
  - Talipes equinovarus
  - Limb reduction defects: amelia, transverse, longitudinal
- Abdominal defects
  - Omphalocele
  - Gastroschisis
- Down syndrome (trisomy 21)

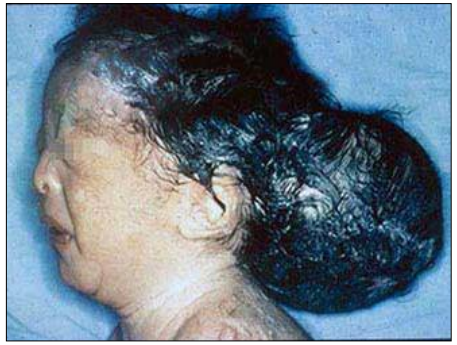
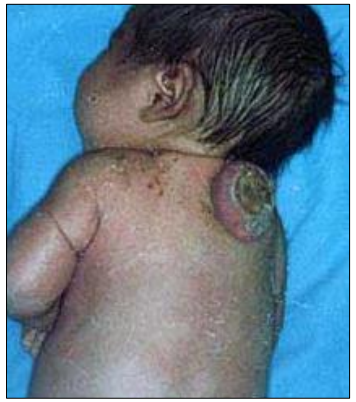
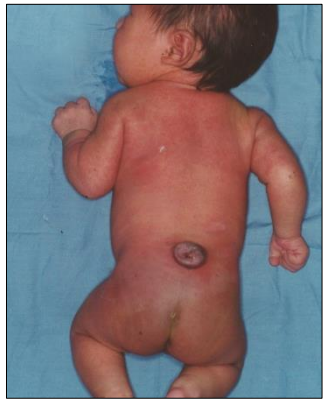
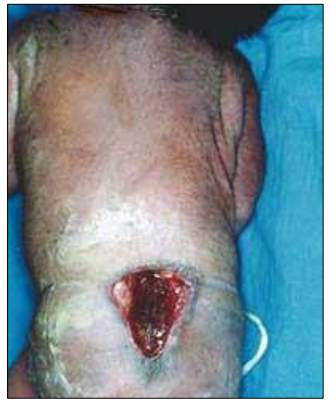
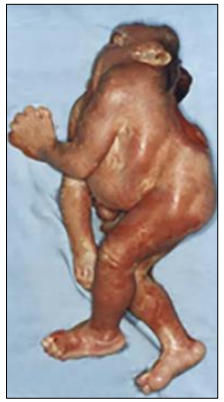
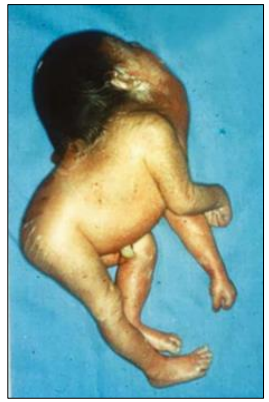
Congenital Infectious Syndromes

- Rubella
- Syphilis
- Cytomegalovirus (CMV)
- Zika

Neural tube defects

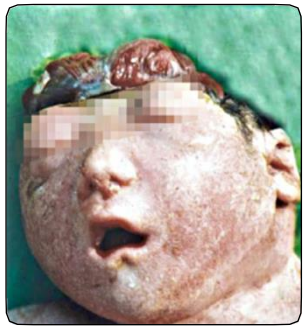
Activity 4.1



Source: Latin American Collaborative Study of Congenital Malformations, CDC, Beijing Medical University collaborative project/Dr Jaime Frias

Neural tube defects

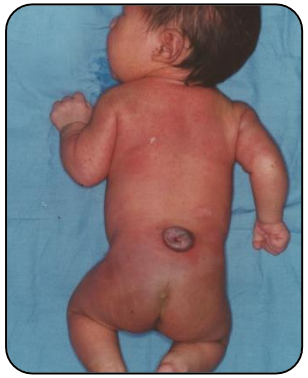


Anencephaly

- Characterized by either total or partial absence of the brain, together with total or partial absence of the cranial vault and the covering skin
- Anatomically normal eyes but appear to be bulging
- Two types of anencephaly can be distinguished:
  - holoanencephaly (total absence)
  - meroanencephaly (partial absence)

Encephalocele

- Skin-covered, pedunculated or sessile cystic lesion protruding through a defect in the skull
- Distinguished based on their location on the skull (e.g., occipital, parietal, frontal, etc.)

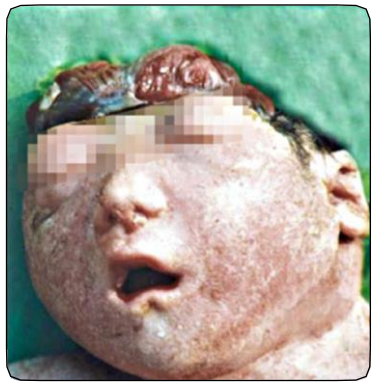


Spina bifida

- A general term used to describe a neural tube defect of the spine, in which part of the meninges or spinal cord, or both, protrude through an opening in the vertebral column
- Lesion can be open or closed; this photograph shows an open lesion (no skin covering) in the lumbar region of the spine

Source: Latin American Collaborative Study of Congenital Malformations

Neural tube defects



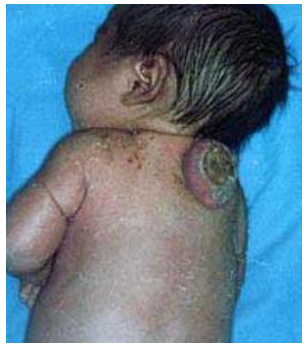
Relevant ICD-10 codes

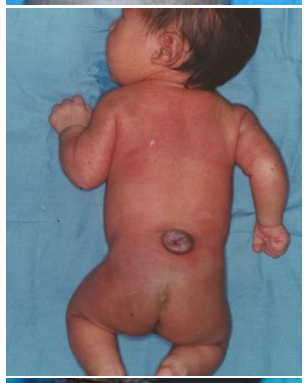
- Q00.0 Anencephaly

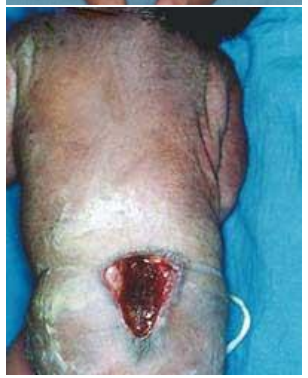
- Q01.0 Frontal encephalocele
- Q01.1 Nasofrontal encephalocele
- Q01.2 Occipital encephalocele
- Q01.80 Parietal encephalocele
- Q01.81 Orbital encephalocele
- Q01.82 Nasal encephalocele
- Q01.83 Nasopharyngeal encephalocele
- Q01.9 Encephalocele, unspecified

Sources*:* Latin American Collaborative Study of Congenital Malformations; Dr Jaime Frias

Neural tube defects





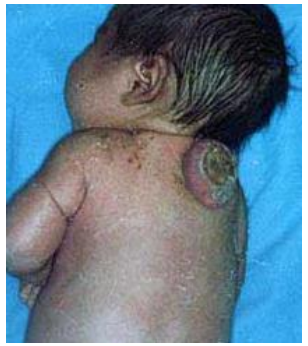


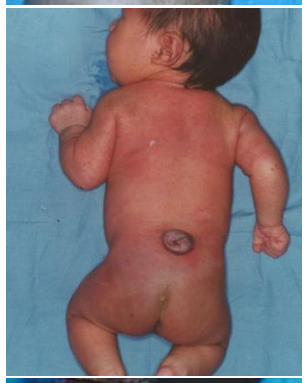
Relevant ICD-10 codes

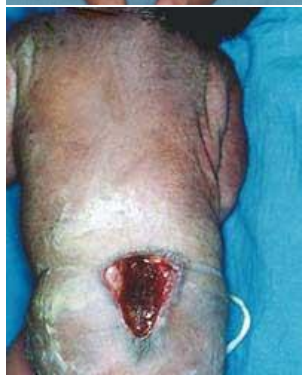
- Q05.0 Cervical spina bifida with hydrocephalus
- Q05.1 Thoracic spina bifida with hydrocephalus
- Q05.2 Lumbar spina bifida with hydrocephalus (inc. lumbosacral spina bifida w/ hydrocephalus)
- Q05.3 Sacral spina bifida with hydrocephalus
- Q05.4 Unspecified spina bifida with hydrocephalus
- Q05.5 Cervical spina bifida without hydrocephalus
- Q05.6 Thoracic spina bifida without hydrocephalus
- Q05.7 Lumbar spina bifida without hydrocephalus (inc. lumbosacral spina bifida w/o hydrocephalus)
- Q05.8 Sacral spina bifida with hydrocephalus
- Q05.9 Spina bifida, unspecified

Source: CDC, Beijing Medical University collaborative project

Neural tube defects





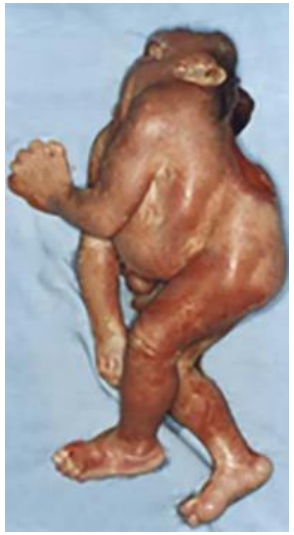


Relevant ICD-10 codes – Order option

- Q05.0 Cervical spina bifida with hydrocephalus
- Q05.5 Cervical spina bifida without hydrocephalus
- Q05.1 Thoracic spina bifida with hydrocephalus
- Q05.6 Thoracic spina bifida without hydrocephalus
- Q05.2 Lumbar spina bifida with hydrocephalus (inc. lumbosacral spina bifida w/ hydrocephalus)
- Q05.7 Lumbar spina bifida without hydrocephalus (inc. lumbosacral spina bifida w/o hydrocephalus)
- Q05.3 Sacral spina bifida with hydrocephalus
- Q05.8 Sacral spina bifida with hydrocephalus
- Q05.4 Unspecified spina bifida with hydrocephalus
- Q05.9 Spina bifida, unspecified

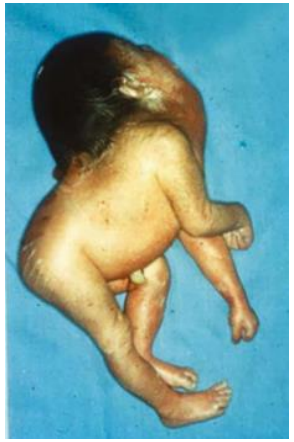
Photograph source: CDC-Beijing Medical University collaborative project

Neural tube defects



Craniorachischisis

- Very rare type of neural tube defect
- Includes anencephaly (absence of brain and cranial vault, without skin covering) with a continuous bony defect of the spine (not covered by meninges)
- May be limited to the cervical spine or can extend more distally
- May be retroflexion of the neck.

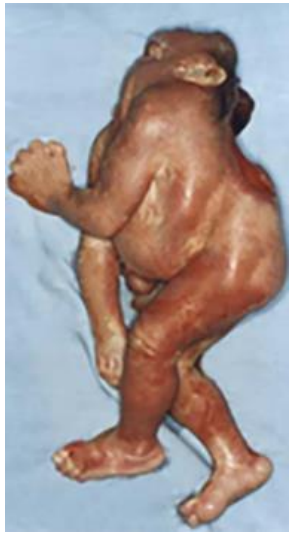


Iniencephaly

- Another very rare type of neural tube defect
- Typically a closed defect (skin covered) that involves the occiput and inion (tip of the occipital protuberance)
- Key findings on external examination are the extreme retroflexion of the head, with skin of the face connected to the chest skin and skin of the scalp connected with the skin of the back

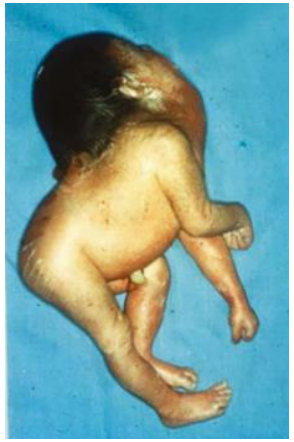
Source: CDC-Beijing Medical University collaborative project

Neural tube defects



Relevant ICD-10 codes

- Q00.1 Craniorachischisis



- Q00.2 Iniencephaly
- Q00.20 Iniencephaly, open
- Q00.21 Iniencephaly, closed

Source: CDC-Beijing Medical University collaborative project

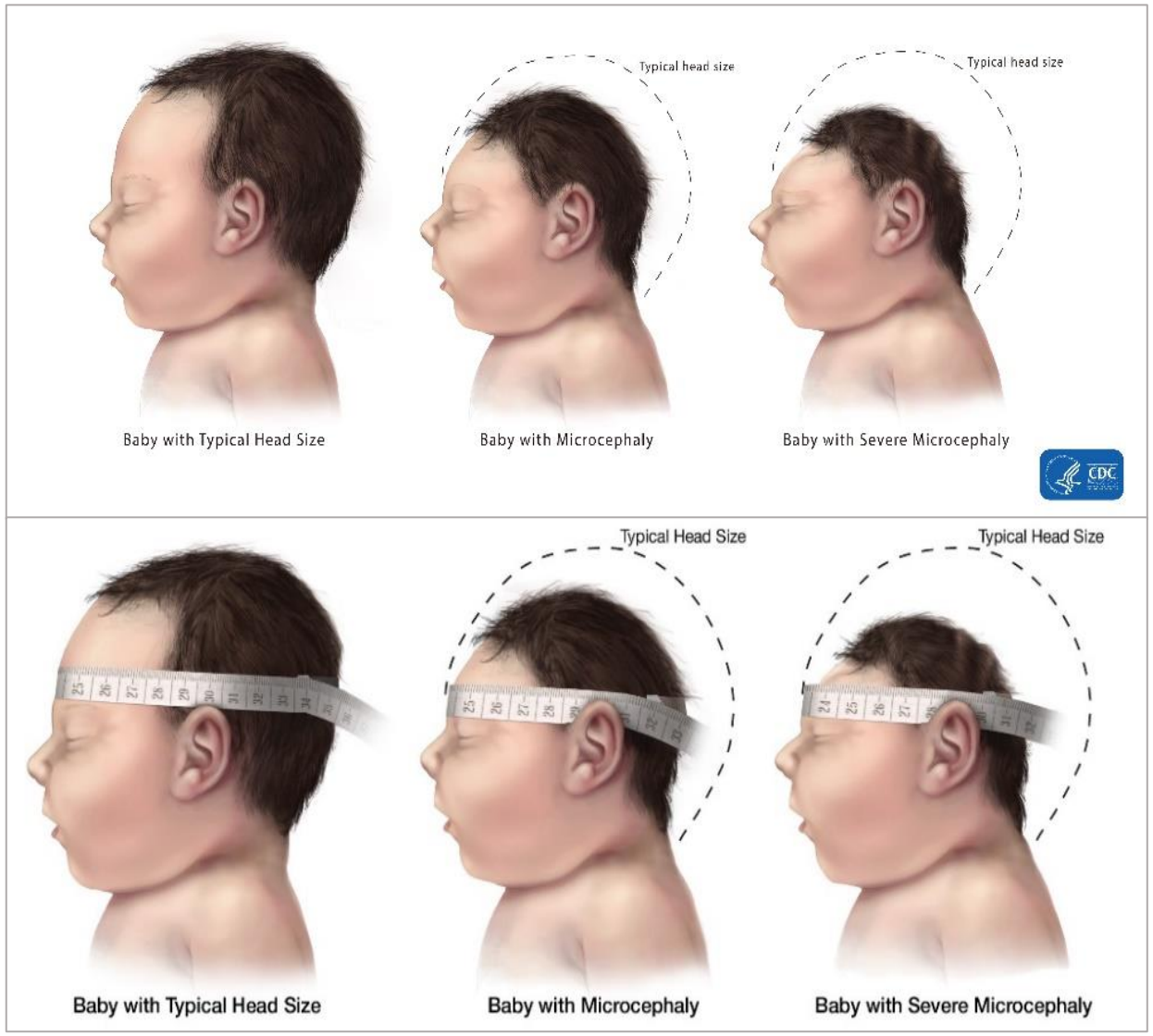
Neural tube defects

Description or documentation checklist for high-quality reporting			
	Anencephaly[a]	**Encephalocele[b]**	**Spina bifida[ab]**
Describe in detail	• Extent (holo vs. mero) • Cervical spine (document no contiguous defects) • Location (whether a non-contiguous spina bifida is present) • Whether amniotic bands are present	• Defect location • Extent (size and whether brain is present in sac) • Skin covering (ruptured or intact) • Other anomalies • Scalp swelling • Whether amniotic bands or limb-body wall anomalies are present	• Defect location • Size of lesion • Skin covering (skin or membrane) • Lesion content (meninges +/- spinal cord) • Hydrocephalus, talipes, other • Other unrelated anomalies
Take or report photographs	• Show clearly missing cranium (skull bones)	• Show cranial lesion	Show level of spina bifida on back and side
Describe evaluations	• Consultations • Protruding eyes but normal development (do not include as an associated anomaly) • Small head circumference	• Consultations and surgery • Imaging • Genetic or chromosomal testing • Head circumference might be small – do not code for microcephaly • Hydrocephalus might be present – do not code as hydrocephalus	Consultations, imaging and surgery Genetic or chromosomal conditions
Autopsy findings	• Report if available	• Report if available	• Report if available

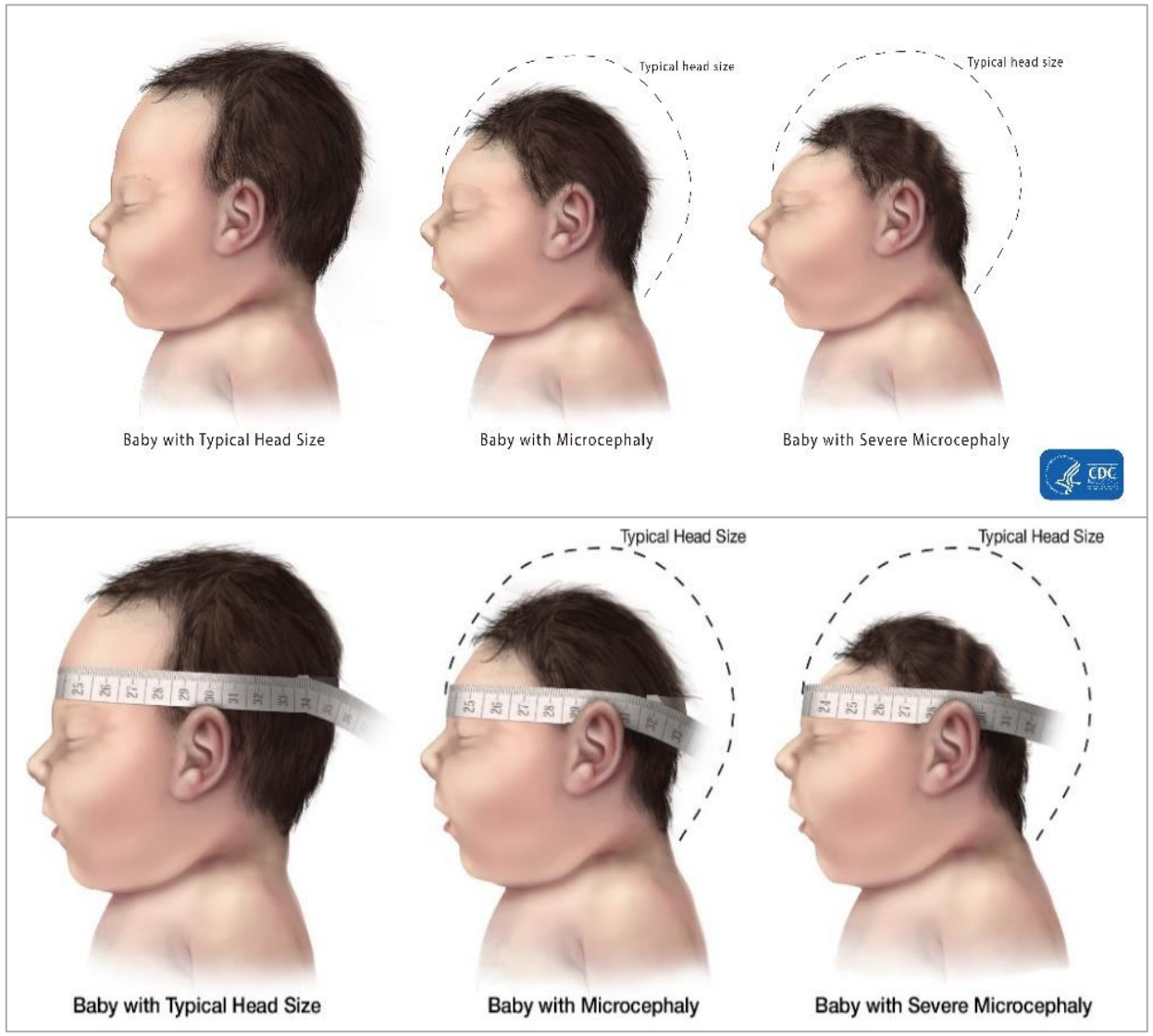
[a]Craniorachischisis: refer to anencephaly and spina bifida descriptions above

[b]Iniencephaly: refer to encephalocele description above and describe if retroflexion, facial and scalp skin connections are present

Microcephaly



Microcephaly



Microcephaly:

- Characterized by a small cranial vault based on the baby's gestational age and sex
- Position of the ears are normal

Measuring the head circumference:

- Use a tape measure that does not stretch
- Measure at the widest part of the head
- Measure three separate times
- Compare head circumference to a known standard for gestational age and sex:
  - INTERGROWTH-21st -- https://intergrowth21.tghn.org/articles/intergrowth-21st-head-circumference-standards-boys/
  - WHO -- *https://www.who.int/tools/child-growth-standards/standards/head-circumference-for-age)*

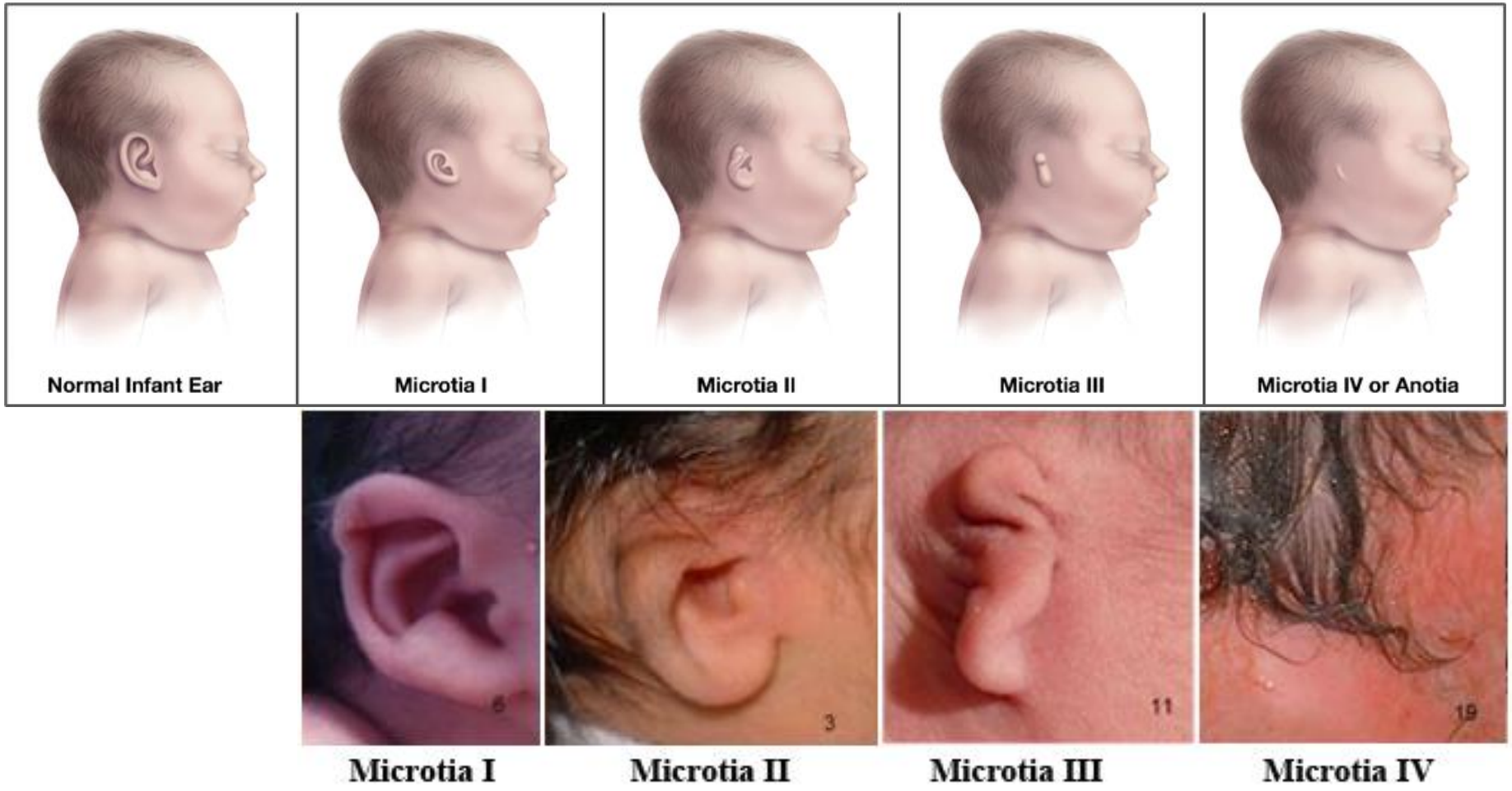
Relevant ICD-10 code:

- Q02 Microcephaly

Microcephaly

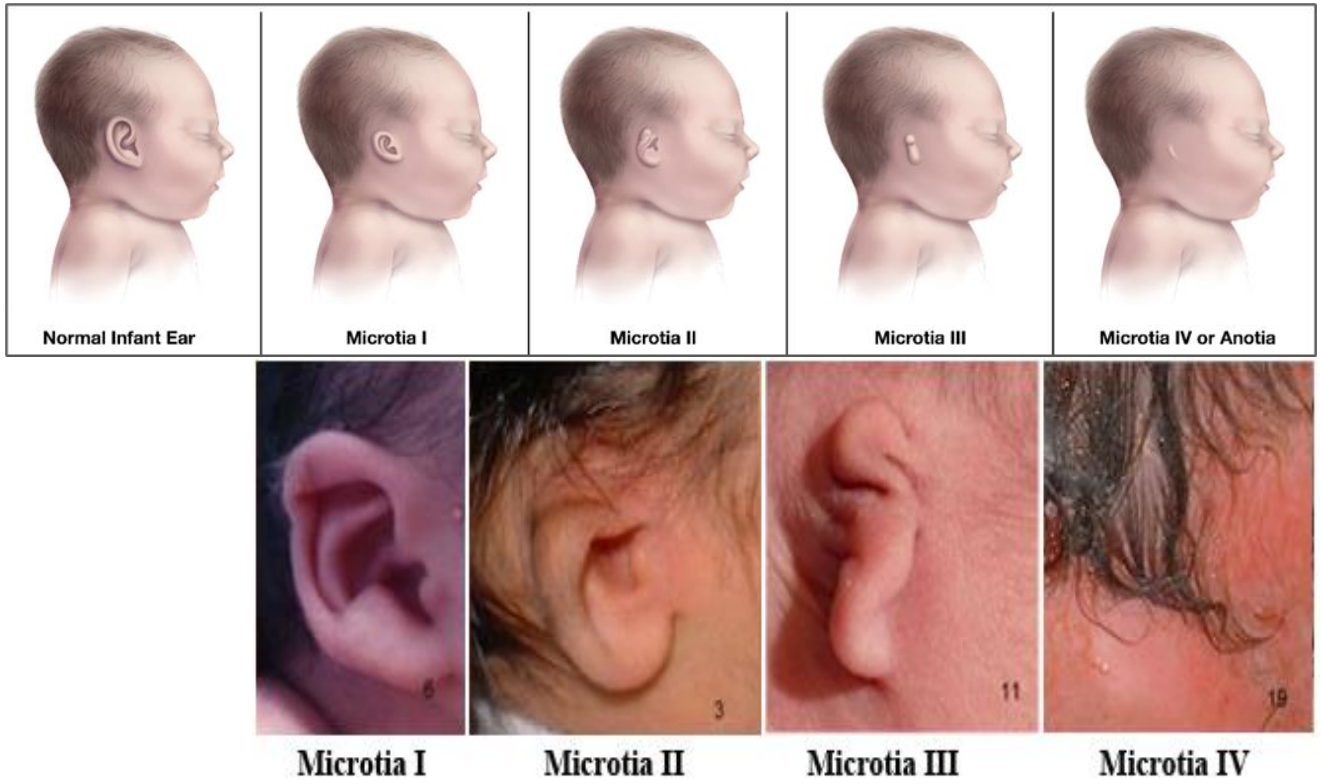
Description or documentation checklist for high-quality reporting	
	Microcephaly and severe microcephaly
Describe in detail	. Measure and document head circumference . Standard: measure head circumference within 24 hours after birth . Document head circumference or SD by gestational age and sex (based on INTERGROWTH-21st standards (https://intergrowth21.tghn.org/articles/intergrowth-21st-head-circumference-standards-boys/) or WHO (https://www.who.int/tools/child-growth-standards/standards/head-circumference-for-age) . Distinguish microcephaly from craniosynostosis . Report neurological status and signs (e.g., tone, seizures, irritability)
Take or report photographs	. Show full face and body
Describe evaluations	. Consultations and brain imaging studies . Genetic or chromosomal conditions . Laboratory findings
Autopsy findings	. Report if available

Normal ear, microtia and anotia



Photographs source: Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

Microtia/anotia

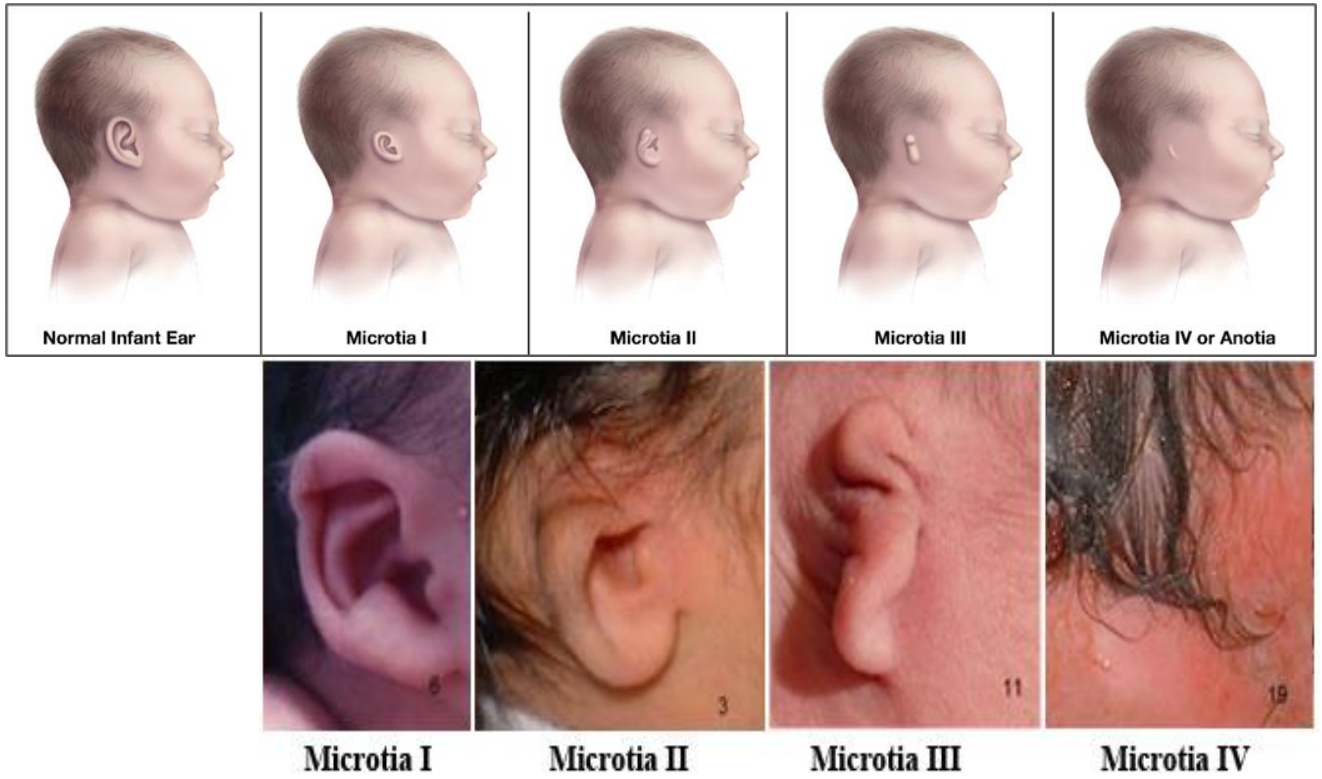


Four types ranging from mild to severe:

- **Microtia I**
  Small ear: generally, not included in public health surveillance system
- **Microtia II**
  Small ear with missing components and abnormally shaped
- **Microtia III**
  Vertical mass of soft tissue and cartilage, associated with external canal atresia
- **Microtia IV (Anotia)**
  Absent external or rudimentary external structure and blind-ending external canal

Photographs source: ECLAMC

Microtia/anotia



Relevant ICD-10 codes

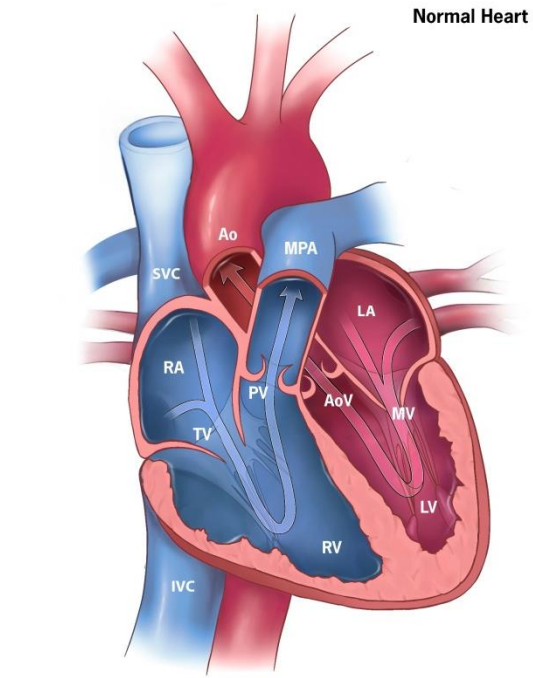
- Q16.0 Congenital absence of external ear (auricle) Type IV/Anotia
- Q17.0 Microtia
- Q17.21 Microtia, first degree
- Q17.22 Microtia, second degree
- Q17.23 Microtia, third degree

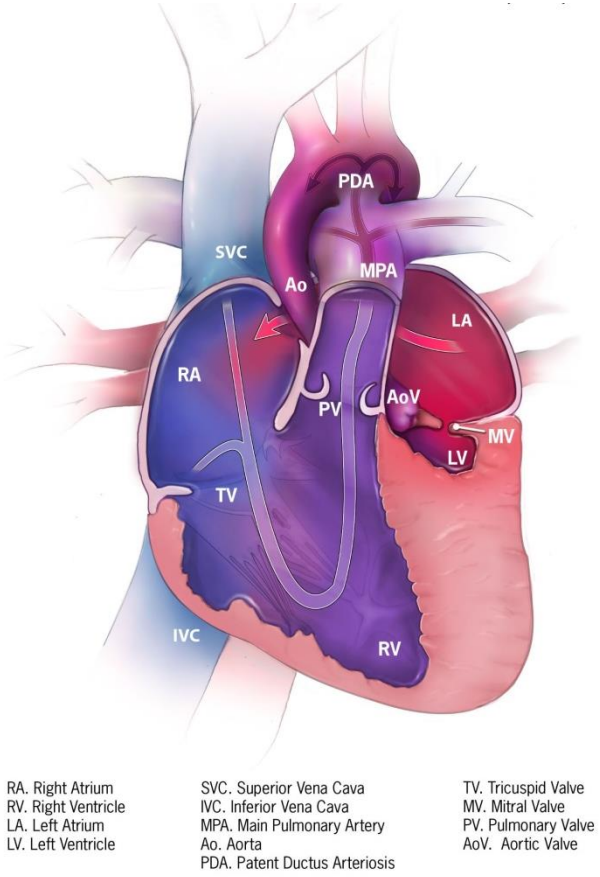
Photographs source: ECLAMC

Microtia/anotia

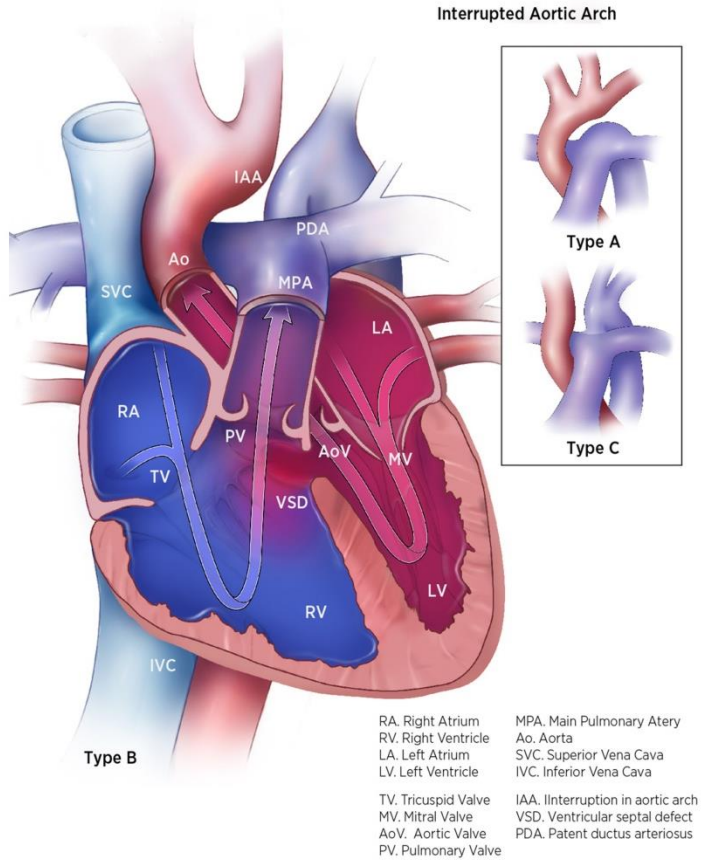
Description or documentation checklist for high-quality reporting	
	Microtia/anotia
Describe in detail	• Unilateral vs. bilateral • Describe external structure and severity • Ear canal present/absent • Check for preauricular tag or pit • Down slanting palpebral fissures, small jaw, eyelid coloboma (suggests syndrome) • Cervical vertebral anomalies – oculo-auriculo-vertebral spectrum (OAVS) suggested
Take or report photographs	Show side and front
Describe evaluations	• Consultations and surgical reports • Hearing tests • Genetic or chromosomal testing
Autopsy findings	Report if available

CHDs: hypoplastic left heart syndrome and interrupted aortic arch



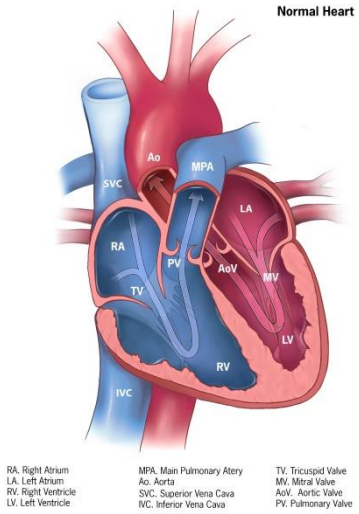


Hypoplastic left heart syndrome

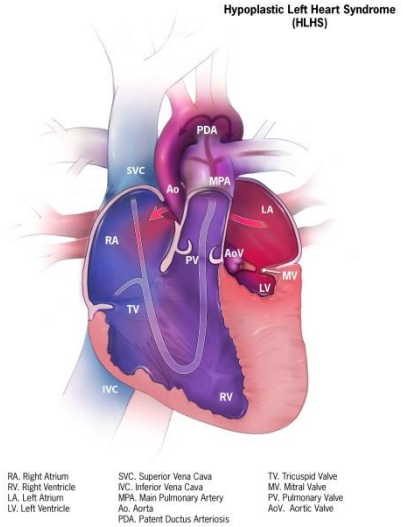


Interrupted aortic arch

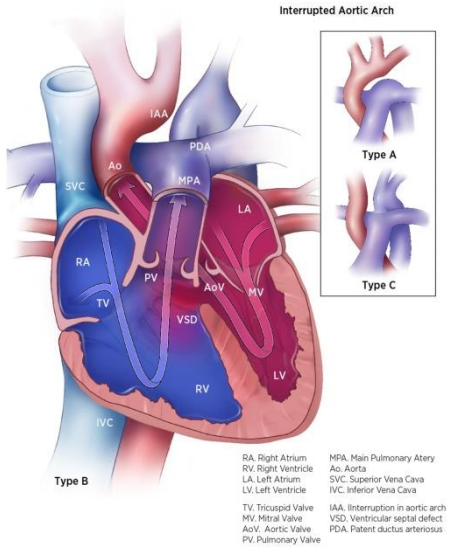
CHDs: hypoplastic left heart syndrome and interrupted aortic arch



Normal heart



Hypoplastic left heart syndrome



Interrupted aortic arch

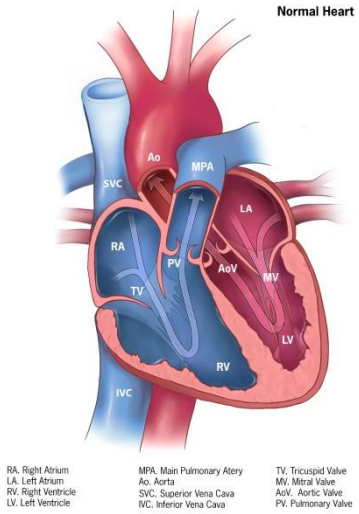
Hypoplastic left heart syndrome (HLHS)

- Small/underdeveloped left ventricle and aorta, mitral and/or aortic valve may be small or atretic

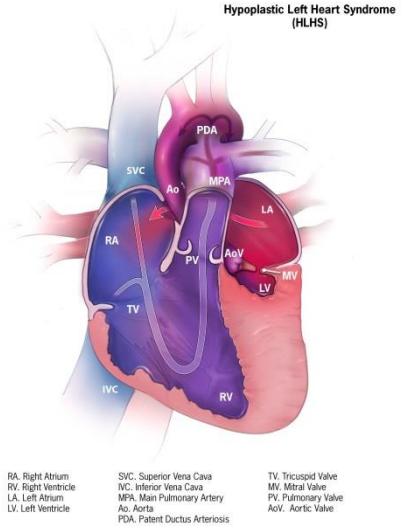
Interrupted aortic arch (IAA)

- Three types of IAA (A: distal; B: proximal [most common]; C: further proximal) with discontinuity along the aortic arch

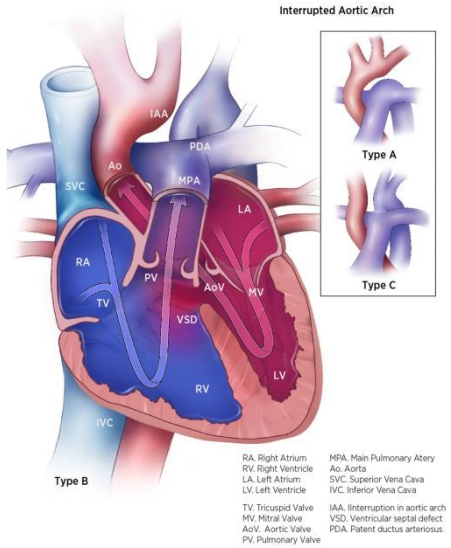
CHDs: hypoplastic left heart syndrome and interrupted aortic arch



Normal heart



Hypoplastic left heart syndrome



Interrupted aortic arch

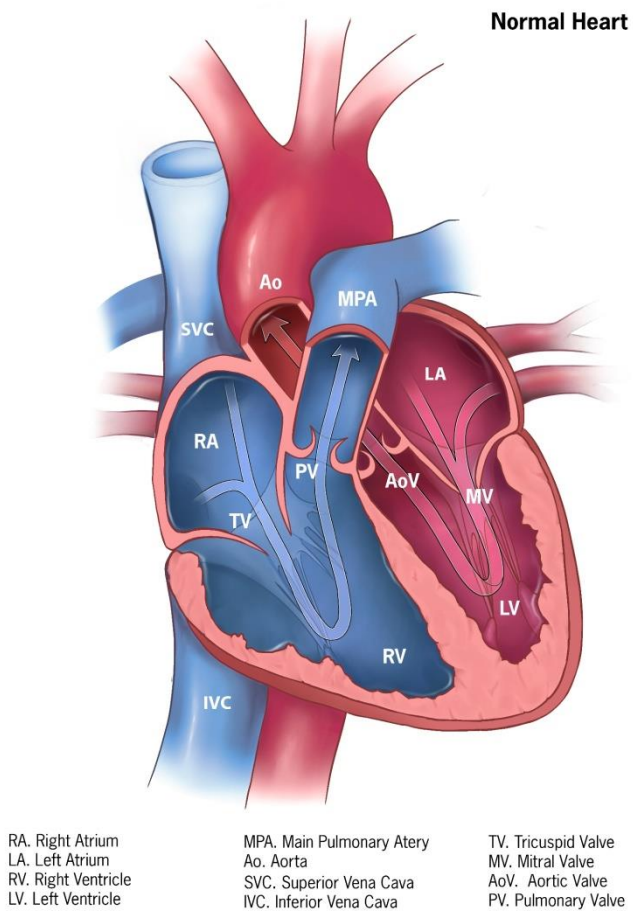
Relevant ICD-10 codes

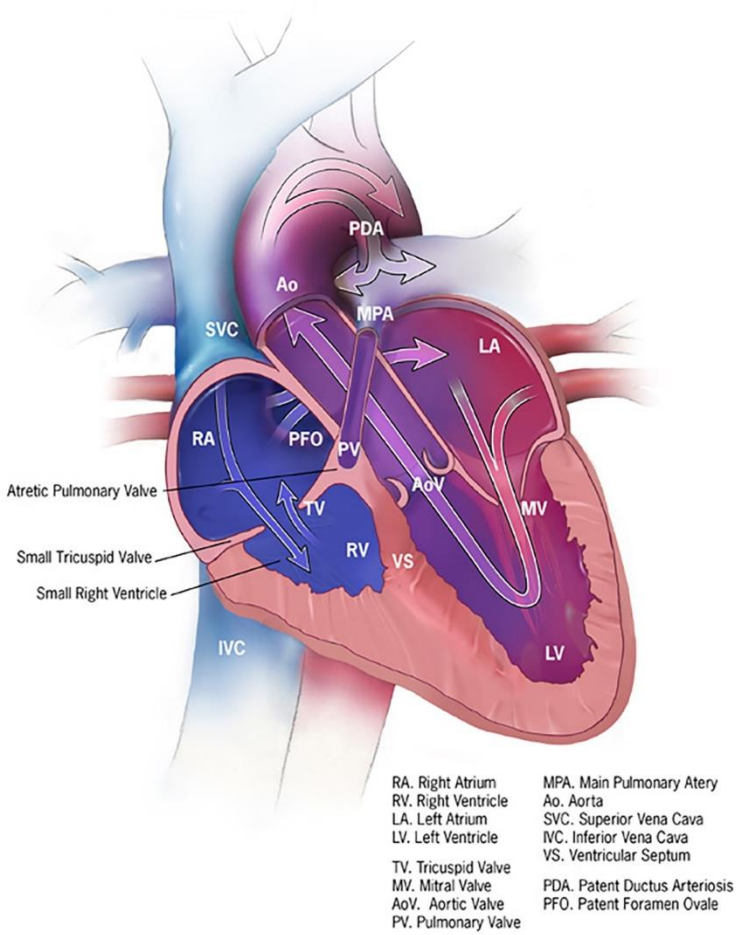
- Q23.4 Hypoplastic left heart syndrome
- Q25.21 Interrupted aortic arch
- Q25.4 Other congenital malformations of the aorta (heterogeneous; might include interrupted aortic arch)

CHDs: hypoplastic left heart syndrome and interrupted aortic arch

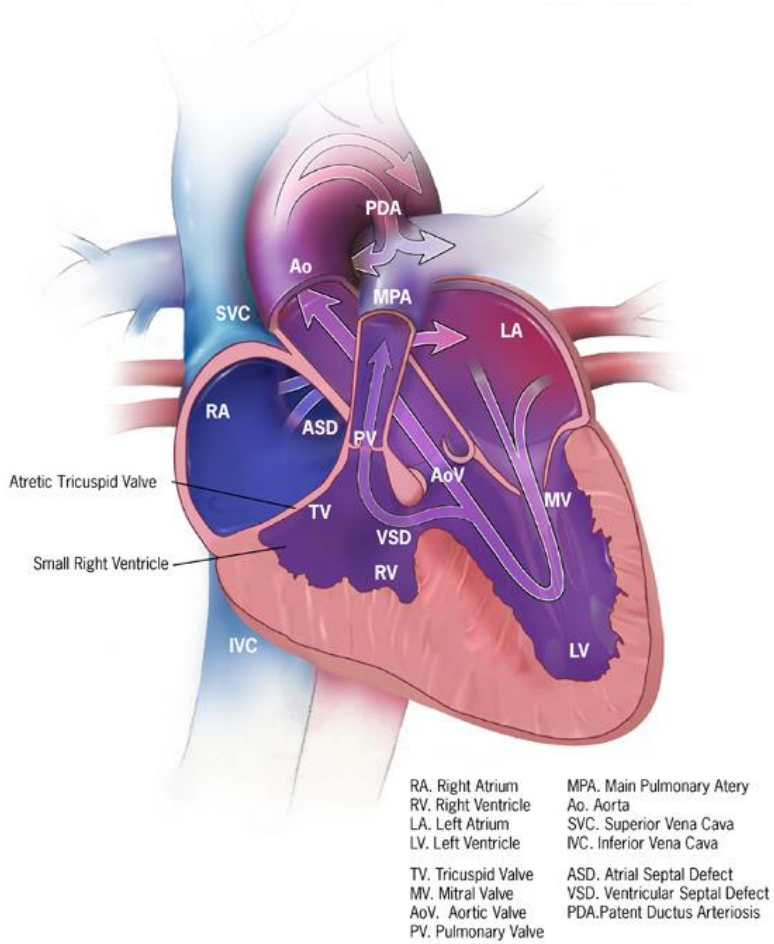
Description or documentation checklist for high-quality reporting		
	Hypoplastic left heart syndrome	**Interrupted aortic arch (IAA)**
Describe in detail	• Anatomy – specify intracardiac anomalies, valves (stenosis/atresia), aorta • Document extracardiac anomalies • Minor anomalies may suggest syndrome (e.g., Turner syndrome)	• Anatomy – specify site of discontinuity, type of IAA, intracardiac anomalies, if ventricular septal defect (VSD) present/type • Document extracardiac anomalies
Describe evaluations and procedures	• For each procedure, describe prenatal vs postnatal diagnosis • Consultations • Surgeries • Genetic testing	• For each procedure, describe prenatal vs postnatal diagnosis • Consultations • Surgeries • Genetic testing
Autopsy findings	Report if available	Report if available

CHDs: pulmonary valve atresia and tricuspid valve atresia



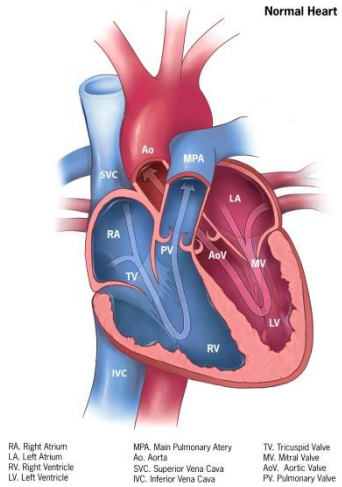


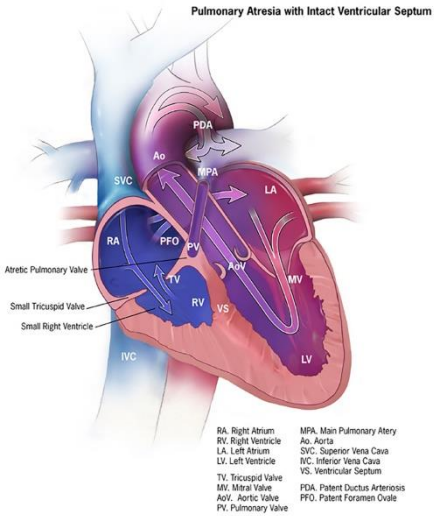
Pulmonary valve atresia

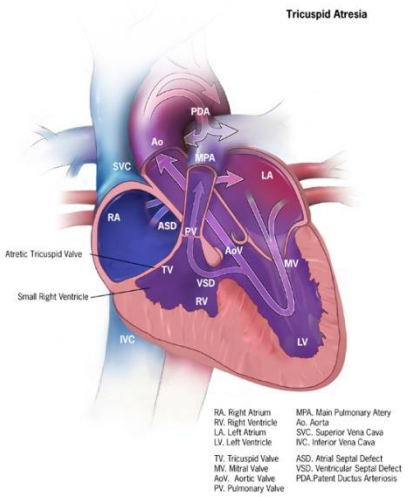


Tricuspid valve atresia

CHDs: pulmonary valve atresia and tricuspid valve atresia







Normal heart **Pulmonary valve atresia** **Tricuspid valve atresia**

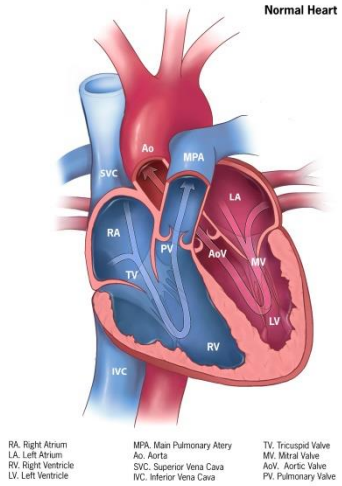
Pulmonary valve atresia

- Imperforate valve, may have ventricular septal defects (VSD), underdeveloped right ventricle

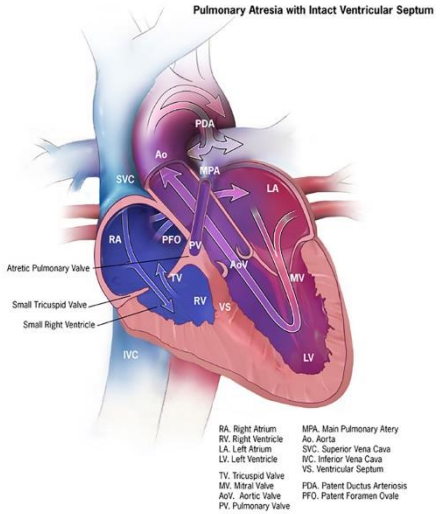
Tricuspid valve atresia

- Atretic valve, may have ventricular septal defects (VSD), stenosis/atresia of pulmonary valve, associated with complex heart anomalies (e.g., heterotaxy, del 22q11)

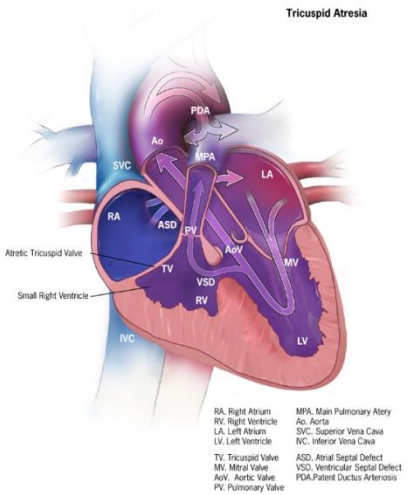
CHDs: pulmonary valve atresia and tricuspid valve atresia



Normal heart



Pulmonary valve atresia



Tricuspid valve atresia

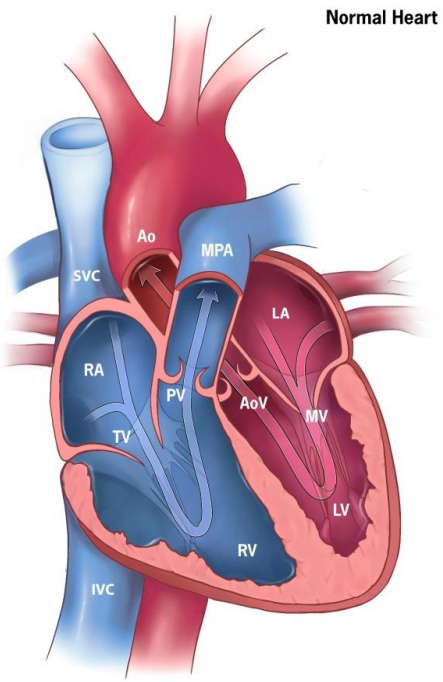
Relevant ICD-10 codes

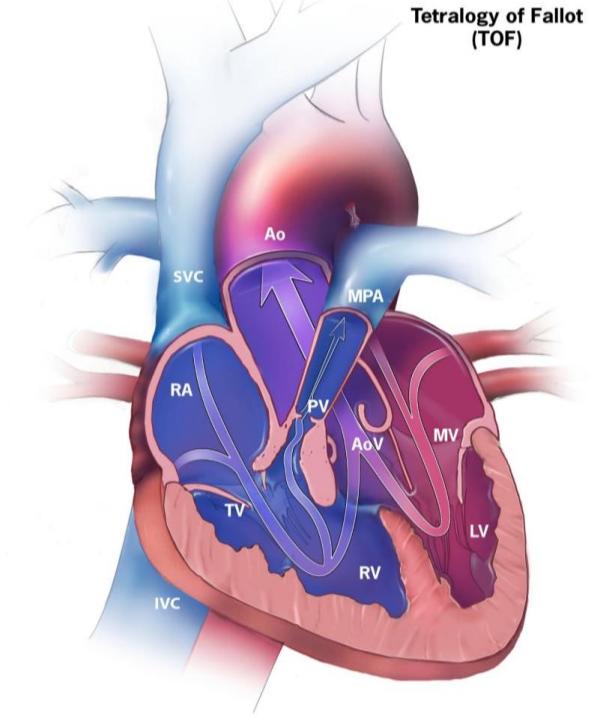
- Q22.0 Pulmonary valve atresia
- Q22.0 Pulmonary valve atresia with Q21.0 (ventricular septal defect)
- Q22.4 Tricuspid valve atresia

CHDs: pulmonary valve atresia and tricuspid valve atresia

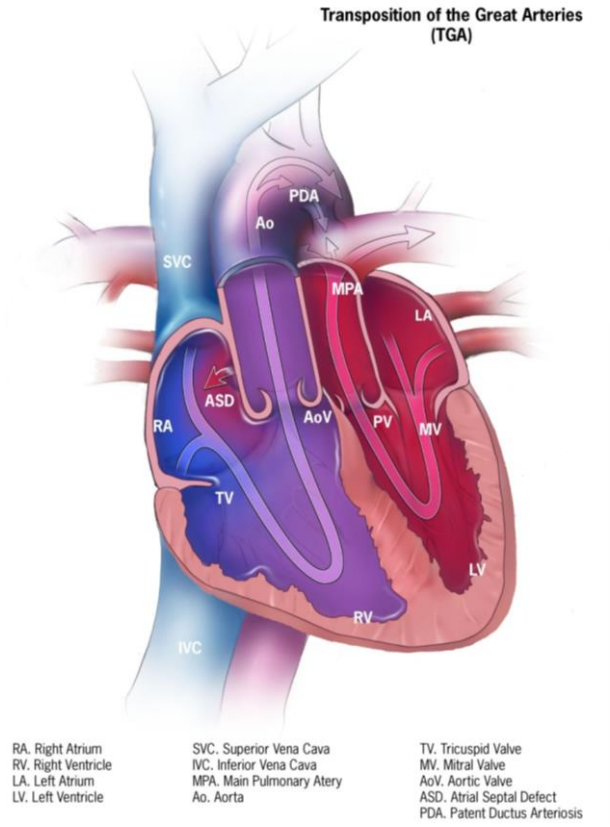
Description or documentation checklist for high-quality reporting		
	Pulmonary valve atresia	Tricuspid valve atresia
Describe in detail	. Intracardiac anatomy, valve atresia, tricuspid valve involvement, right ventricle, ventricular septal defect (VSD) . Document extracardiac anomalies	. Intracardiac anatomy, stenosis/atresia of pulmonary valve, right ventricle, atrial septal defect (ASD), ventricular septal defect (VSD) . Document extracardiac anomalies
Describe evaluations and procedures	. For each procedure, describe prenatal vs postnatal diagnosis . Consultations . Surgeries . Genetic testing	. For each procedure, describe prenatal vs postnatal diagnosis . Consultations . Surgeries . Genetic testing
Autopsy findings	. Report if available	. Report if available

CHDs: tetralogy of Fallot, transposition of great arteries, and common truncus

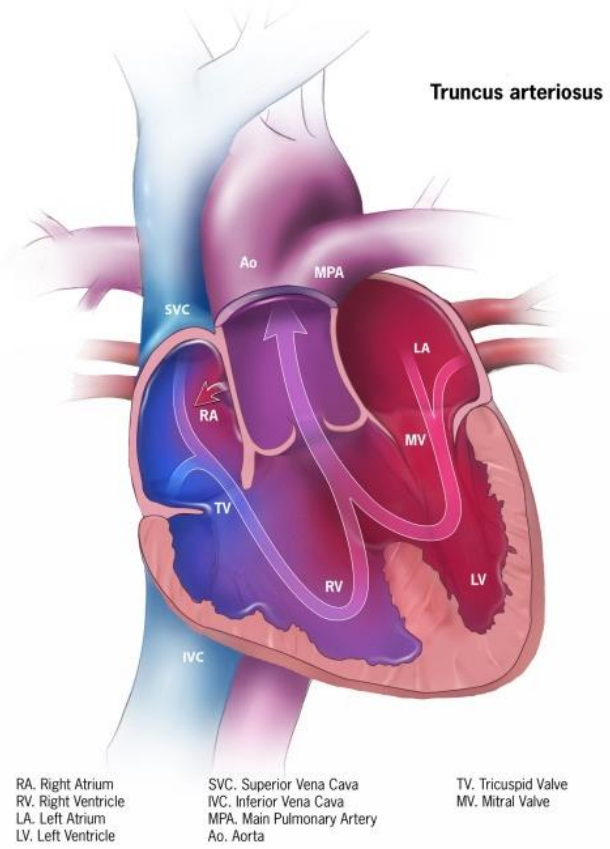




Tetralogy of Fallot

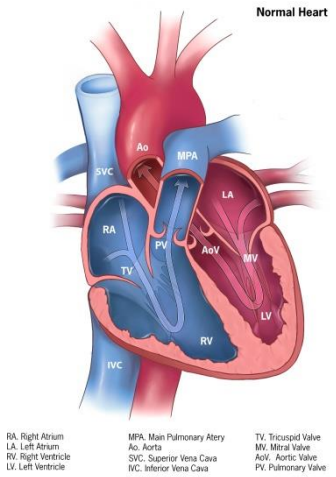


Transposition of great arteries

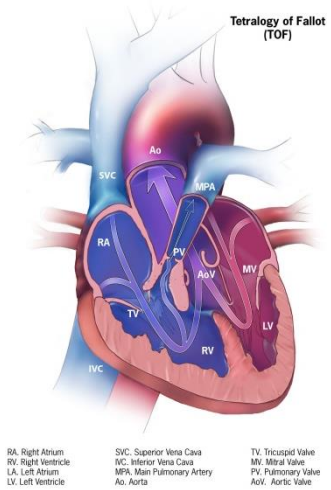


Common truncus

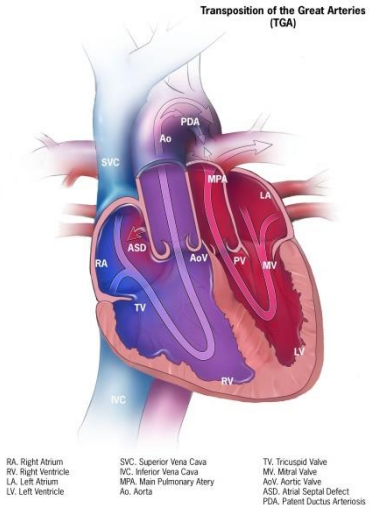
CHDs: tetralogy of Fallot, transposition of great arteries, and common truncus



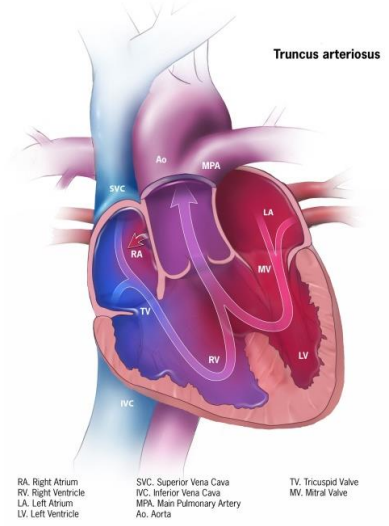
Normal heart



Tetralogy of Fallot



Transposition of great arteries



Common truncus

Tetralogy of Fallot

- Right ventricular outflow tract obstruction (with stenosis at or below the pulmonary valve, or pulmonary valve atresia or absent pulmonary valve) and ventricular septal defects (VSD)
    - Infants may not be cyanotic at first

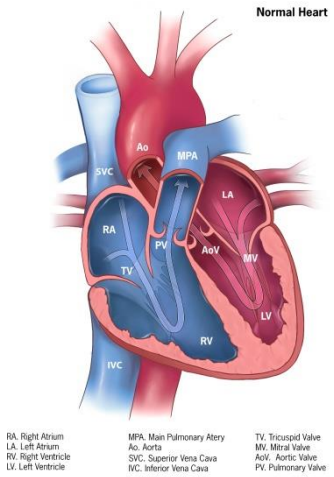
Transposition of great arteries

- Transposition of the arteries (aorta and main pulmonary artery switched)
    - Infants may not show signs of cyanosis if a ventricular septal defects (VSD), is present; if no VSD is present, infants may be cyanotic; check for heterotaxy

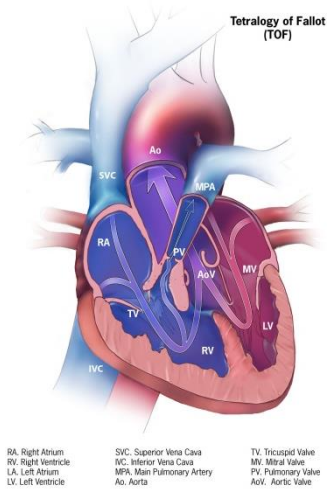
Truncus arteriosus (common truncus)

- No separate aorta or main pulmonary valve, one common truncal valve, ventricular septal defect (VSD)

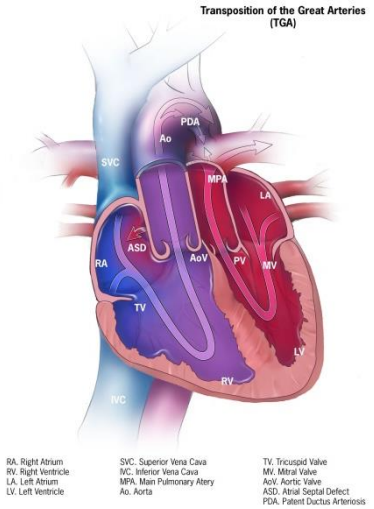
CHDs: tetralogy of Fallot and transposition of great arteries



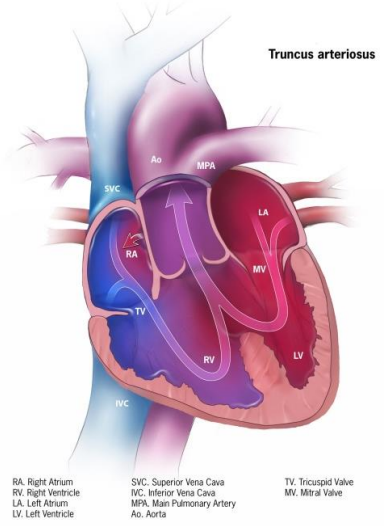
Normal heart



Tetralogy of Fallot



Transposition of great arteries



Common truncus

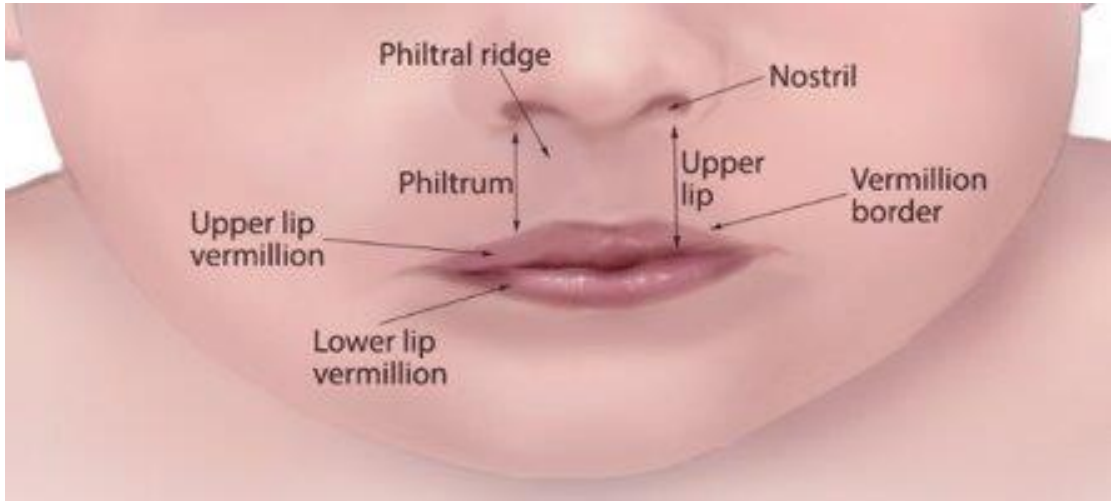
Relevant ICD-10 codes

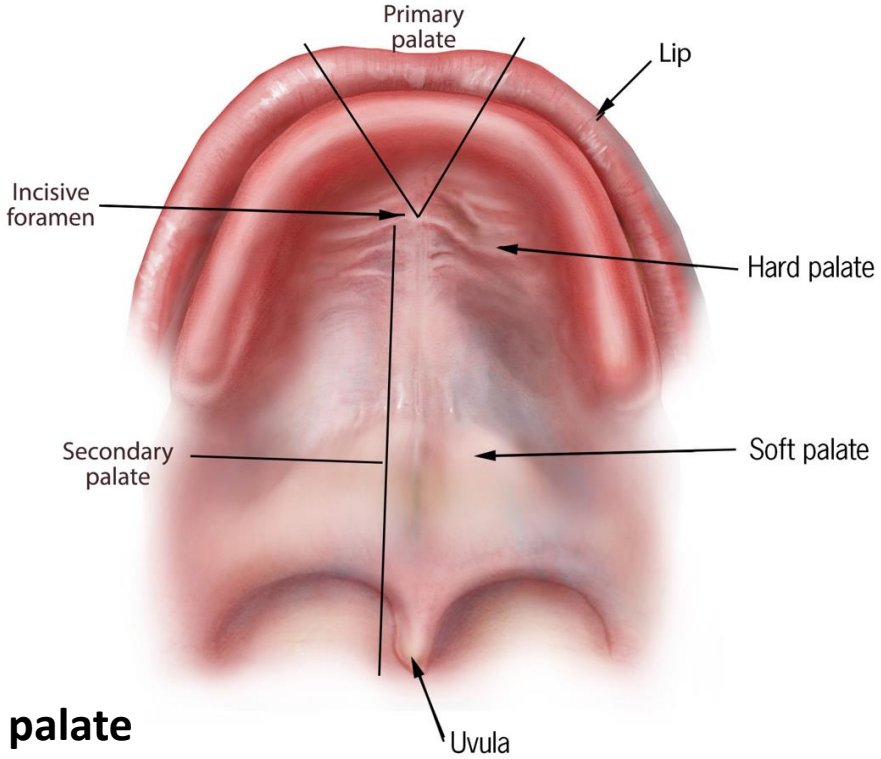
- Q21.3 Tetralogy of Fallot
- Q22.0 Pulmonary valve atresia with Q21.0 (ventricular septal defect)
- Q20.3 Transposition of great arteries
- Q20.0 Common truncus

CHDs: tetralogy of Fallot and transposition of great arteries

Description or documentation checklist for high-quality reporting			
	Transposition of great arteries	**Tetralogy of Fallot**	**Common truncus**
Describe in detail	• Intracardiac anatomy, valvar involvement, presence and type of VSD • Additional cardiac findings (e.g., ASD, atrial isomerism) • Document extracardiac anomalies • Check for evidence of heterotaxy, double outlet right ventricle (DORV)	• Intracardiac anatomy, severity of pulmonary valve involvement (stenosis or atresia); presence and type of VSD • Document extracardiac anomalies	• Anatomy – specify intracardiac anomalies, if VSD present (nearly always)/type, truncal valve morphology • Document extracardiac anomalies
Describe evaluations and procedures	• For each procedure, describe prenatal vs postnatal diagnosis • Consultations • Surgeries • Genetic testing	• For each procedure, describe prenatal vs postnatal diagnosis • Consultations • Surgeries • Genetic testing	• For each procedure, describe prenatal vs postnatal diagnosis • Consultations • Surgeries • Genetic testing
Autopsy findings	• Report if available	• Report if available	Report if available

Orofacial clefts



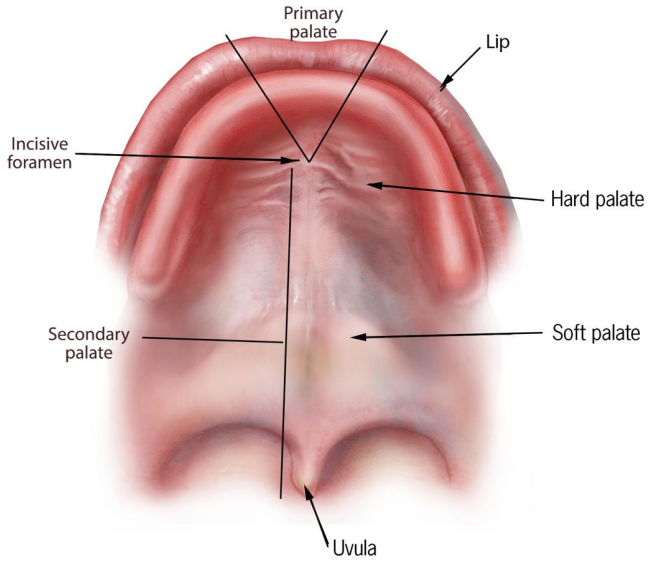


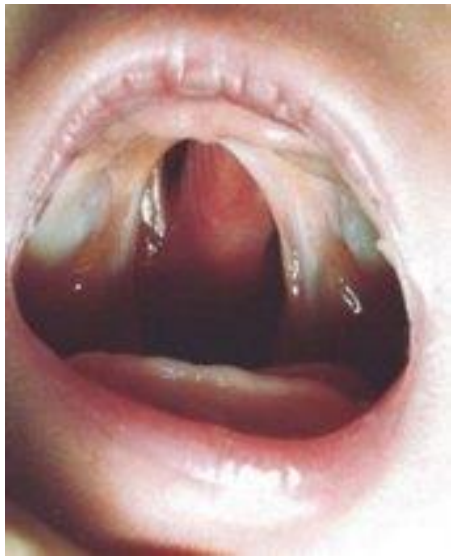
Anatomy of the lip and palate

Orofacial clefts

Photographs source: CDC-Beijing Medical University collaborative project; Dr Jaime Frias; Dr Pedro Santiago and Dr Miguel Yanez

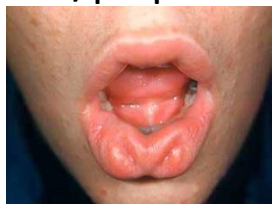
Cleft palate





Cleft palate

- Opening in the secondary palate (hard and soft palate)
- Easily missed in oral cavity and palate is not visible/palpated
- Check for lip pits – suggest a genetic condition (van der Woude syndrome)

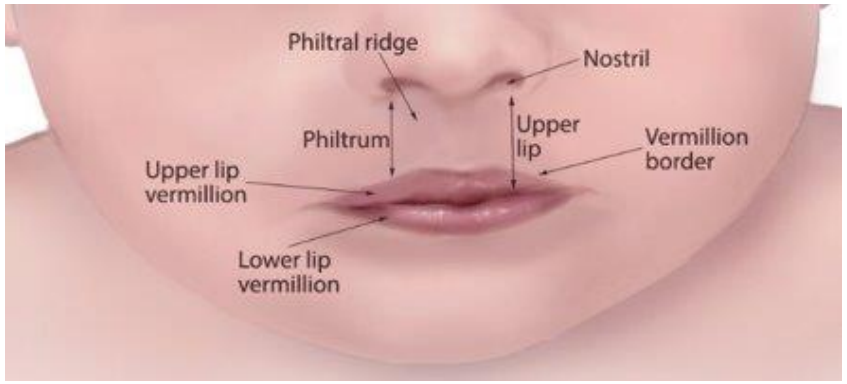


Relevant ICD-10 codes

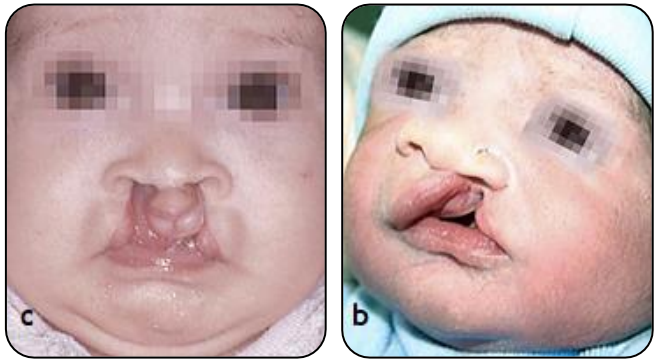
- Q35.1 Cleft hard palate
- Q35.3 Cleft soft palate
- Q35.5 Cleft hard palate with cleft soft palate
- Q35.59 Complete cleft
- Q35.9 Cleft palate, unspecified
- Q35 Cleft palate: (best to use more specific code if possible)

Photographs source: CDC-Beijing Medical University collaborative project;
https://elementsofmorphology.nih.gov/index.cgi?tid=54f2dee42a1f9ef7

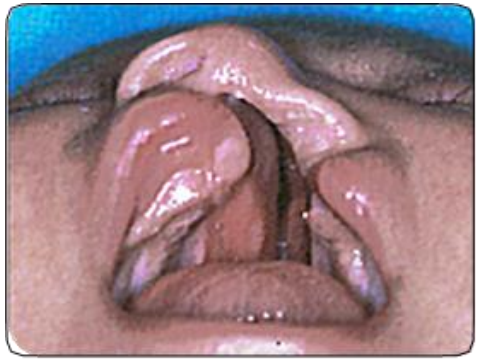
Cleft lip with or without cleft palate



Anatomy of the lip



Cleft lip



Cleft lip with cleft palate

Cleft lip

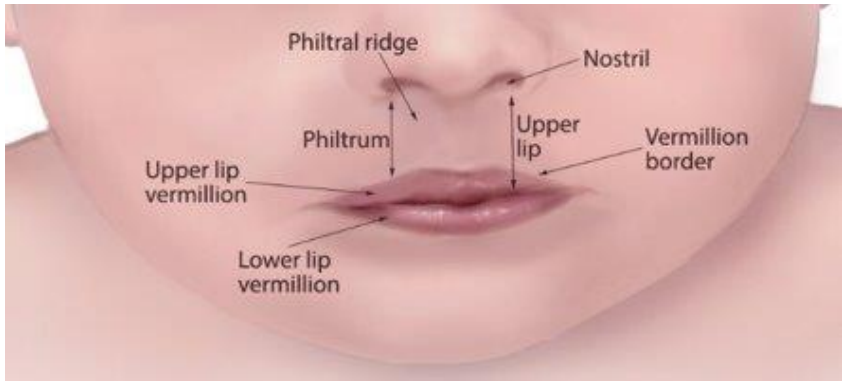
- Can be unilateral, bilateral, or median
- Partial or complete fissure
- Can extend through the gum

Cleft lip with cleft palate

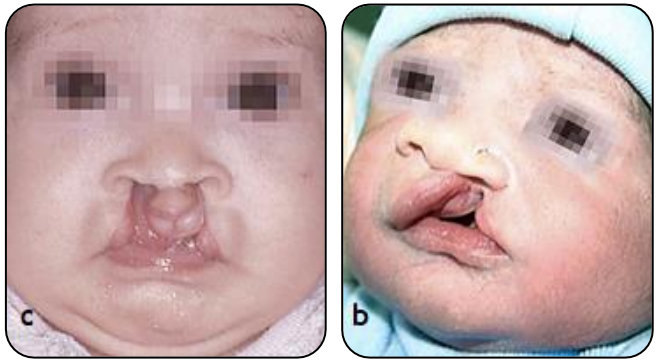
- Involves cleft of the upper lip extending into the hard palate and may extend through the soft palate

Photographs source: Dr Jaime Frias; Dr Pedro Santiago and Dr Miguel Yanez

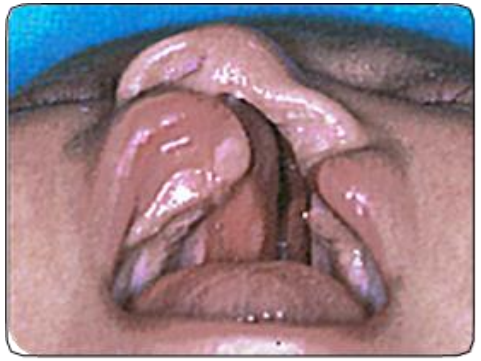
Cleft lip with or without cleft palate



Anatomy of the lip



Cleft lip



Cleft lip with cleft palate

Relevant ICD-10 codes

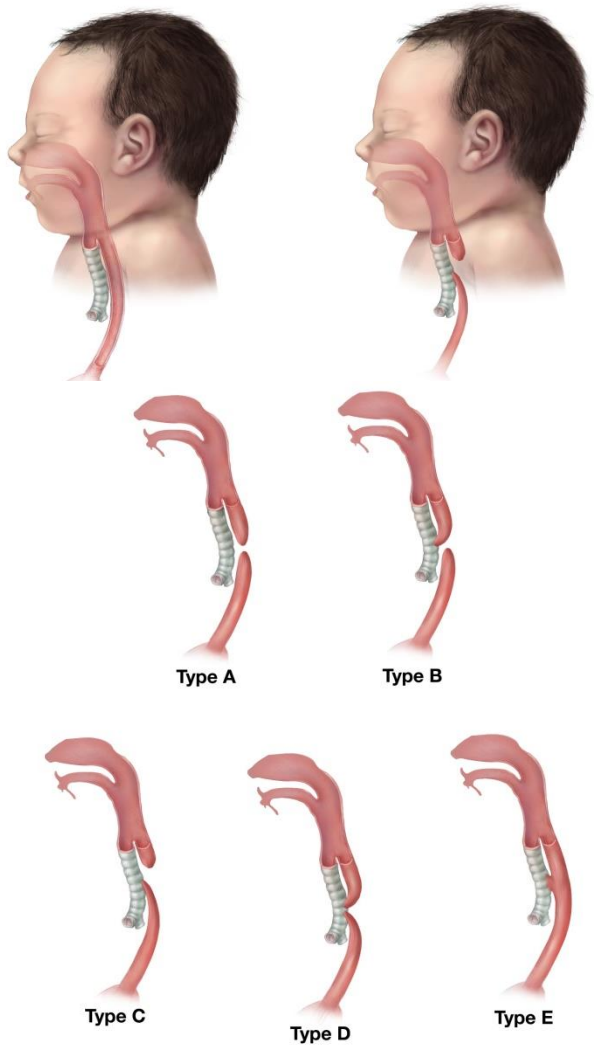
- Q36.0 Cleft lip, bilateral
- Q36.1 Cleft lip, median
- Q36.9 or Q36.90 Cleft lip, specified as unilateral
- Q36.99 Cleft lip, unspecified
- Q37.0 Cleft hard palate with bilateral cleft lip
- Q37.10 Cleft hard palate with cleft lip, specified as unilateral
- Q37.19 Cleft hard palate with cleft lip, unspecified
- 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 of the hard and soft palate with unilateral cleft lip
- Q37.59 Cleft hard palate and soft palate with cleft lip, unspecified
- Q37.8 Unspecified cleft palate with bilateral cleft lip
- Q37.9 Unspecified cleft palate with unilateral cleft lip
- Q37.99 Cleft palate with cleft lip, unspecified

Photographs source: Dr Jaime Frias; Dr Pedro Santiago and Dr Miguel Yanez

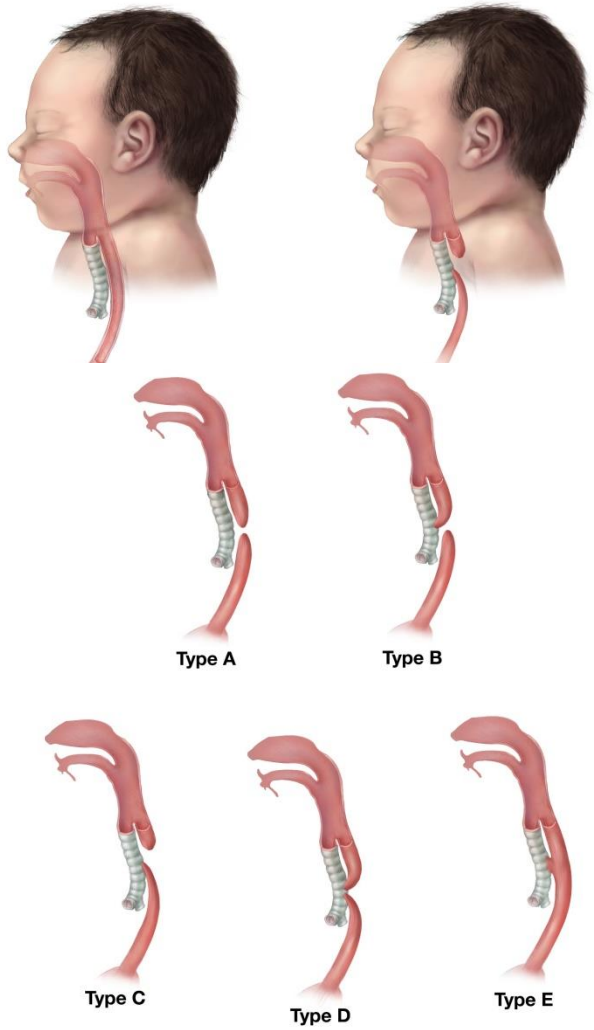
Orofacial clefts

Description or documentation checklist for high-quality reporting			
	Cleft Palate	**Cleft Lip**	**Cleft Palate and Cleft Lip**
Describe in detail	• Hard palate, soft palate • Lower lip pits present • Microretrognathia • Glossoptosis • Respiratory obstruction	• Median, lateral (unilateral [side] or bilateral) • Side of cleft • Extension of cleft (nose, gum involved) • Lower lip pits present • Check for normal palate	• Unilateral or bilateral • Side of cleft lip • Extension of cleft into palate • Lower lip pits present
Take or report photographs	Show face front	Show face front	Show face front
Describe evaluations	• Consultations • Additional unrelated anomalies, especially heart • Genetic testing	• Consultations • Additional unrelated anomalies	• Consultations • Plastic surgery • Additional unrelated anomalies • Genetic testing

Digestive system: oesophageal atresia/tracheo-oesophageal fistula



Digestive system: oesophageal atresia/tracheo-oesophageal fistula



Oesophageal atresia:

- Oesophagus ends in a blind pouch, not connected to the stomach

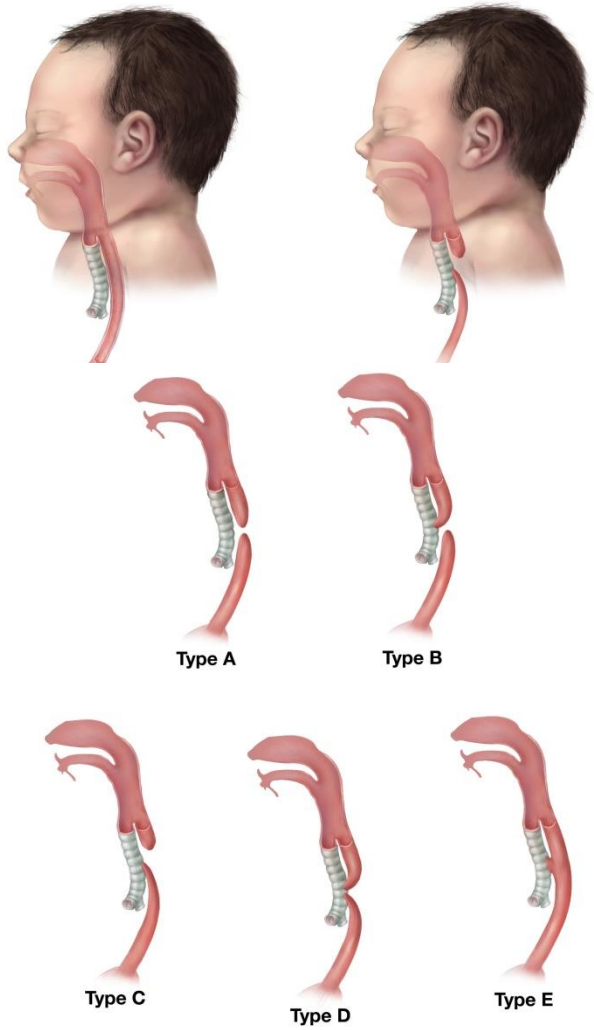
Tracheo-oesophageal fistula:

- An abnormal connection between the oesophagus and trachea

There are five types that involve the oesophagus and/or trachea:

- **A** – oesophageal atresia without tracheo-oesophageal fistula
- **B** – oesophageal atresia with proximal tracheo-oesophageal fistula
- **C** – oesophageal atresia with distal tracheo-oesophageal fistula (*most common*)
- **D** – oesophageal atresia with proximal and distal tracheo-oesophageal fistula
- **E** – tracheo-oesophageal fistula only

Digestive system: oesophageal atresia/tracheo-oesophageal fistula



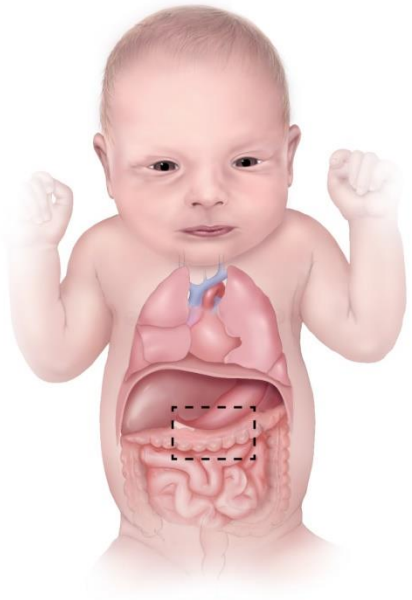
Relevant ICD-10 codes

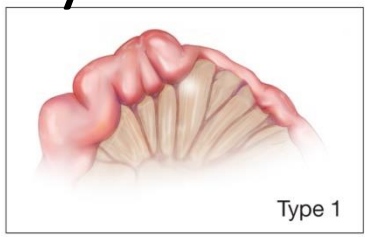
- Q39.0 Atresia of oesophagus without fistula
- Q39.1 Atresia of oesophagus with trachea-oesophageal atresia
- Q39.2 Congenital trachea-oesophageal fistula without atresia

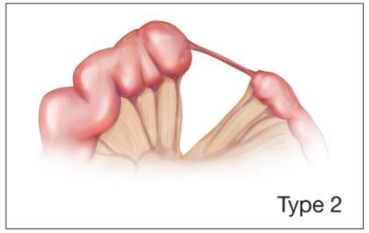
Digestive system: oesophageal atresia/tracheo-oesophageal fistula

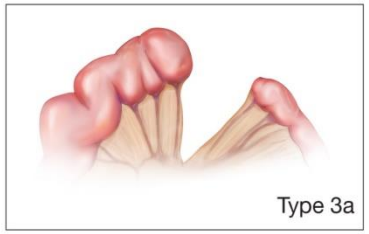
Description or documentation checklist for high-quality reporting	
	Oesophageal atresia/tracheo-oesophageal fistula
Describe in detail	• Anatomical type of oesophageal atresia and if tracheo-oesophageal fistula is present • Assess whether other defects are present (heart, anorectal/urogenital, vertebral); check for a diagnosis of OAV, VATER/VACTERL, trisomies 18 or 21, CHARGE, Feingold, other genetic syndromes
Take or report photographs	Show face front and side view and if external anomalies are present
Describe evaluations and procedures	• Specialty consultations • Surgery report • Radiographs
Testing	• Genetic tests • Chromosomal studies
Autopsy findings	Report if available

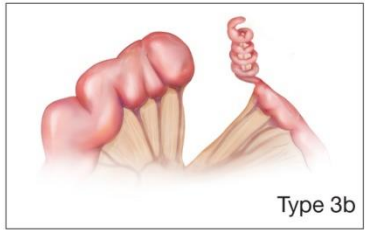
Digestive system: large intestinal atresia/stenosis

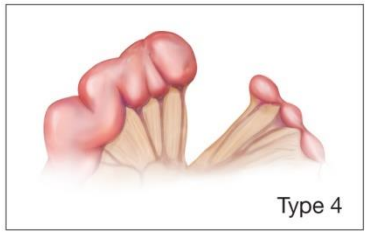




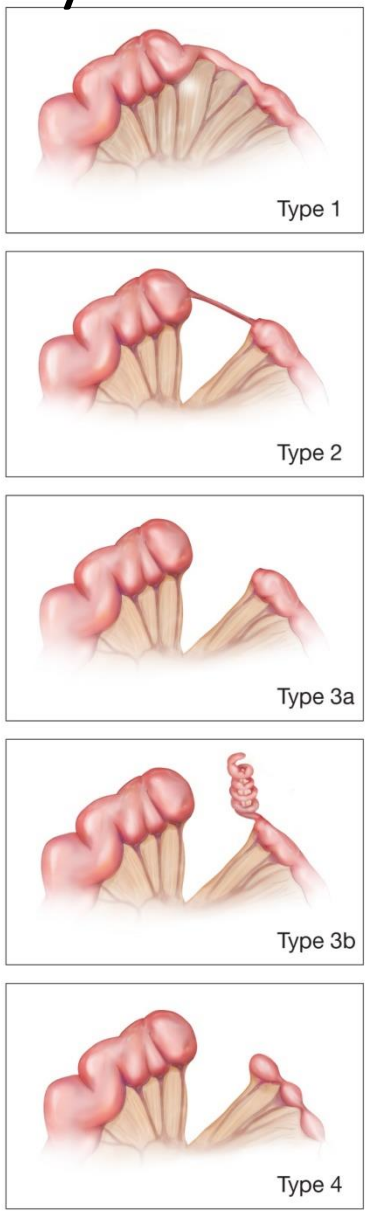








Digestive system: large intestinal atresia/stenosis



Large intestinal atresia:

- Has either complete atresia or
- Partial obstruction of the opening (lumen) within the colon (also called colonic atresia)

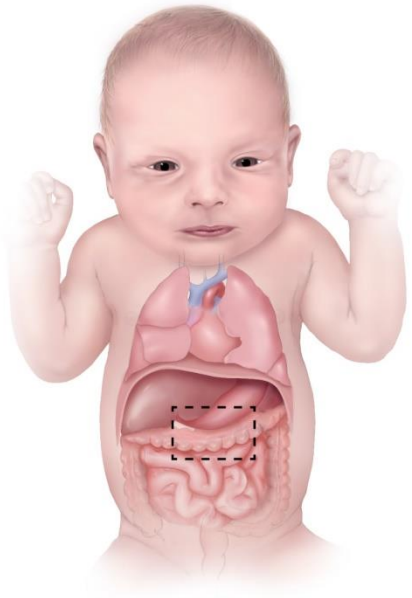
Obstruction or atresia:

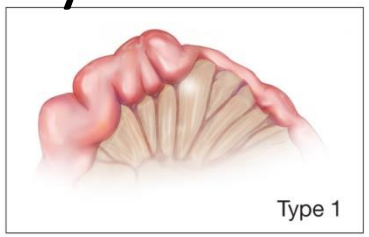
- May involve a portion of the ascending, transverse, and descending large intestine or sigmoid region

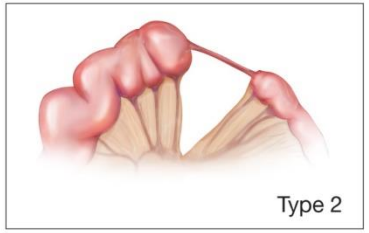
There are four types:

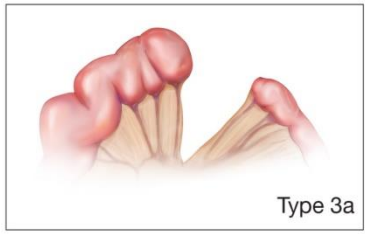
1. Lumen is blocked by tissue (mucosal web), mesentery intact
2. Atretic segment is a fibrous cord connecting the ends of the large intestine
3. Atretic segment has a V-shaped mesenteric gap defect (**3a**) or an apple peel portion (**3b**)
4. Two or more regions involved with a string of sausage type appearance

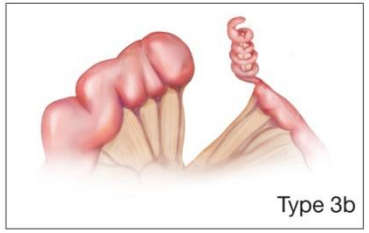
Digestive system: large intestinal atresia/stenosis

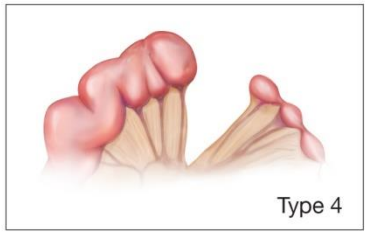












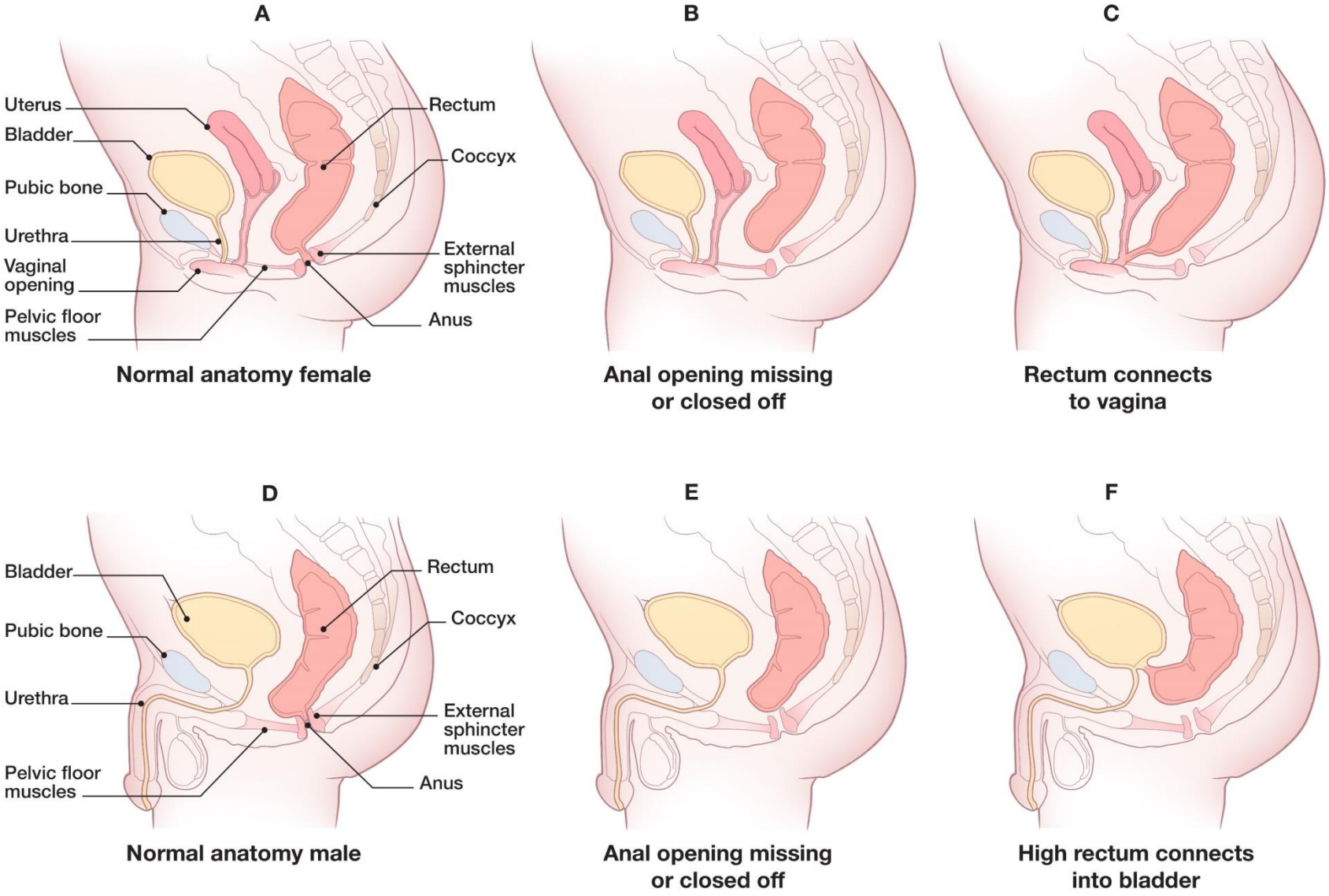
Relevant ICD-10 codes

- Q42.8 Large intestinal atresia or stenosis
- Q42.8 Sigmoid atresia or stenosis
- Q42.8 Agenesis of the recto-sigmoid portion of the colon
- Q42.9 Congenital absence, atresia and stenosis of large intestine
- Q42.90 Colonic atresia or stenosis

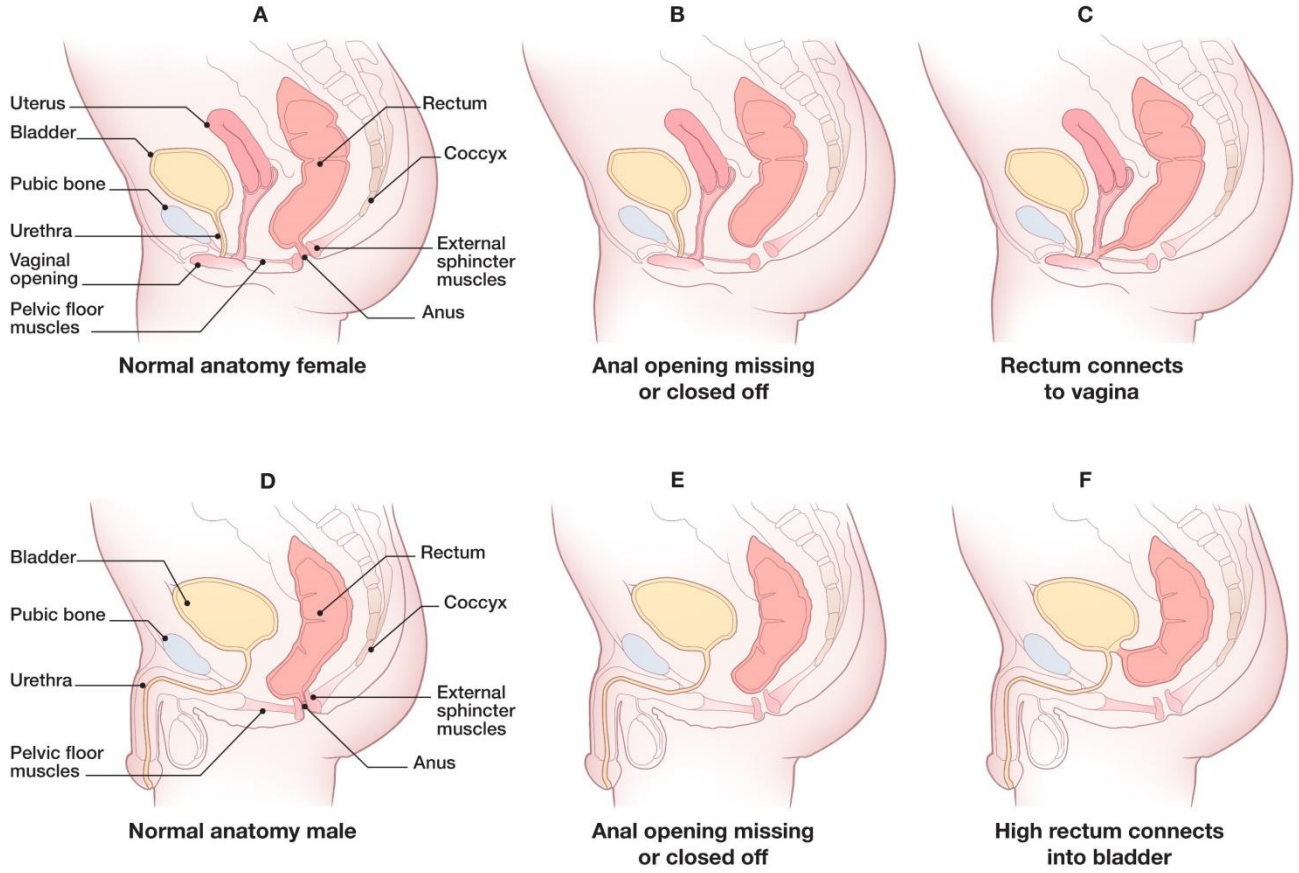
Digestive system: large intestinal atresia/stenosis

Description or documentation checklist for high-quality reporting	
	Large intestinal atresia/stenosis
Describe in detail	• Distinguish between congenital and acquired atresia (after birth) • Anatomical location/level of atresia/stenosis • Single vs. multiple atresias • Presence of fistula and location • Check for jejunal atresia • Craniofacial or ocular anomalies (seen with maternal varicella infection)
Take or report photographs	Show face front and side view and if anomalies are present
Describe evaluations	• Specialty consultations (malrotation, aganglionosis, volvulus) • Radiographs • Surgery report
Autopsy findings	Report if available

Digestive system: anal atresia/stenosis



Digestive system: anal atresia/stenosis



Anorectal atresia or stenosis:

- Involves the anus alone or an additional segment of the rectum

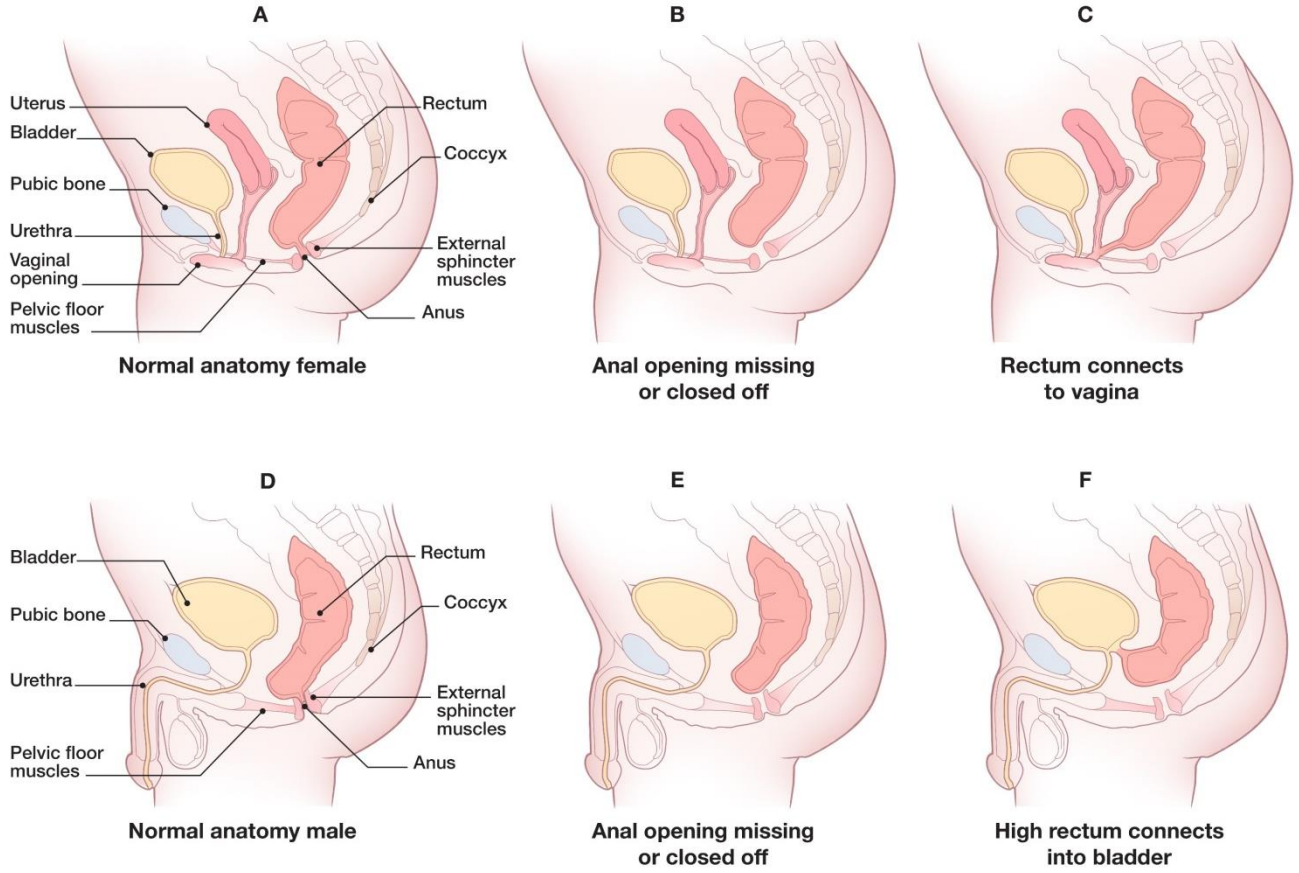
Simple classification based on descent of rectum into sphincter complex:

- Low lesion (panel E)
- High lesion (panel F)
  - High level lesions – greater frequency of associated anomalies

Rectum may connect to:

- The vagina in females (panel C)
- The bladder in males (panel F)

Digestive system: anal atresia/stenosis



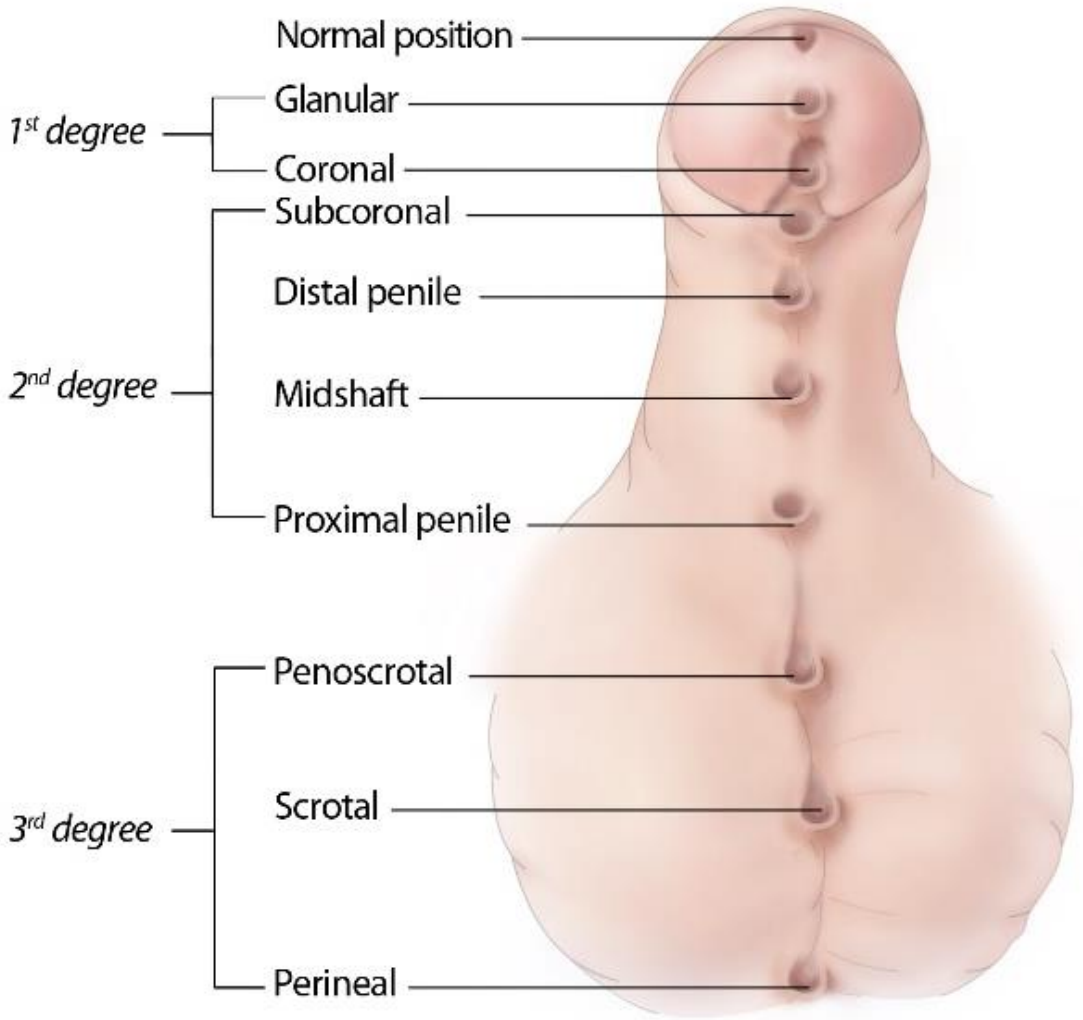
Relevant ICD-10 codes

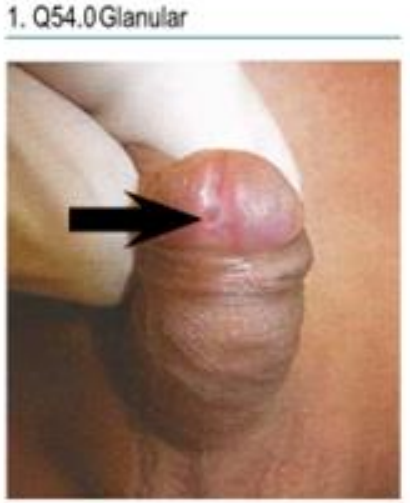
- Q42.0 Congenital absence, atresia and stenosis of rectum with fistula
- Q42.1 Congenital absence, atresia and stenosis of rectum without fistula
- Q42.2 Congenital absence, atresia and stenosis of anus with fistula
- Q42.3 Congenital absence, atresia and stenosis of anus without fistula
- Q42.3 Imperforate anus

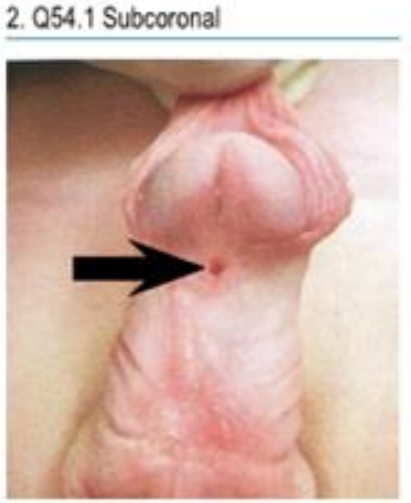
Digestive system: anal atresia/stenosis

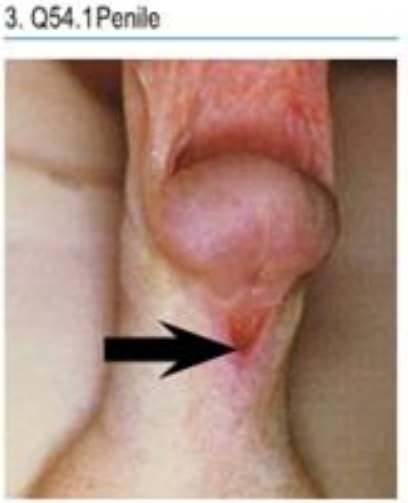
Description or documentation checklist for high-quality reporting	
	Anorectal atresia
Describe in detail	• Level of lesion (high or low) • Atresia (anus or rectum), fistula type if present • Additional anomalies present – urinary tract, caudal regression, components of the OEIS complex (Omphalocele, cloacal Exstrophy, Imperforate anus, Spinal anomalies), components of the VATER/VACTERL association
Take or report photographs	• Anal area • Additional anomalies if present
Describe evaluations	• Specialty consultations and procedures to assess additional anomalies • Radiographs • Surgery report • Genetic testing • Chromosomal studies
Autopsy findings	Report if available

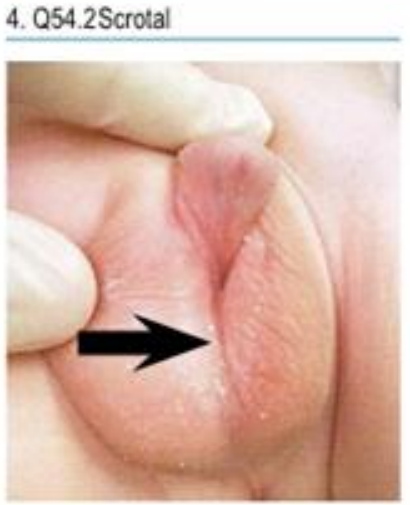
Genital and urinary system: hypospadias

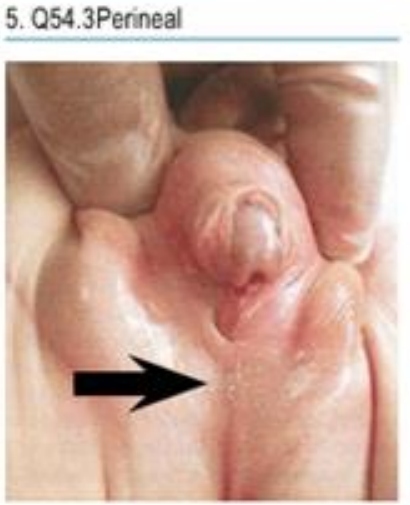






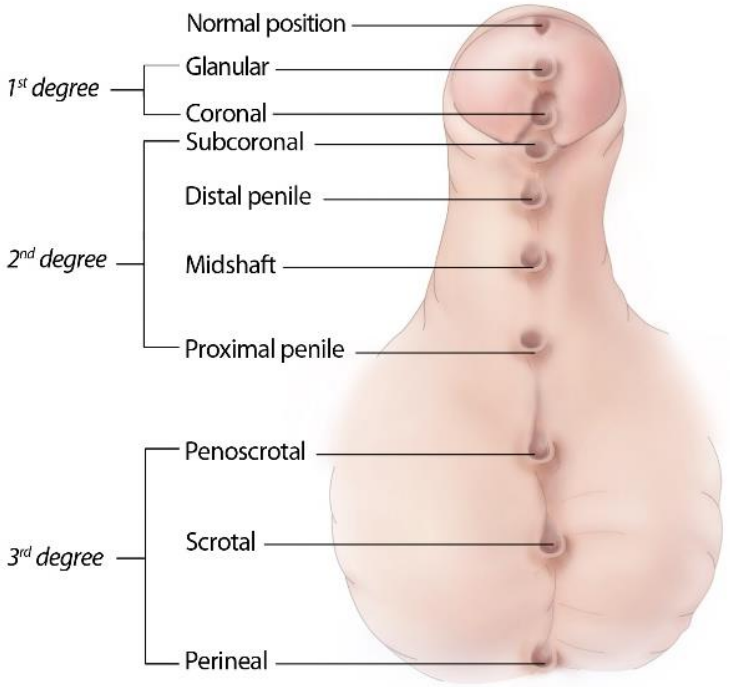


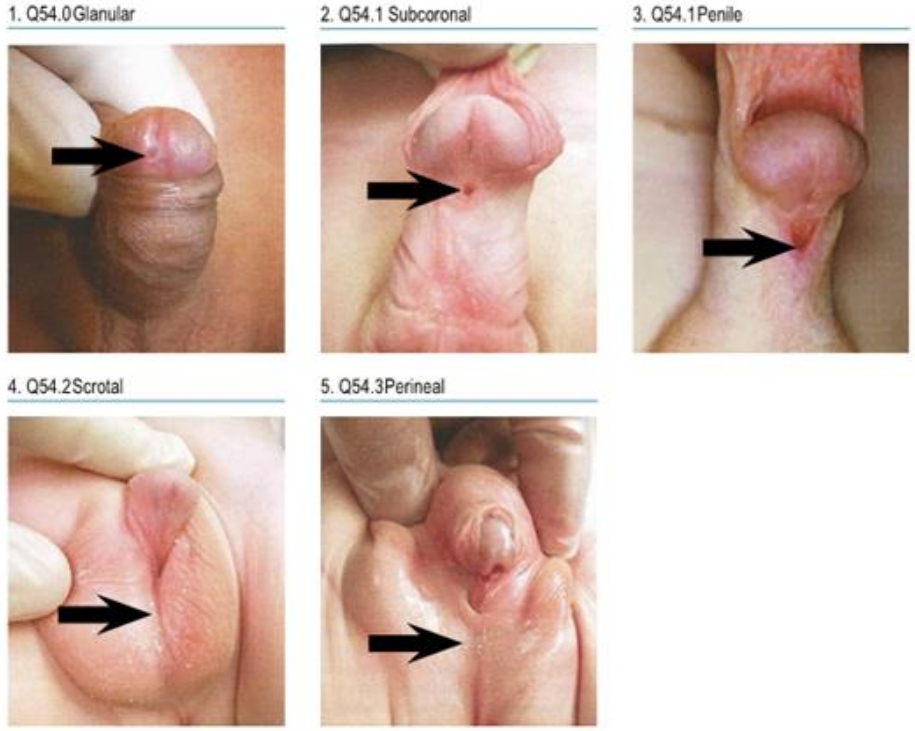




Photographs source: Kliegman RM, St. Geme III JW, editors. Nelson textbook of pediatrics, 2-volume set. Philadelphia, PA: Elsevier; 2024.

Genital and urinary system: hypospadias

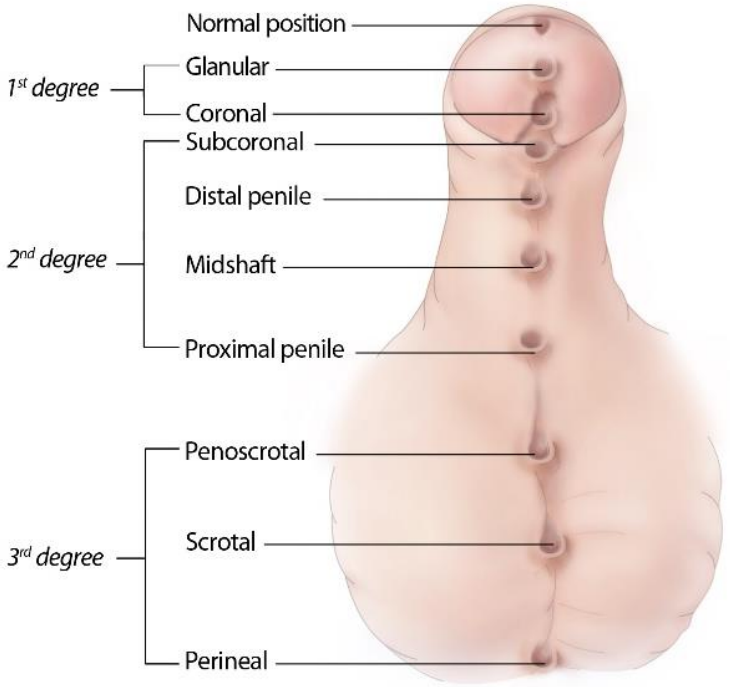


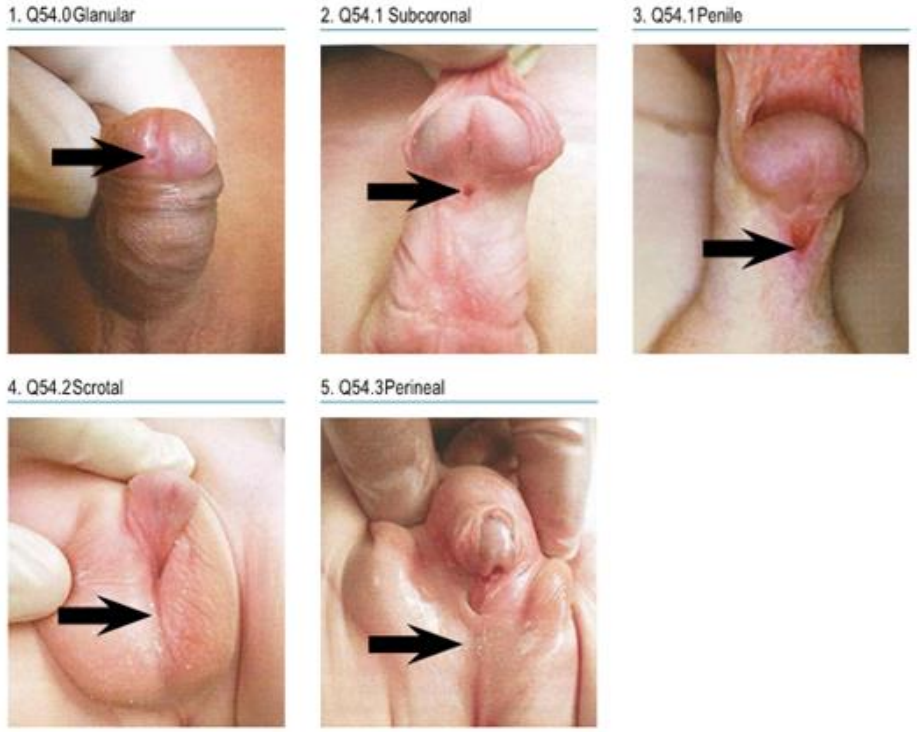


- Normal position for the urethral meatus is on the tip of the penis
- Severity of the hypospadias increases as the location of the urethral meatus becomes more proximal
- Shortening of the ventral side of the penis can result in a curvature of the penis (chordee)

Photographs source: Kliegman RM, St. Geme III JW, editors. Nelson textbook of pediatrics, 2-volume set. Philadelphia, PA: Elsevier; 2024.

Genital and urinary system: hypospadias





Relevant ICD-10 codes

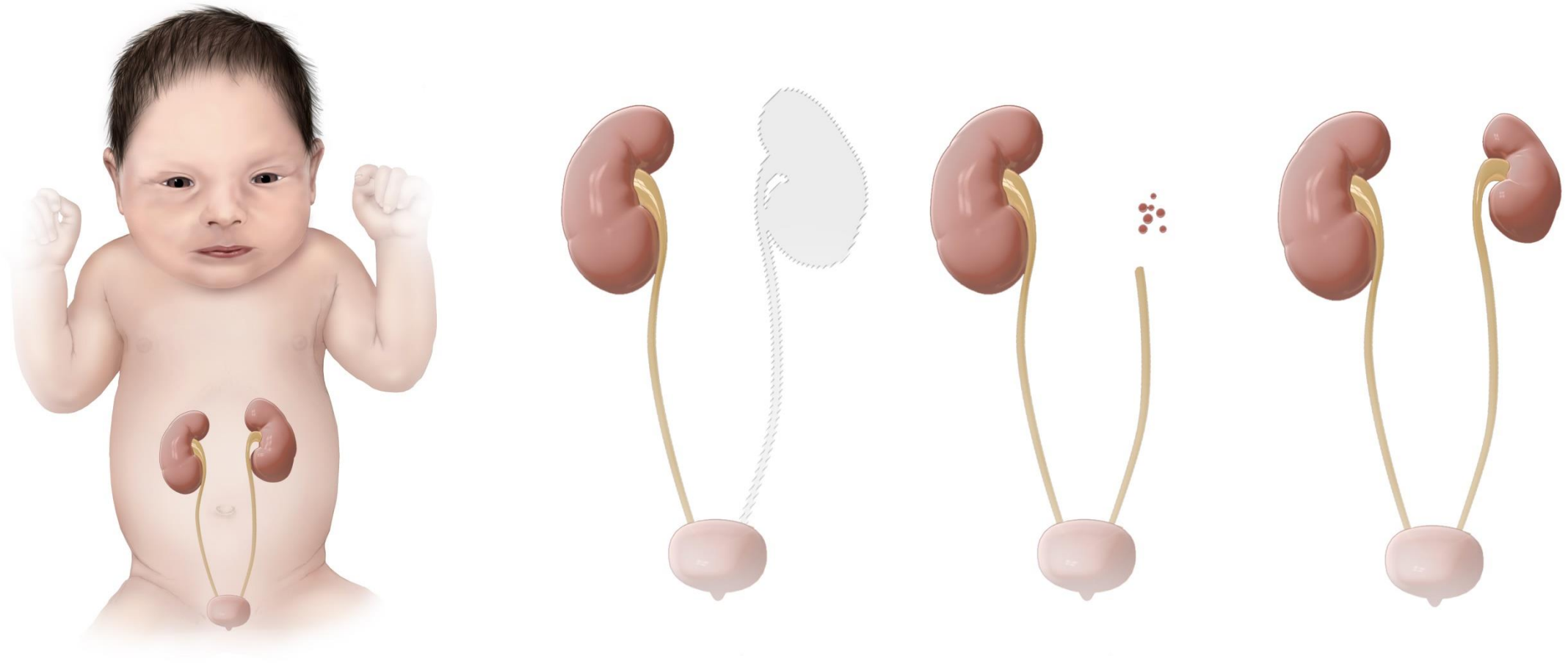
- Q54.0 Hypospadias, balanic (coronal, glanular)
- Q54.1 Hypospadias, penile (includes subcoronal and midshaft)
- Q54.2 Hypospadias, penoscrotal
- Q54.3 Hypospadias, perineal
- Q54.8 Hypospadias, other
- Q54.9 Hypospadias, unspecified

Photographs source: Kliegman RM, St. Geme III JW, editors. Nelson textbook of pediatrics, 2-volume set. Philadelphia, PA: Elsevier; 2024.

Genital and urinary system: hypospadias

Description or documentation checklist for high-quality reporting	
	Hypospadias
Describe in detail	• Placement of the urethral meatus (glanular, coronal, subcoronal, shaft, scrotal, perineal) • Presence of chordee (curvature) and testes (undescended – unilateral, bilateral) • Other anomalies of the urinary tract or genitalia
Take or report photographs	• Male genitalia • If additional anomalies are present
Describe evaluations	• Surgery report • Specialty consultations • Genetic tests • Chromosomal studies • Biochemical tests
Autopsy findings	Report if available

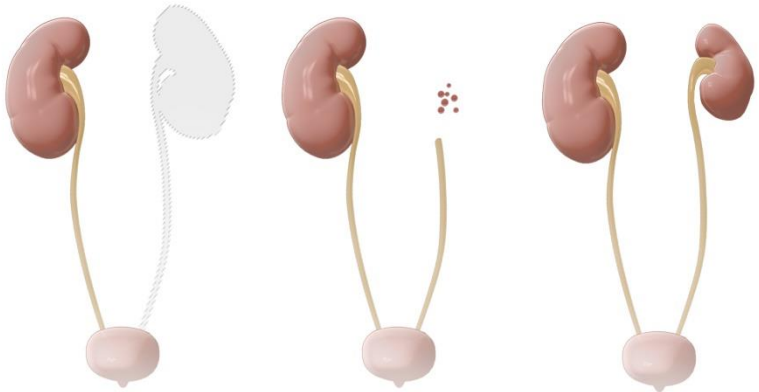
Genital and urinary system: renal agenesis and hypoplasia



Normal kidneys **Agenesis** **Aplasia** **Hypoplasia**

Genital and urinary system: renal agenesis and hypoplasia

Normal kidneys



Agenesis **Aplasia** **Hypoplasia**

Renal agenesis:

- Can be bilateral or unilateral
- Bilateral renal agenesis is a lethal condition and infants are either stillborn or die shortly after birth

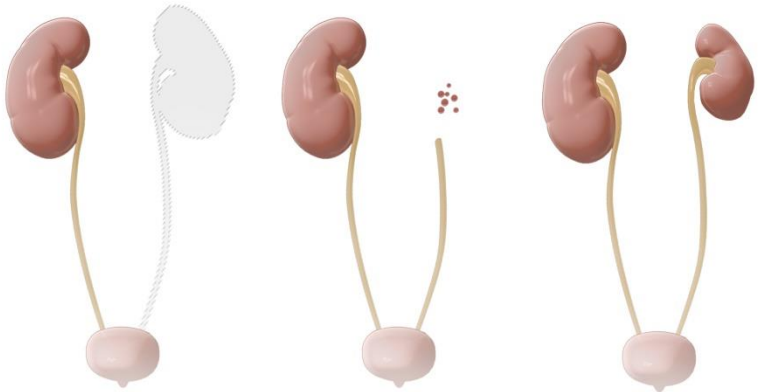
Unilateral cases may be:

- Complete agenesis (no kidney or ureter)
- Aplasia (ureter present, no kidney)
- Hypoplasia (small kidney)

Agenesis and/or hypoplasia (unilateral renal agenesis with contralateral renal hypoplasia) may occur

Genital and urinary system: renal agenesis and hypoplasia

Normal kidneys



Agenesis Aplasia Hypoplasia

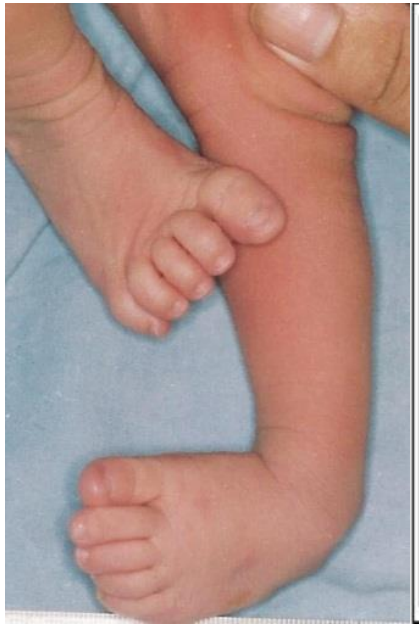
Relevant ICD-10 codes

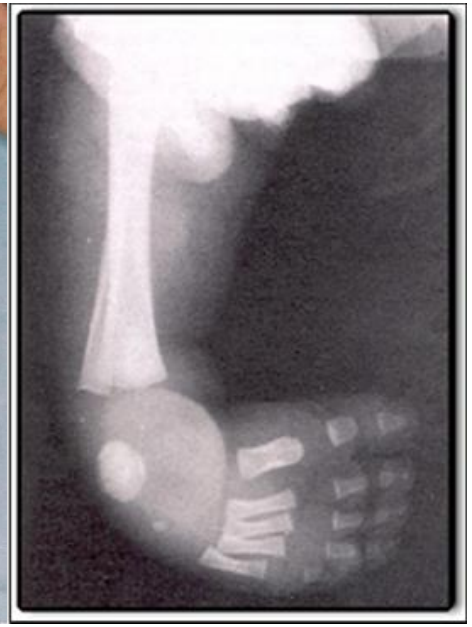
- Q60.0 Unilateral renal agenesis
- Q60.1 Bilateral renal agenesis
- Q60.2 Unspecified renal agenesis
- Q60.3 Renal hypoplasia, unilateral
- Q60.4 Renal hypoplasia, bilateral
- Q60.5 Renal hypoplasia, unspecified
- Q60.6 Potter sequence with renal agenesis

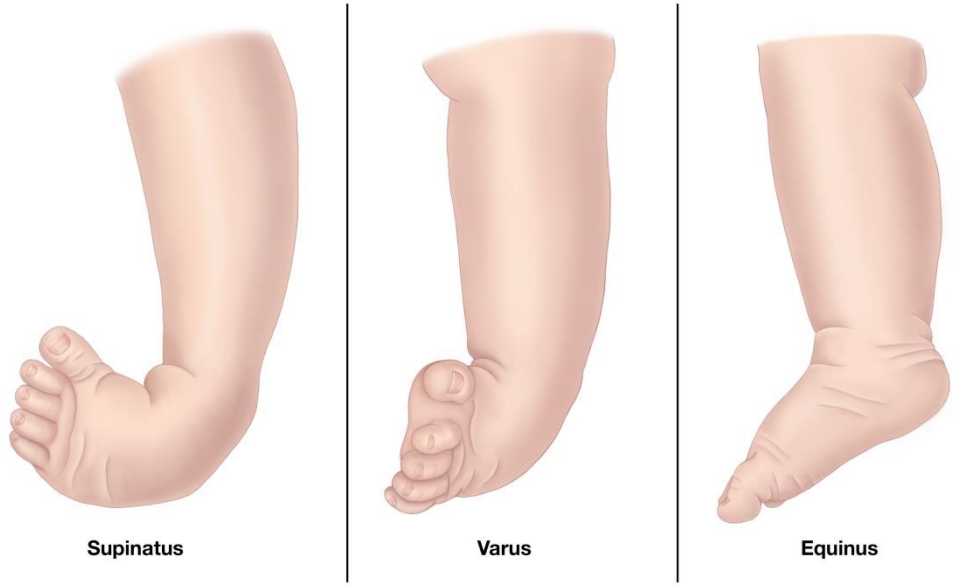
Genital and urinary system: renal agenesis and hypoplasia

Description or documentation checklist for high-quality reporting	
	Renal agenesis and/or hypoplasia
Describe in detail	• Distinguish unilateral or bilateral, right or left • Agenesis and/or hypoplasia (unilateral renal agenesis with contralateral renal hypoplasia) • Other anomalies of the urinary tract, face, or unrelated anomalies (VATER/VACTERL)
Take or report photographs	Face (Potter sequence), talipes, contractures
Describe evaluations	• Specialty consultations • Surgery, procedure, and radiology reports • Genetic tests • Chromosomal studies
Autopsy findings	Report if available

Musculoskeletal system: talipes equinovarus

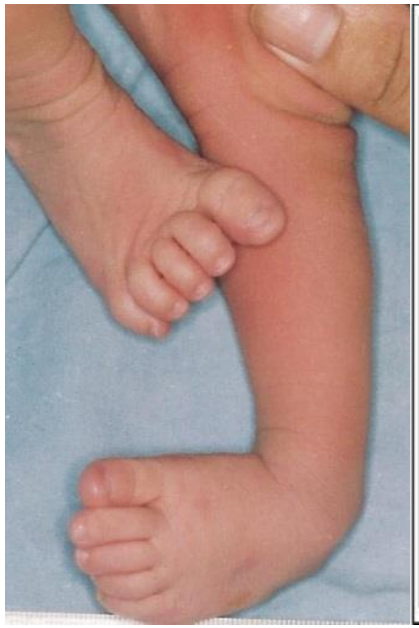


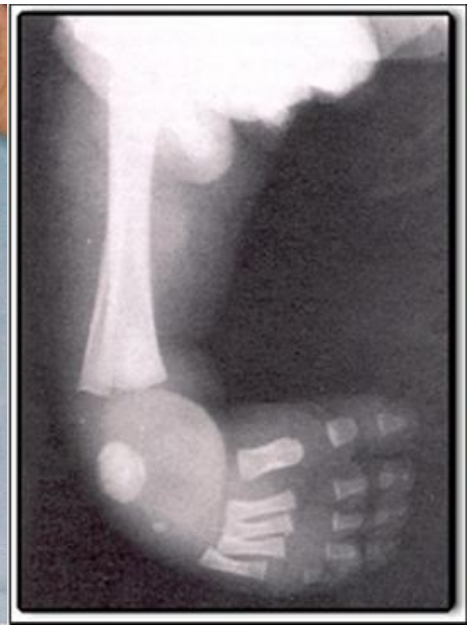


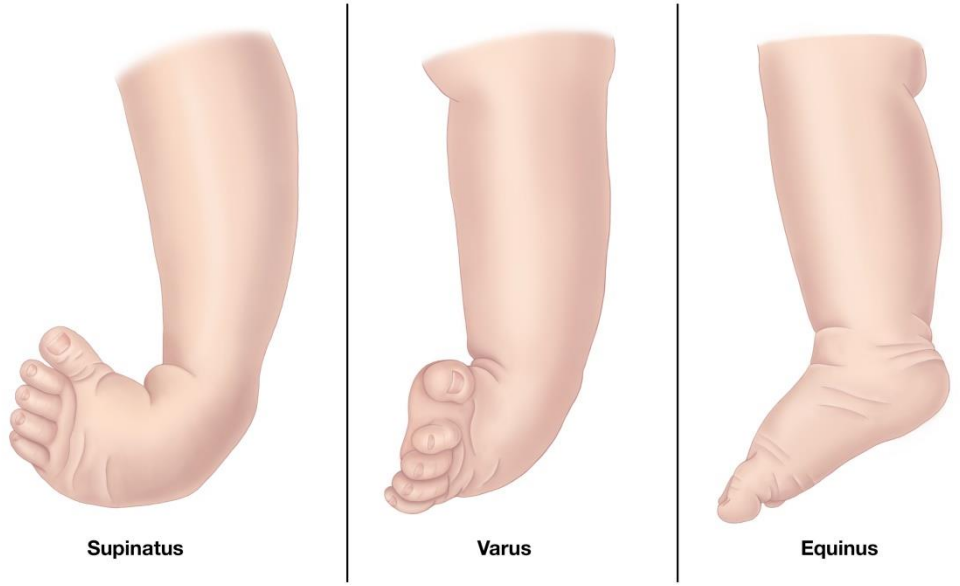


Photograph and x-ray source: Dr Idalina Montes and Dr Rajael Longo

Musculoskeletal system: talipes equinovarus





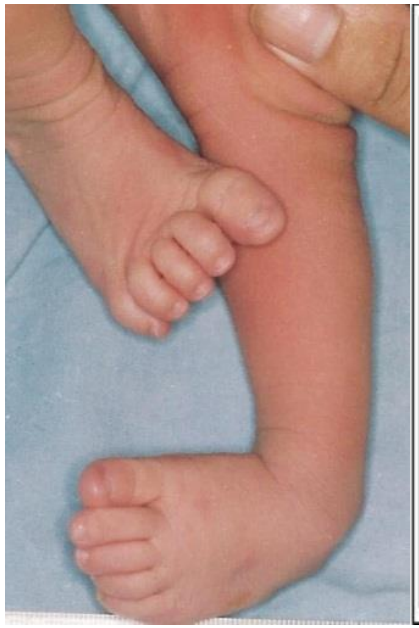


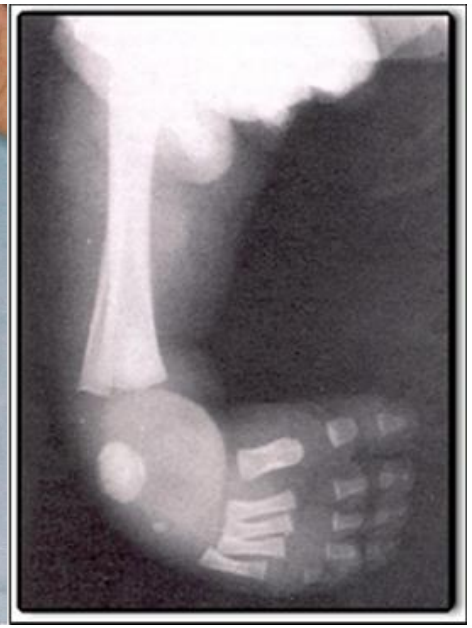
Talipes equinovarus:

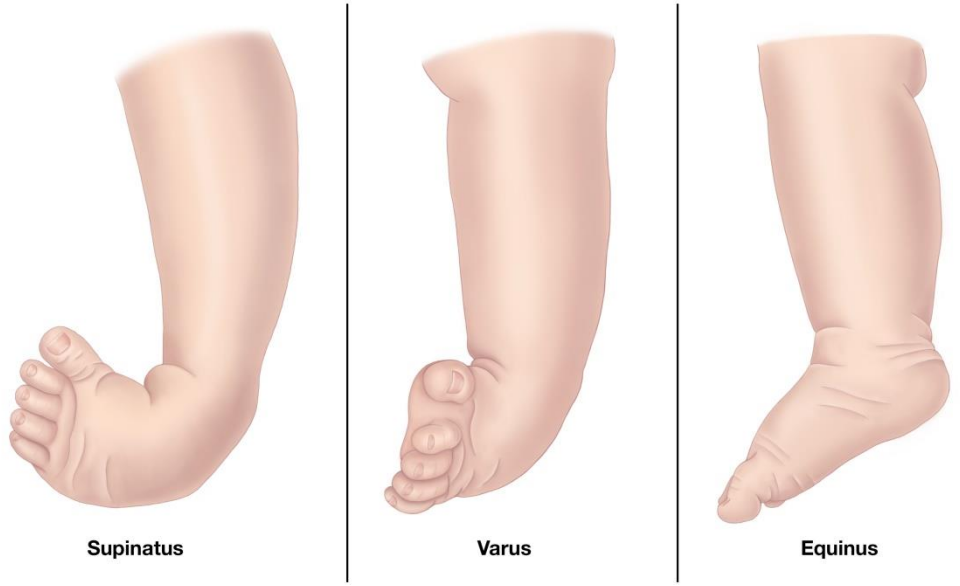
- May be bilateral or unilateral
- The foot can be rigid (cannot be manipulated) or positional (can be manipulated into normal position)
- The position of the foot can vary and includes supinatus, varus and equinovarus

Photograph and x-ray source: Dr Idalina Montes and Dr Rajael Longo

Musculoskeletal system: talipes equinovarus







Relevant ICD-10 codes

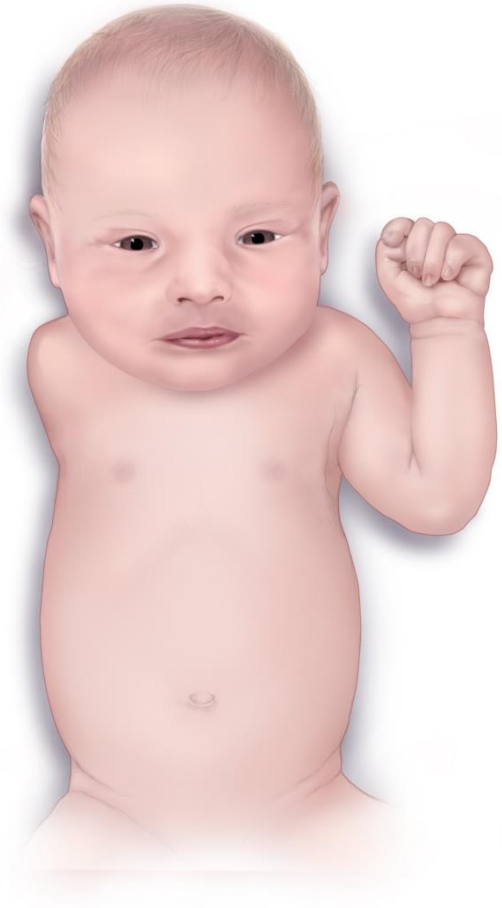
- Q66.0 Talipes equinovarus
- Q66.8 Other congenital deformities of feet, clubfoot NOS
- Q66.1 Talipes calcaneovarus
- Q66.4 Talipes calcaneovalgus

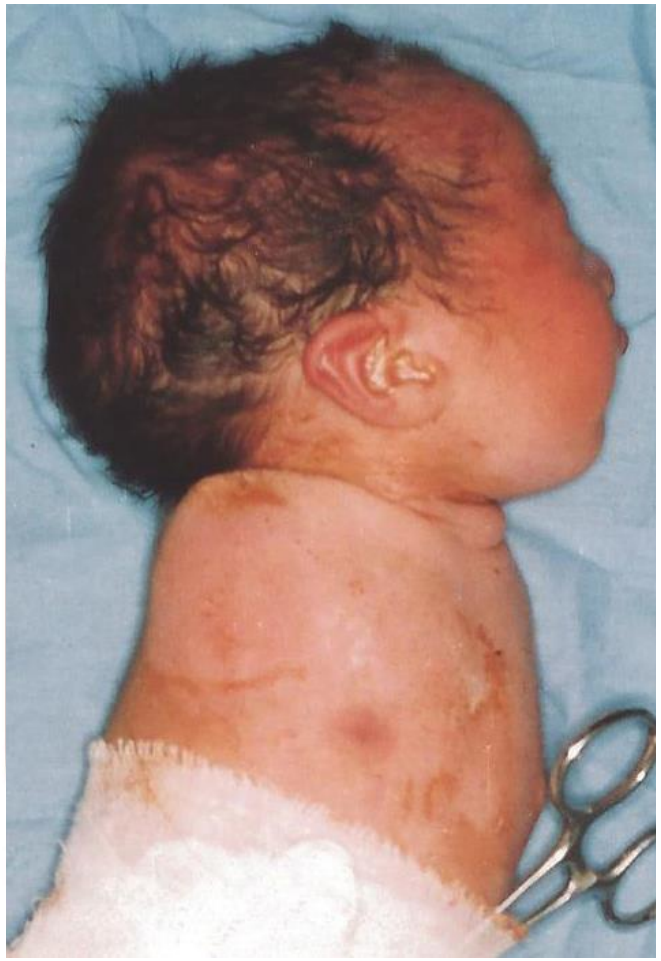
Photograph and x-ray source: Dr Idalina Montes and Dr Rajael Longo

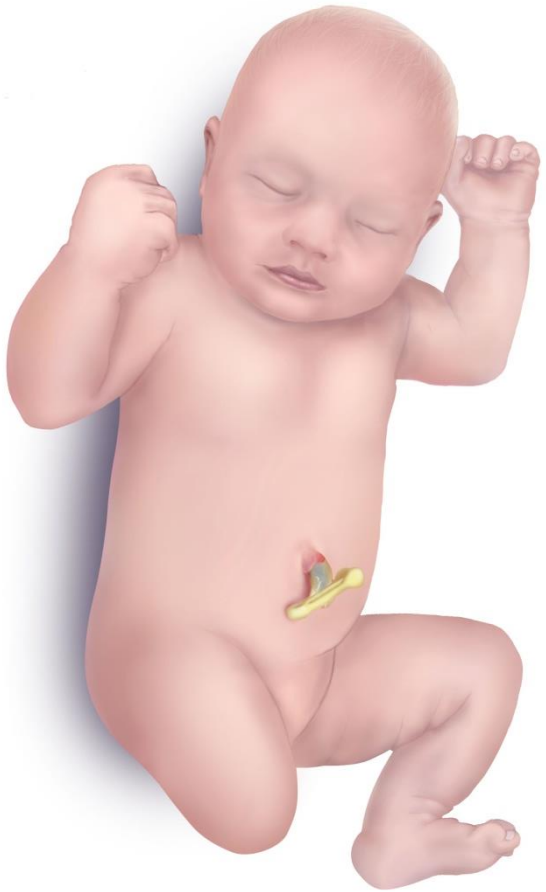
Musculoskeletal system: talipes equinovarus

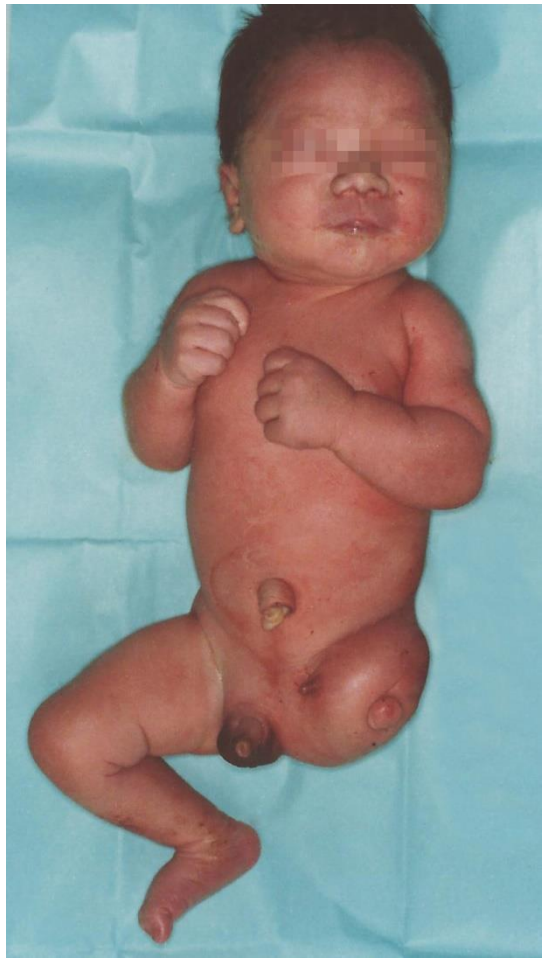
Description or documentation checklist for high-quality reporting	
	Talipes equinovarus
Describe in detail	• Mobility of the foot – rigid vs flexible • Bilateral or unilateral, right or left • Position (plantar flexion, varus deformity) • Other anomalies of the musculoskeletal system (neck, hip, joints), brain or spine
Take or report photographs	• Entire infant • Both feet
Describe evaluations	• Specialty consultations (e.g., genetics, orthopedics) • Surgery, procedure and radiology reports • Genetic tests • Chromosomal studies
Autopsy findings	Report if available

Limb deficiency: amelia



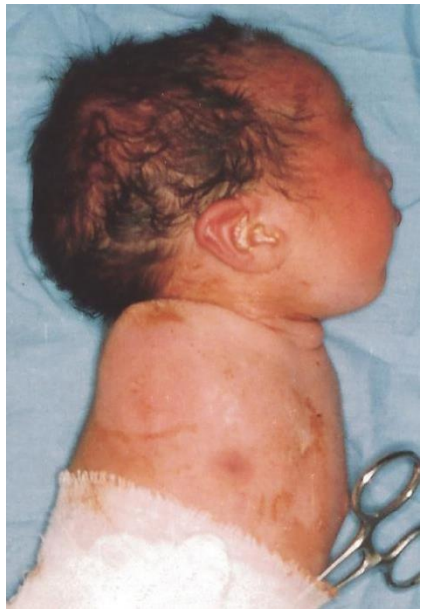
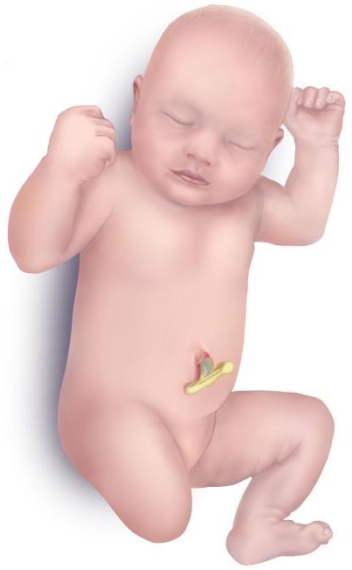
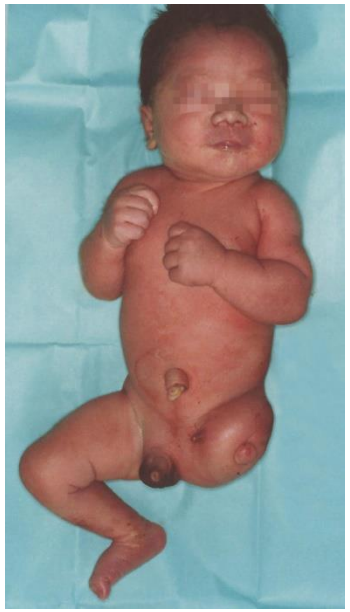






Photograph source: CDC-Beijing Medical University collaborative project

Limb deficiency: amelia

Amelia

- The complete absence of one or more limbs
- May occur in upper limbs and/or lower limbs and may be bilateral or unilateral

Relevant ICD-10 codes

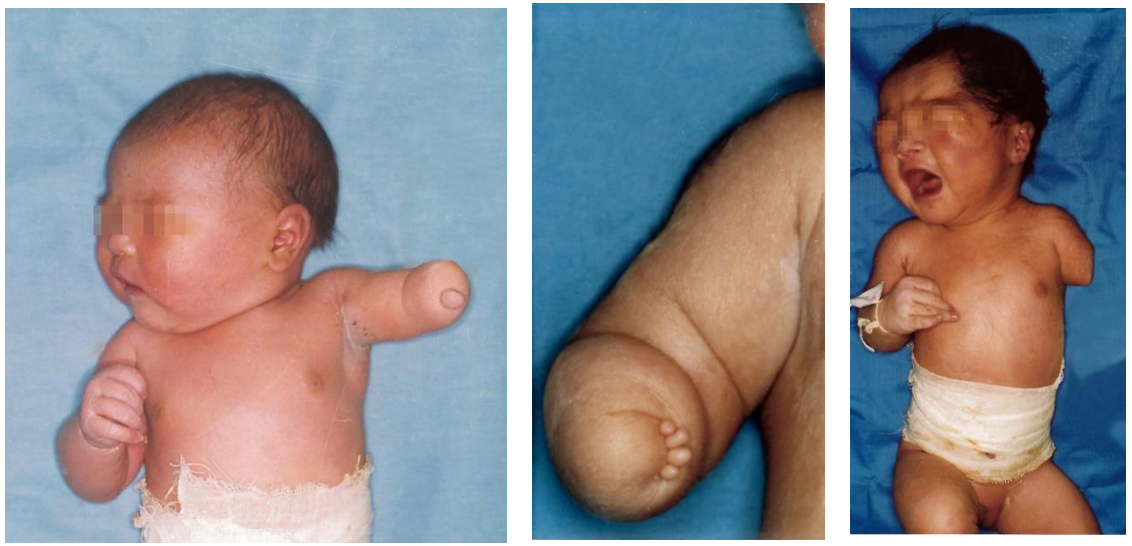
- Q71.0 Congenital complete absence of upper limb(s); amelia of upper limb
- Q72.0 Congenital complete absence of lower limb(s); amelia of lower limb
- Q73.0 Congenital complete absence of unspecified limb(s); amelia NOS

Photograph source: CDC-Beijing Medical University collaborative project

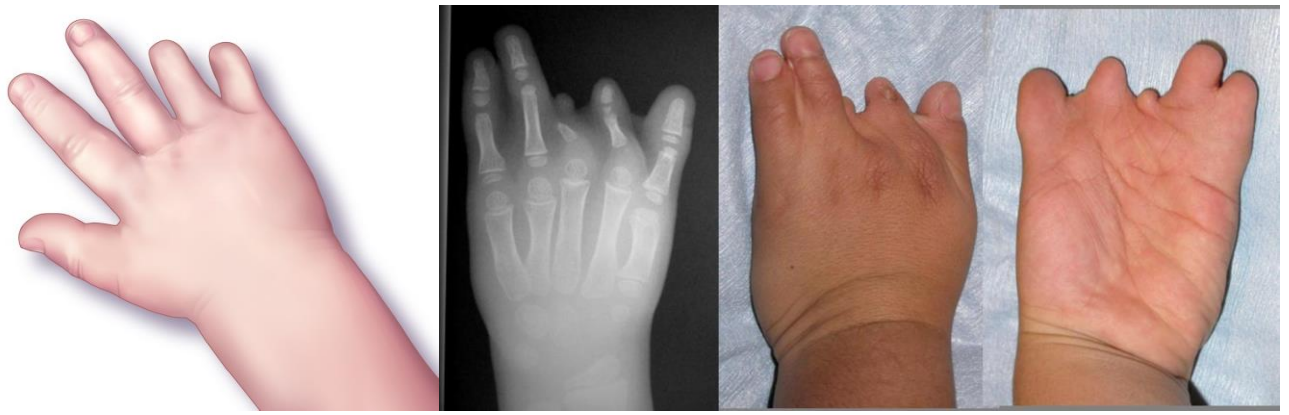
Limb deficiency: amelia

Description or documentation checklist for high-quality reporting	
	Amelia
Describe in detail	• Limb involvement (upper, lower, both) • Bilateral, unilateral (right or left) • Involved segment (complete or partial) • Other unrelated anomalies if present
Take or report photographs	• Entire infant • All extremities
Describe evaluations	• Specialty consultations (e.g., genetics, orthopedics) • Procedure and radiology reports • Genetic tests • Chromosomal studies
Autopsy findings	Report if available

Limb deficiency: transverse terminal



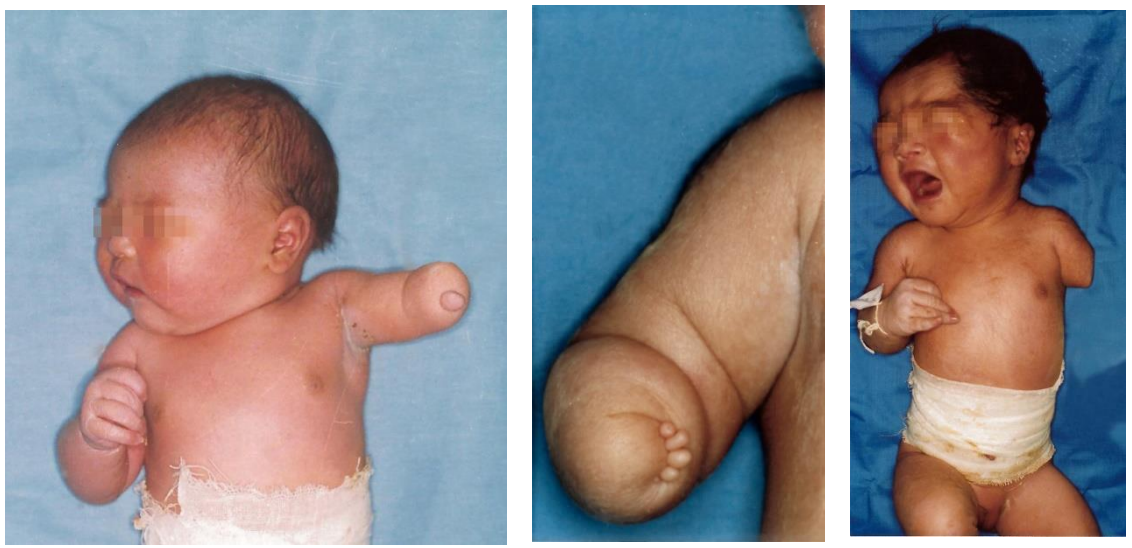
Transverse terminal – congenital absence of both forearm and hand



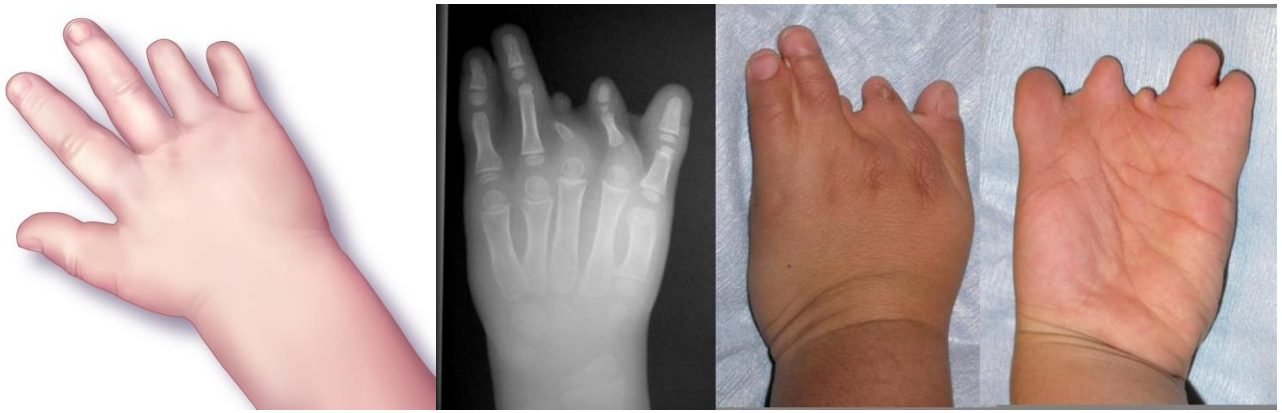
Transverse terminal – congenital absence of finger(s)

Photographs and imaging sources: CDC-Beijing Medical University collaborative project; Dr. E Gene Deune

Limb deficiency: transverse terminal



Transverse terminal – congenital absence of both forearm and hand



Transverse terminal – congenital absence of finger(s)

Transverse Terminal:

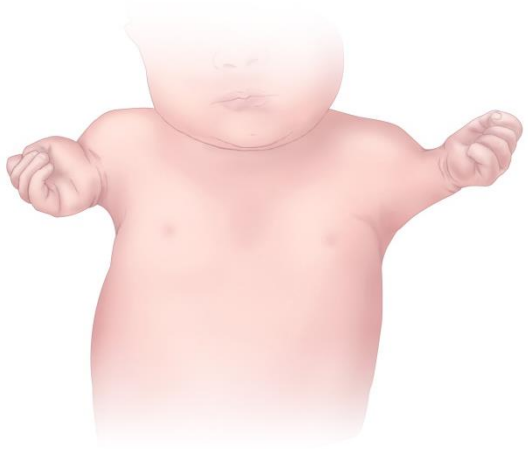
- The affected limb appears amputated (arm, leg, toes, or fingers)
- The affected limb/digit is missing the distal segments and the proximal segments remain intact

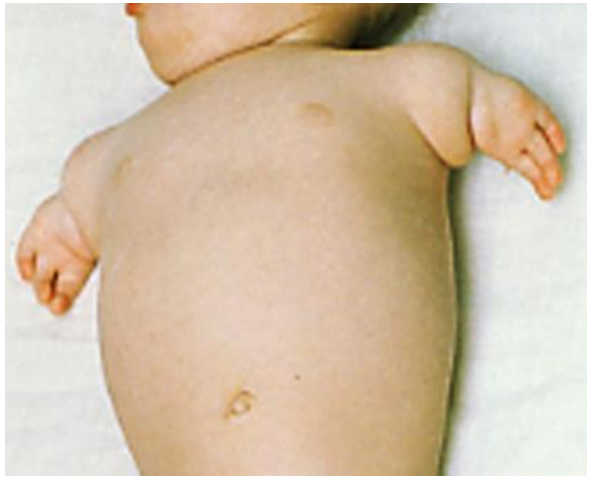
Relevant ICD-10 codes

- Q71.2 Congenital absence of both forearm and hand
- Q71.3 Congenital absence of hand and finger(s)
- Q71.30 Congenital absence of finger(s)
- Q72.2 Congenital absence of both lower leg and foot
- Q72.3 Congenital absence of foot and toe(s)
- Q72.30 Congenital absence or hypoplasia of toe(s) with remainder of foot intact

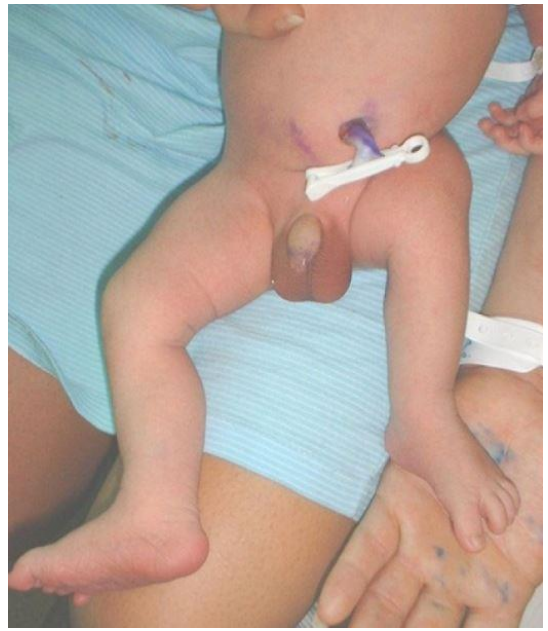
Photographs and imaging sources: CDC-Beijing Medical University collaborative project; Dr. E Gene Deune

Limb deficiency: transverse terminal

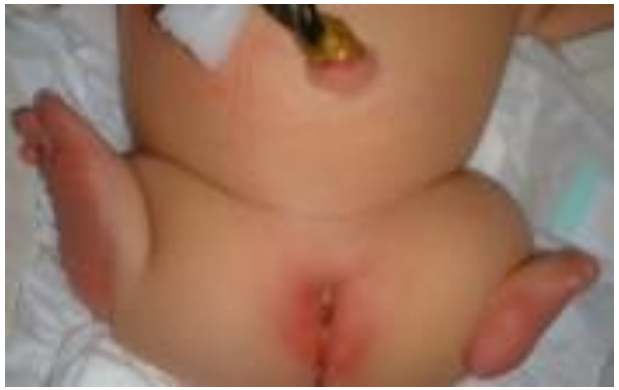




Congenital absence of upper arm and forearm with hand present



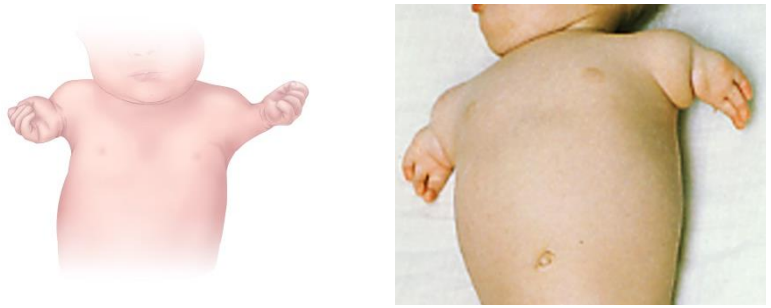
Longitudinal reduction defect of femur



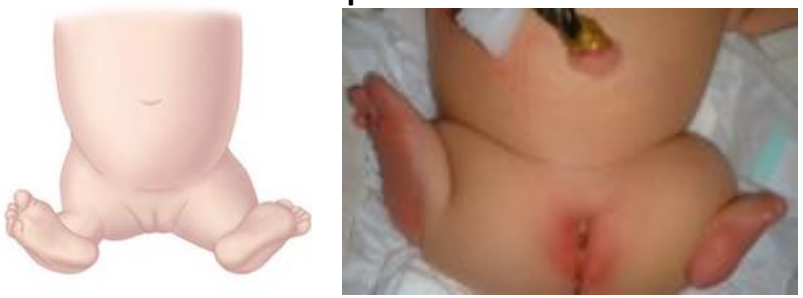
Congenital absence of thigh and lower leg with foot present

Photographs sources: Dr. Jaime Frias, ECLAMC

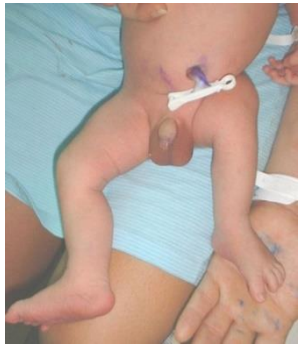
Limb deficiency: transverse intercalary



Congenital absence of upper arm and forearm with hand present



Congenital absence of thigh and lower leg with foot present



Longitudinal reduction defect of femur

Transverse intercalary:

- Absence of the proximal or middle segments of a limb, with all or part of the distal segment present

Relevant ICD-10 codes

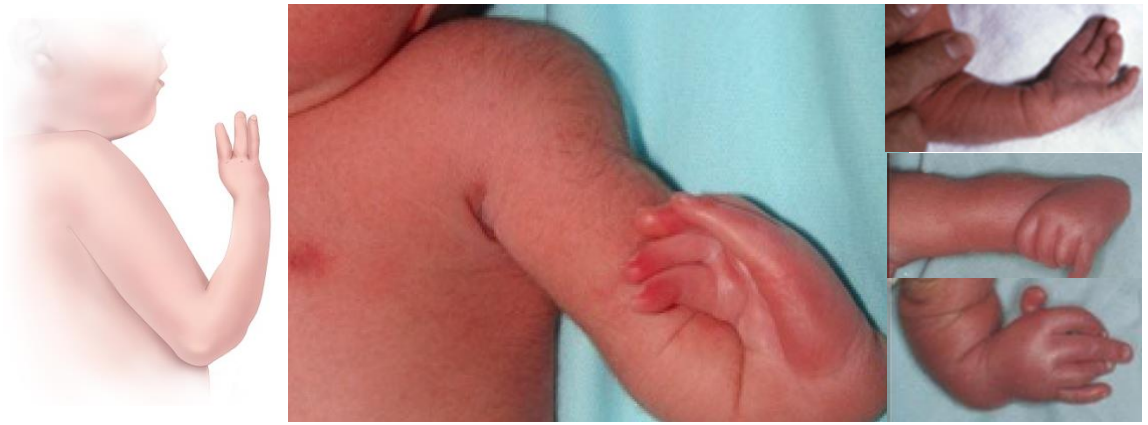
- Q71.1 Congenital absence of upper arm and forearm with hand present (phocomelia of upper limb)
- Q72.1 Congenital absence of thigh and lower leg with foot present (phocomelia of lower limb)
- Q72.4 Longitudinal reduction defect of femur (proximal femoral focal deficiency)

Photographs sources: Dr. Jaime Frias, ECLAMC

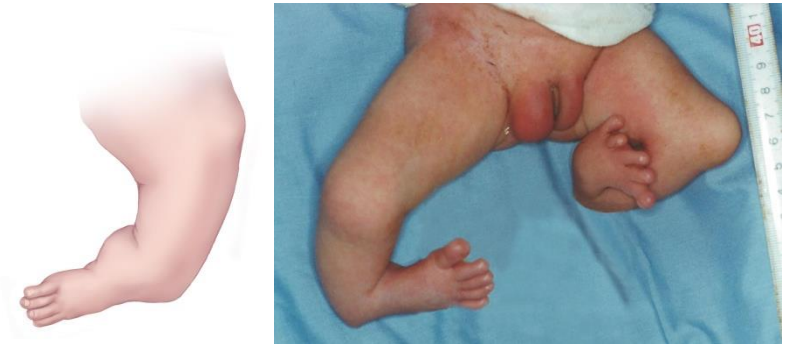
Limb deficiency: transverse terminal and transverse intercalary

Description or documentation checklist for high-quality reporting		
	Transverse terminal	**Transverse intercalary**
Describe in detail	• Involved limb • Bilateral or unilateral (right, left) • Soft tissue present • Amniotic bands present • Constriction rings present	• Involved limb and the missing segment • Bilateral or unilateral (right, left) • Other unrelated anomalies if present
Take or report photographs	• Entire infant • All extremities	• Entire infant • All extremities
Describe evaluations and procedures	• Specialty consultations (e.g., genetics, orthopedics) • Procedure and radiology reports • Genetic tests • Chromosomal studies	• Specialty consultations (e.g., genetics, orthopedics) • Procedure and radiology reports • Genetic tests • Chromosomal studies
Autopsy findings	Report if available	Report if available

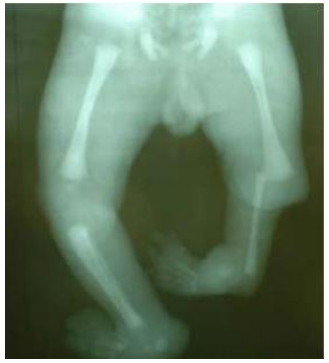
Limb deficiency: longitudinal preaxial



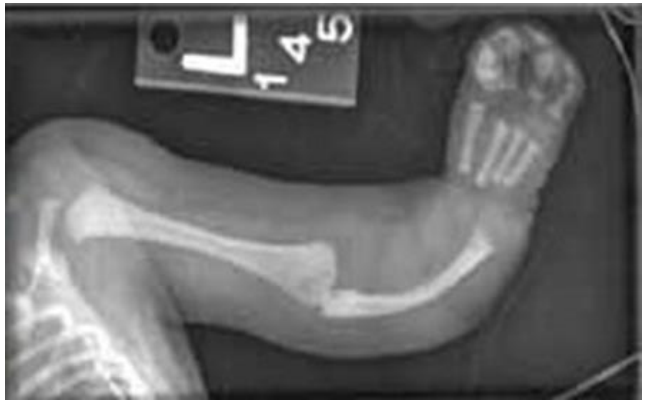
Absence/hypoplasia of thumb



Hypoplasia of first toe with other digits present



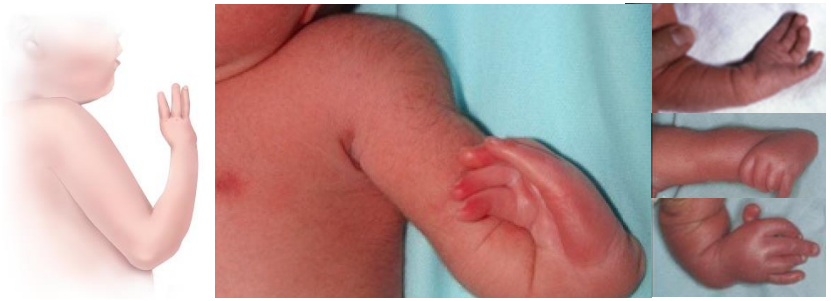
Longitudinal reduction defect of tibia



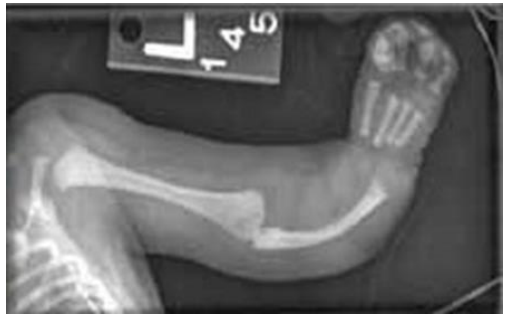
Longitudinal reduction defect of radius

Photographs and imaging sources: CDC-Beijing Medical University collaborative project, ECLAMC

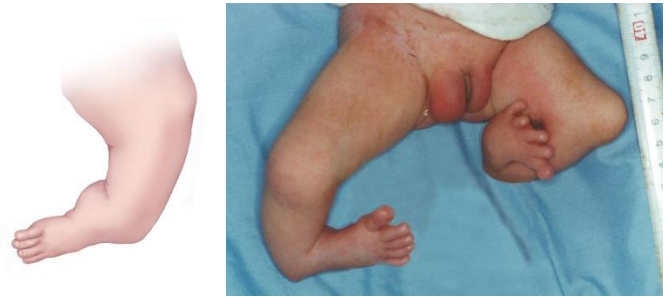
Limb deficiency: longitudinal preaxial



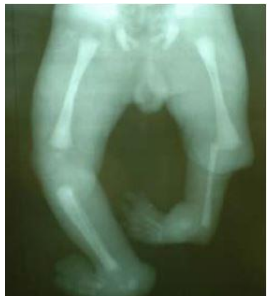
Absence/hypoplasia of thumb



Longitudinal reduction defect of radius



Hypoplasia of first toe with other digits present



Longitudinal reduction defect of tibia

Longitudinal Preaxial:

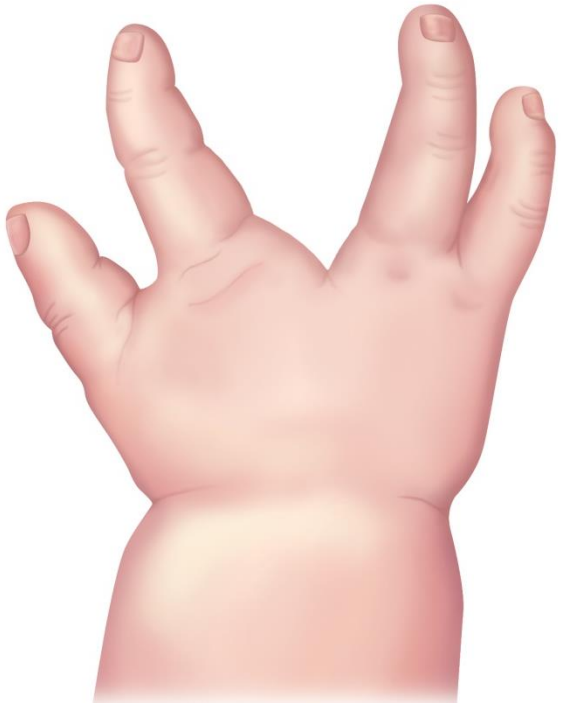
- Absence or hypoplasia of the preaxial side of the upper or lower extremity
- Affected segment may be absent or hypoplastic
- Preaxial defects include the thumb and/or radius, big toe and/or tibia

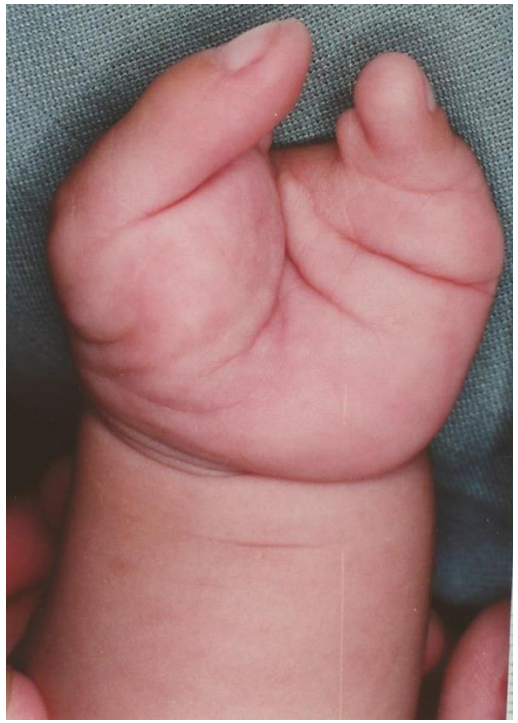
Relevant ICD-10 codes

- Q71.31 Absence or hypoplasia of thumb (other digits intact)
- Q71.4 Longitudinal reduction defect of radius (congenital clubhand, radial clubhand, absent radius)
- Q72.31 Absence or hypoplasia of first toe with other digits present
- Q72.5 Longitudinal reduction defect of tibia (absent tibia)

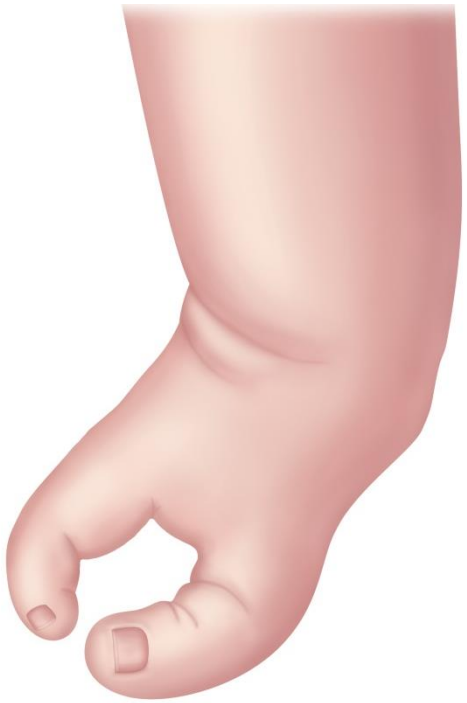
Photographs and imaging sources: CDC-Beijing Medical University collaborative project, ECLAMC

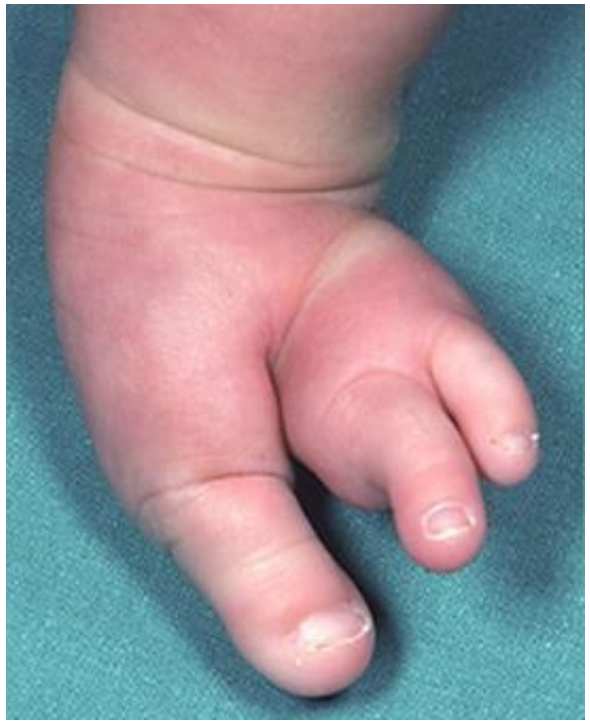
Limb deficiency: longitudinal axial





Congenital cleft hand

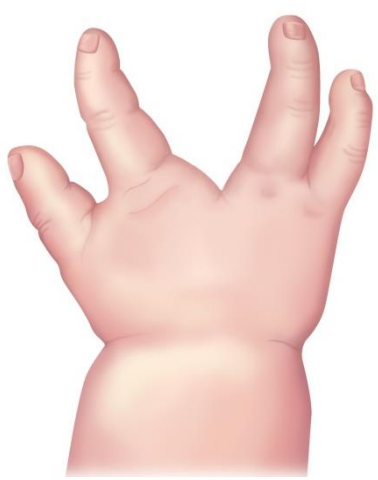


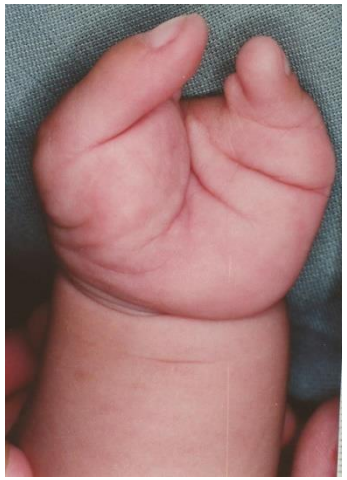


Split foot

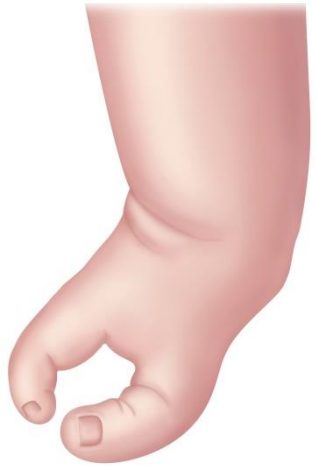
Photographs sources: CDC-Beijing Medical University collaborative project

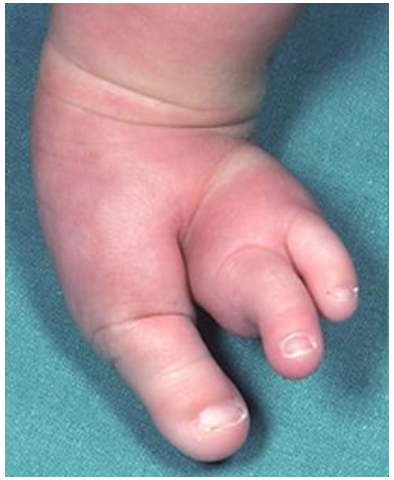
Limb deficiency: longitudinal axial





Congenital cleft hand





Split foot

Longitudinal axial:

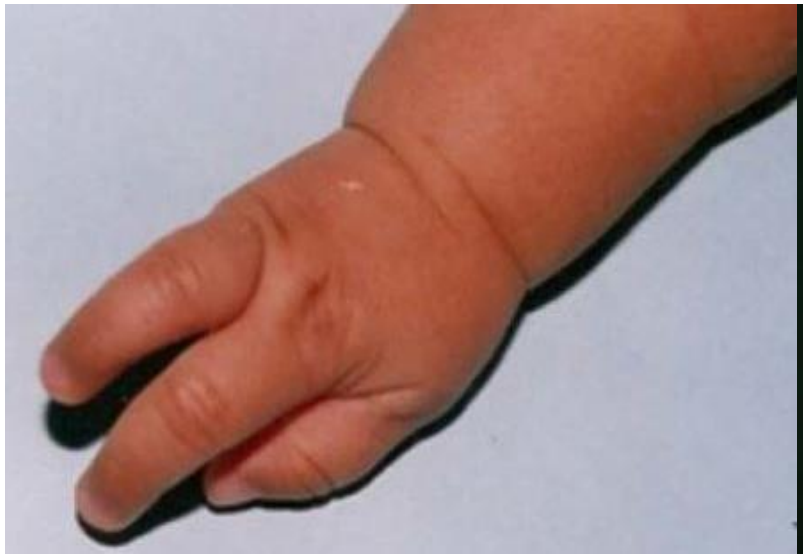
- Digital (digits and toes) are deficient centrally
- Carpal or tarsal bones may be involved which give the appearance of a split hand or split foot – may occur with deficiency of the adjacent long bones of the affected limb

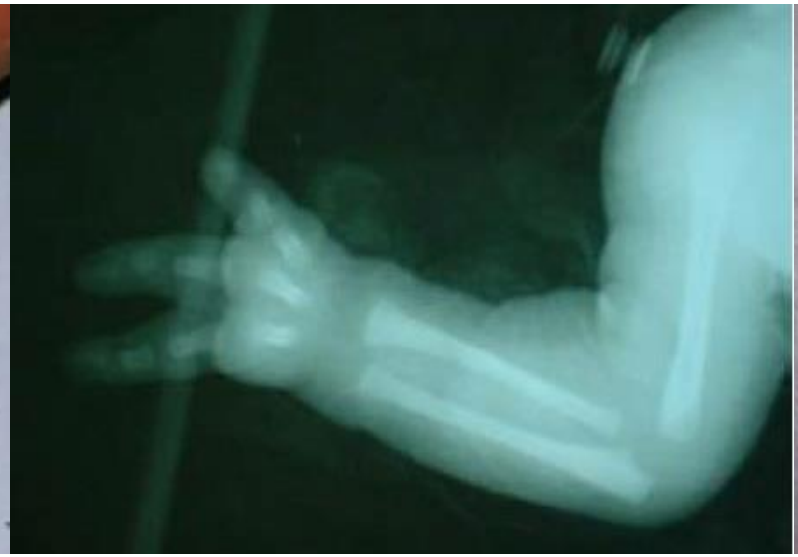
Relevant ICD-10 codes

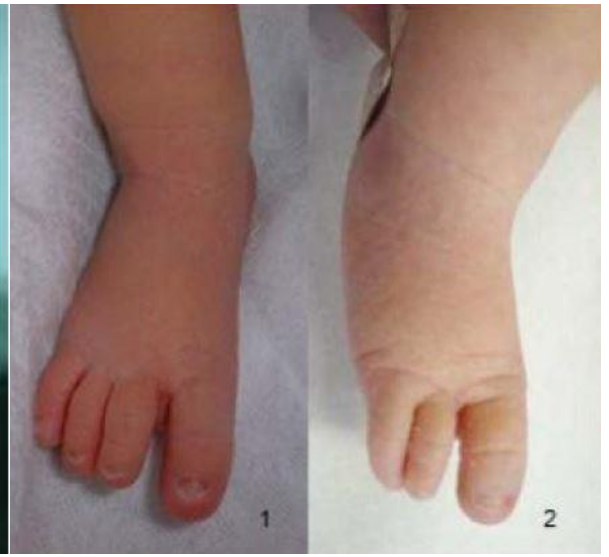
- Q71.6 Congenital cleft hand
- Q72.7 Split foot

Photographs sources: CDC-Beijing Medical University collaborative project

Limb deficiency: longitudinal postaxial





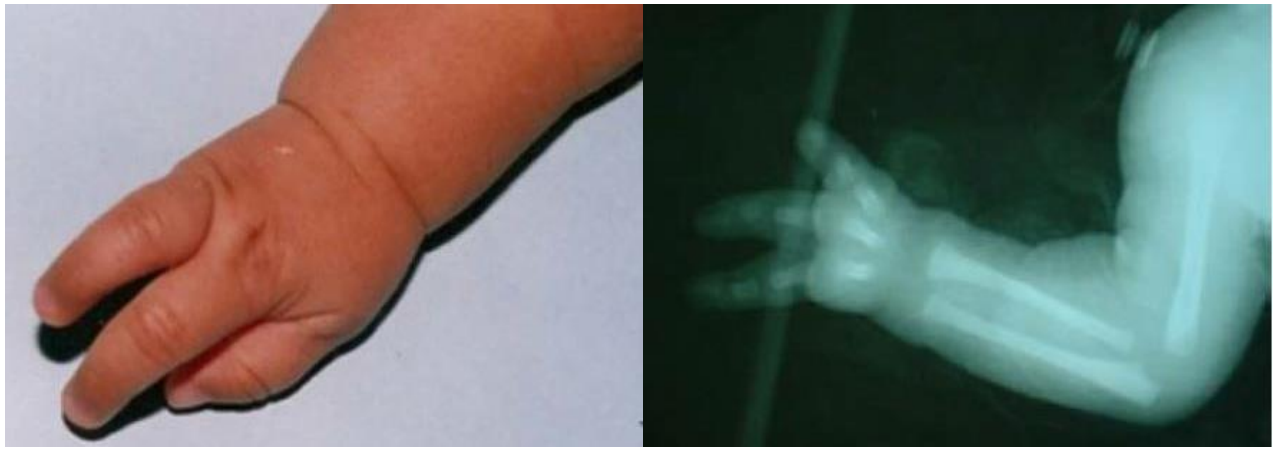


Congenital absence of fourth and fifth fingers

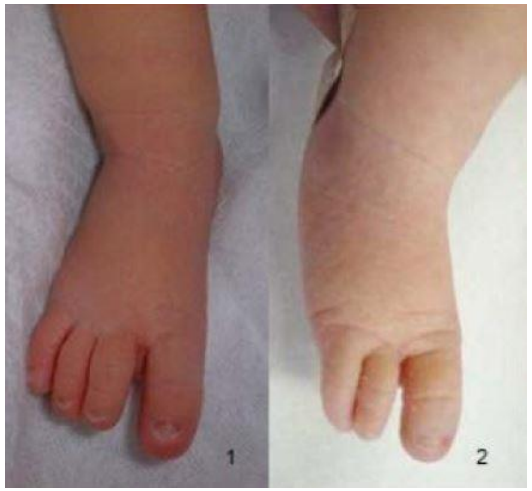
Congenital absence of toe(s) with remainder of foot intact

Photographs and imaging sources: ECLAMC

Limb deficiency: longitudinal postaxial



Congenital absence of fourth and fifth fingers



Congenital absence of toe(s) with remainder of foot intact

Longitudinal postaxial:

- Absence or hypoplasia of the fifth toe/finger and may involve the fibula/ulna

Relevant ICD-10 codes

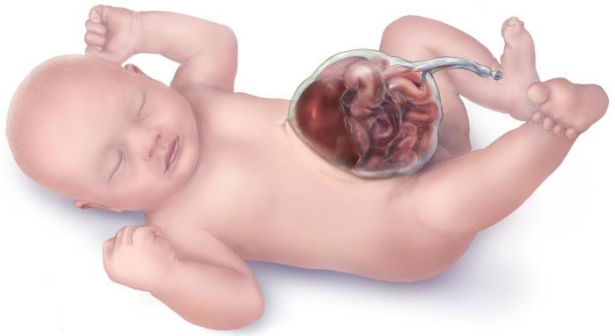
- Q71.30 Congenital absence of finger(s) with remainder of hand intact
- Q71.5 Longitudinal reduction defect of ulna
- Q72.30 Congenital absence or hypoplasia of toe(s) with remainder of foot intact
- Q72.6 Longitudinal reduction defect of fibula (absent fibula)

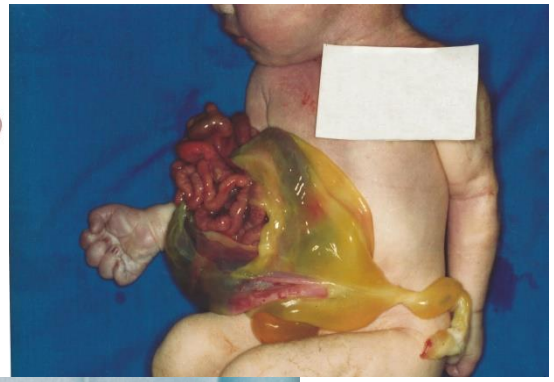
Photographs and imaging sources: ECLAMC

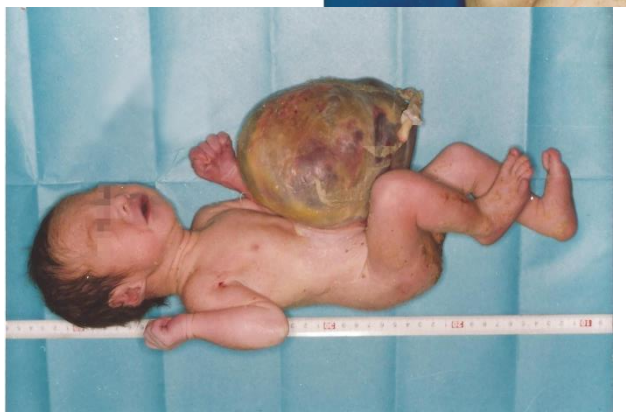
Limb deficiency: longitudinal preaxial, axial and postaxial

Description or documentation checklist for high-quality reporting			
	Longitudinal Preaxial	**Longitudinal Axial**	**Longitudinal Postaxial**
Describe in detail	• Involved limb(s) • Bilateral or unilateral (right, left) • Other anomalies (e.g., VATER/VACTERL, trisomy 18, other genetic syndromes)	• Involved limb(s) • Bilateral or unilateral (right, left) • Other anomalies if present (e.g., orofacial clefts)	• Involved digit(s) and if affected, forearm/leg • Bilateral or unilateral (right, left) • Other unrelated anomalies if present
Take or report photographs	• Entire infant • All extremities	• Entire infant • All extremities	• Entire infant • All extremities
Describe evaluations	• Specialty consultations (e.g., genetics, orthopedics) • Procedure and radiology reports • Genetic tests • Chromosomal studies	• Specialty consultations (e.g., genetics, orthopedics) • Procedure and radiology reports • Genetic tests • Chromosomal studies	• Specialty consultations (e.g., genetics, orthopedics) • Procedure and radiology reports • Genetic tests • Chromosomal studies
Autopsy	Report if available	Report if available	Report if available

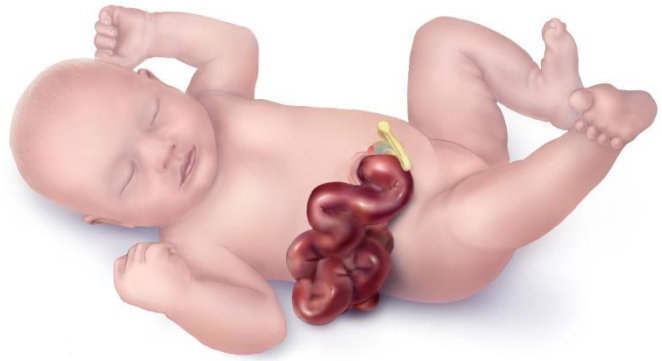
Abdominal defects: omphalocele and gastroschisis

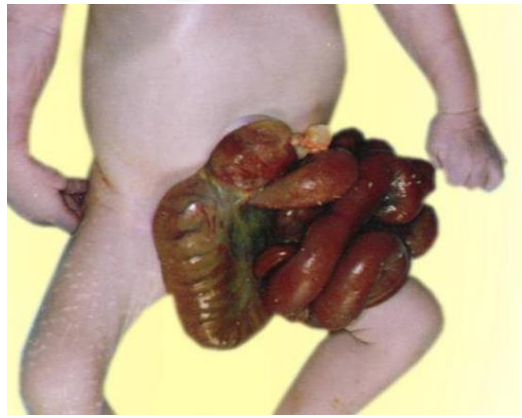






Omphalocele

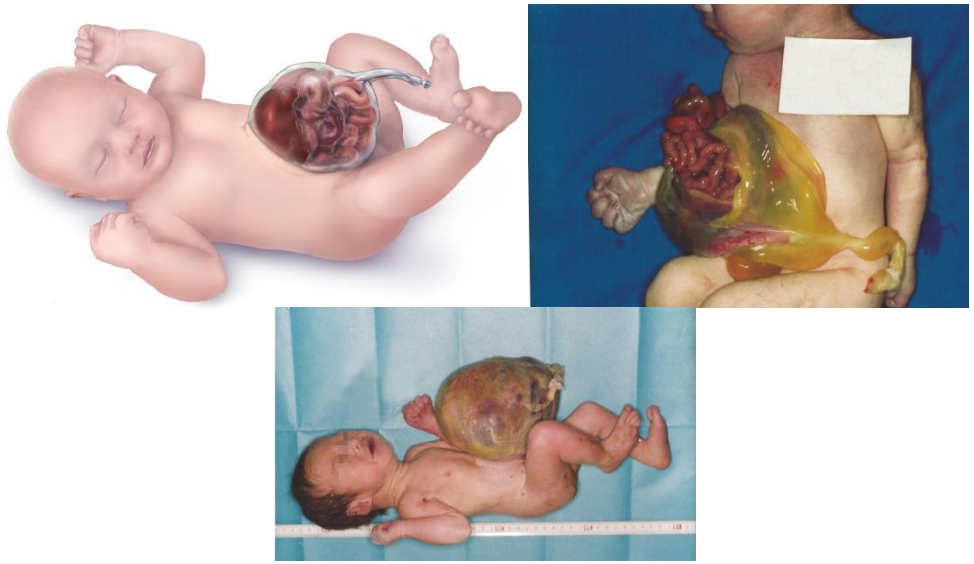




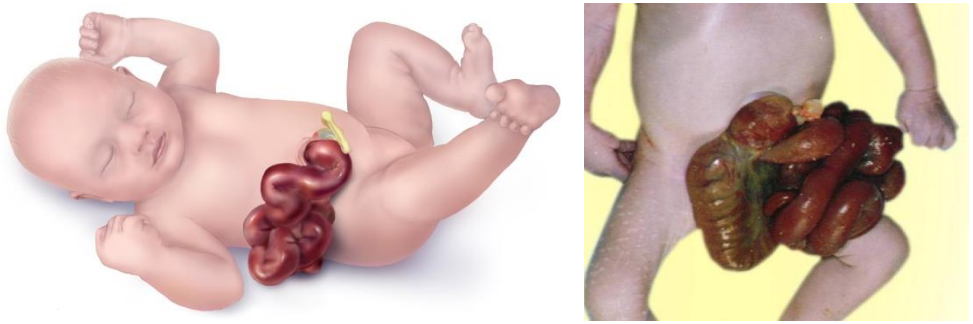
Gastroschisis

Photographs sources: CDC-Beijing Medical University collaborative project

Abdominal defects: omphalocele and gastroschisis



Omphalocele



Gastroschisis

Photographs Source: CDC-Beijing Medical University collaborative project.

Omphalocele:

- Abdominal contents are herniated through the enlarged umbilical ring
- Umbilical cord is inserted in the distal part of the membrane covering the defect
- Ruptured omphalocele is rare – difficult to distinguish between a gastroschisis
- If the umbilical ring is intact, this suggests omphalocele

Gastroschisis:

- Involves an opening, **usually** to right side of the umbilical cord which is not attached (at edge of umbilical ring)
- Umbilical cord is attached on the left side
- Extruded organs are not covered by any membrane
- Usually, an isolated anomaly
- Intestinal atresia may be present as a result of the defect (acquired, congenital)

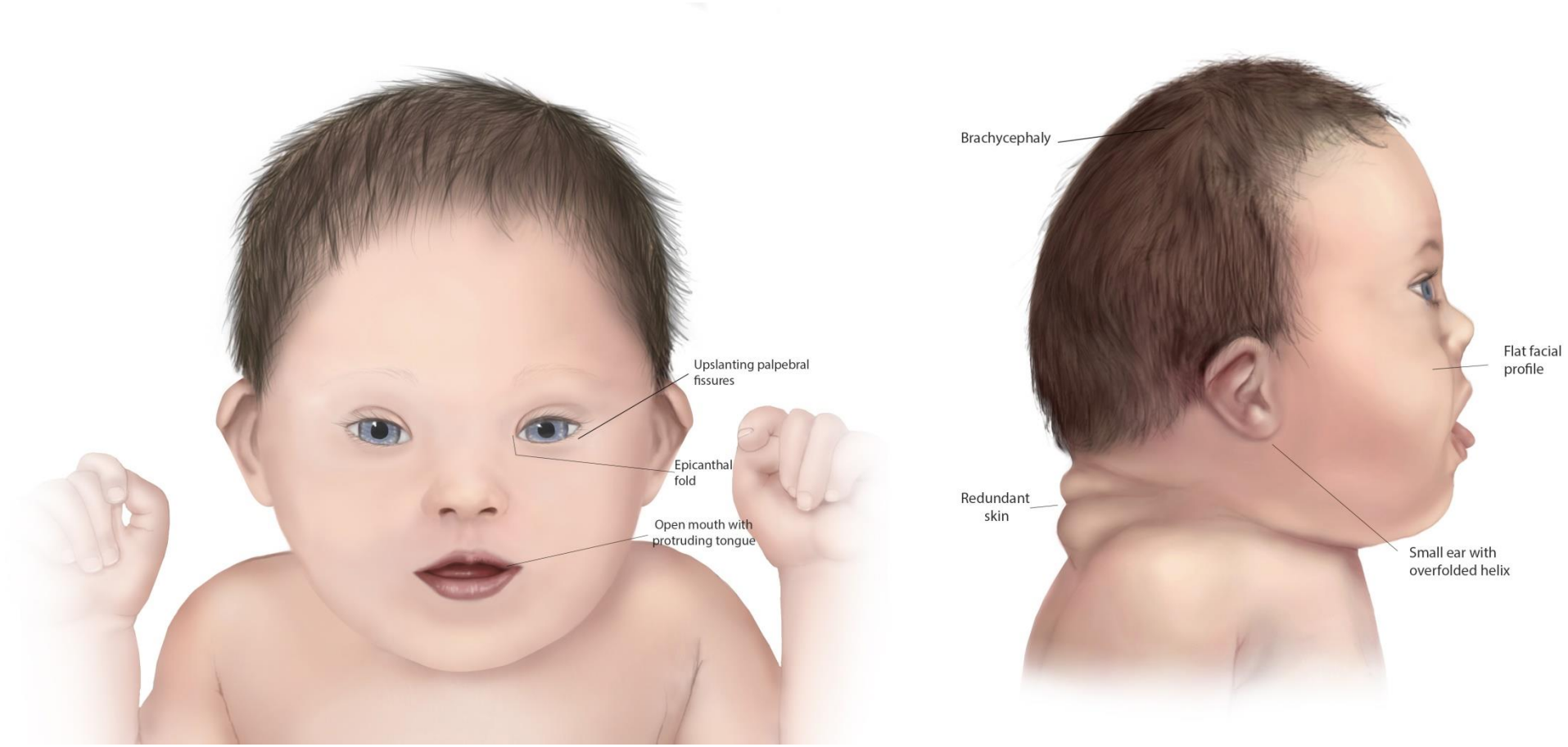
Relevant ICD-10 codes

- Q79.2 Omphalocele (exomphalos)
- Q79.3 Gastroschisis

Abdominal defects: omphalocele and gastroschisis

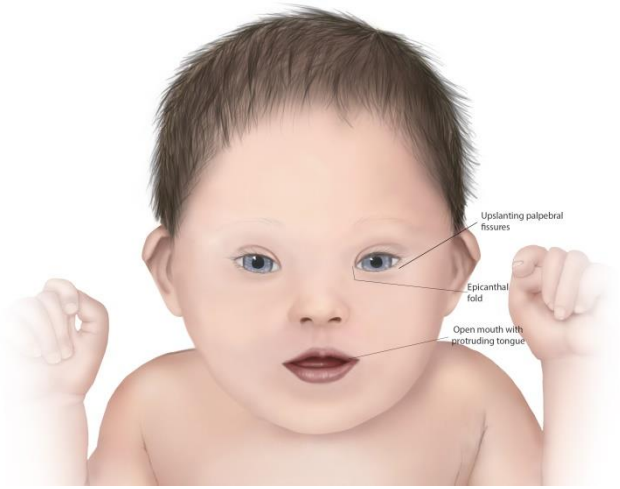
Description or documentation checklist for high-quality reporting		
	Omphalocele	**Gastroschisis**
Describe in detail	•Membrane intact and covering extruded organs •Organs extruded •Description of umbilical cord placement, size of defect •Other unrelated anomalies	•Side and size of opening •Organs extruded •Condition of intestines •Other unrelated anomalies
Take or report photographs	Show front of abdomen	Show front of abdomen
Describe evaluations and procedures	•Consultations •Surgery •Additional anomalies common •Chromosomal or genetic testing	•Consultations •Surgery
Autopsy findings	Report if available	Report if available

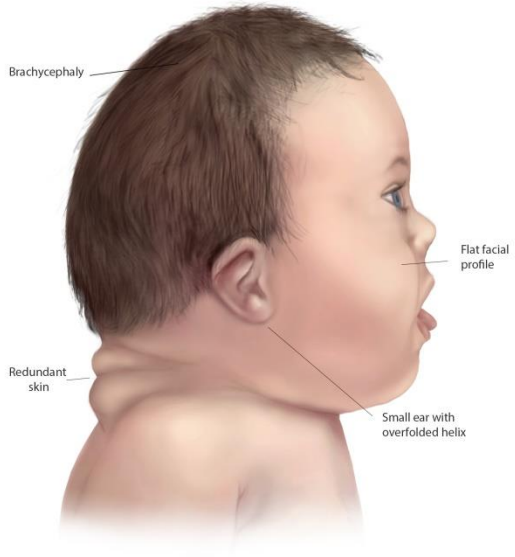
Chromosomal abnormalities: trisomy 21



Down syndrome

Chromosomal abnormalities: trisomy 21





Typical characteristics of an infant with trisomy 21 seen in these drawings include:

- Upslanting palpebral fissures
- Flat nasal bridge
- Brachycephaly
- Helix of ear folded over
- Short neck with redundant skin on back of the neck
- Other physical findings include broad/short hands and feet, single transverse palmar crease, wider spaced 1st and 2nd toes, nystagmus, clinodactyly, narrow palate, and hypotonia

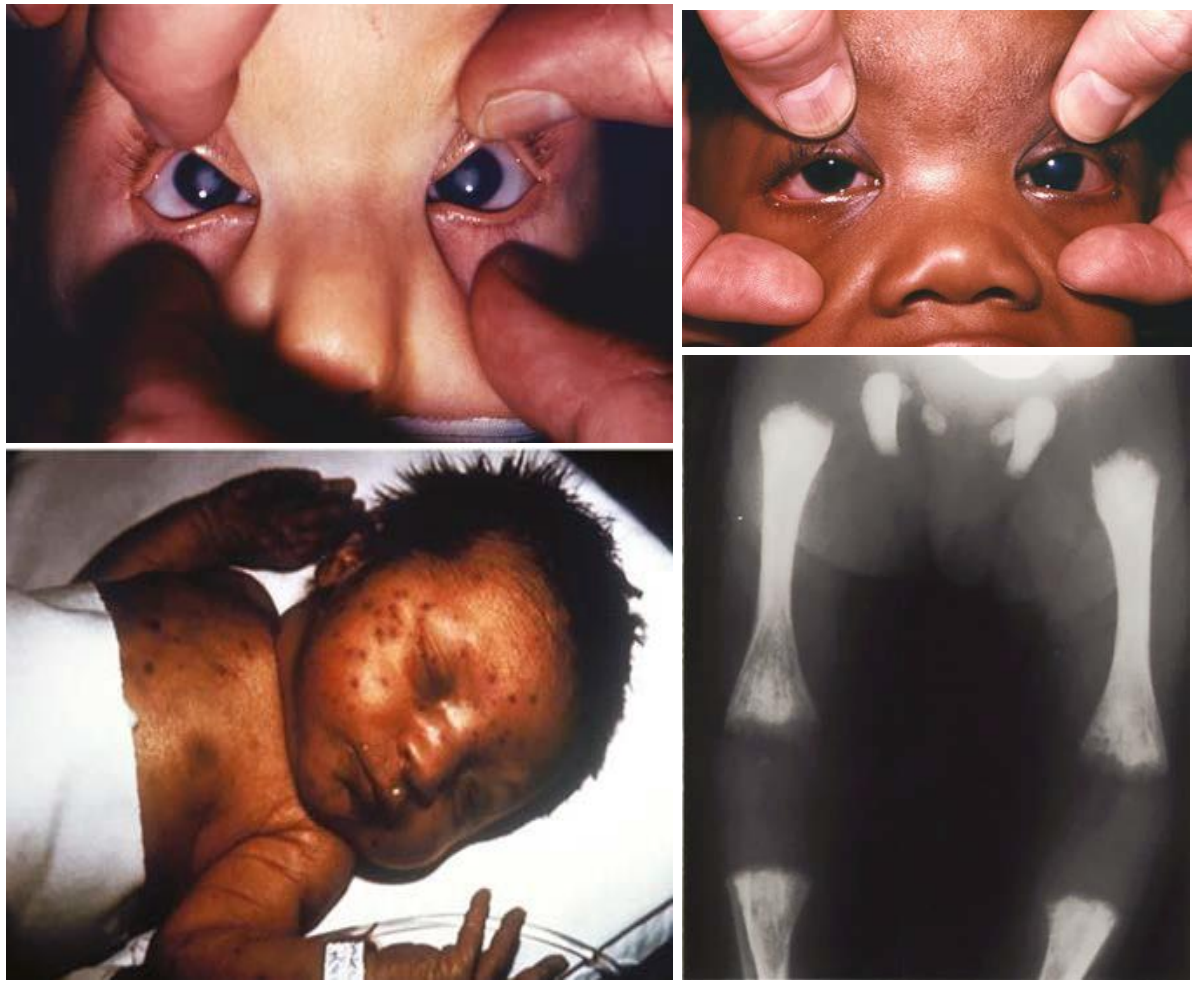
Relevant ICD-10 codes

- Q90.0 Trisomy 21, nonmosaicism (meiotic nondisjunction)
- Q90.1 Trisomy 21, mosaicism (mitotic nondisjunction)
- Q90.2 Trisomy 21, translocation
- Q90.9 Down syndrome, unspecified
- P50.9 Congenital syphilis, unspecified

Chromosomal abnormalities: trisomy 21

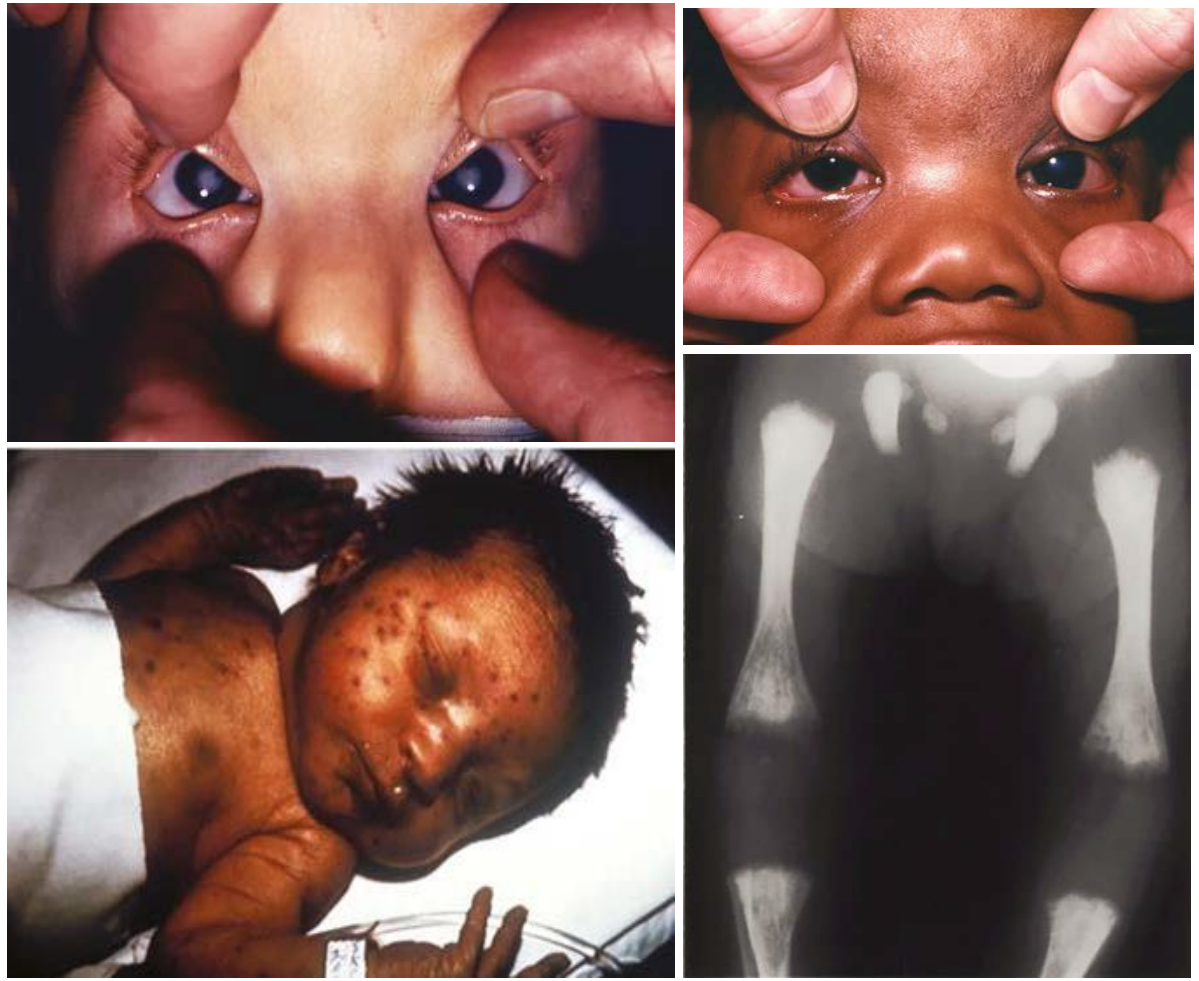
Description or documentation checklist for high-quality reporting	
	Down syndrome (trisomy 21)
Describe in detail	•Major anomalies (e.g., cardiac, duodenal or oesophageal atresia, vertebral) •Minor anomalies (e.g., facial characteristics, neck, hands/feet) •Hypotonia
Take or report photographs	•Show side and front of face •Extremities
Describe evaluations	•Consultations (e.g., genetics, cardiology) • Examinations (e.g., echocardiogram, radiology) •Surgery •Chromosomal or genetic testing
Autopsy findings	Report if available

Congenital infectious syndromes: rubella



Photograph source: CDC public health image library;
X-ray source: Government of Canada webpage (www.canada.ca/en/public-health/services/diseases/rubella/information-health-professionals-rubella.html)

Congenital infectious syndromes: rubella



Infants with congenital rubella syndrome (CRS) may have:

- Cataracts
- Chorioretinitis
- Congenital glaucoma
- Skin rash (blueberry muffin skin lesions)
- Bone disease (radiolucent)
- Other findings include congenital heart defects (branch pulmonary artery stenosis, PDA), hearing impairment, splenomegaly, microcephaly, developmental delay, low birth weight and jaundice

Relevant ICD-10 codes

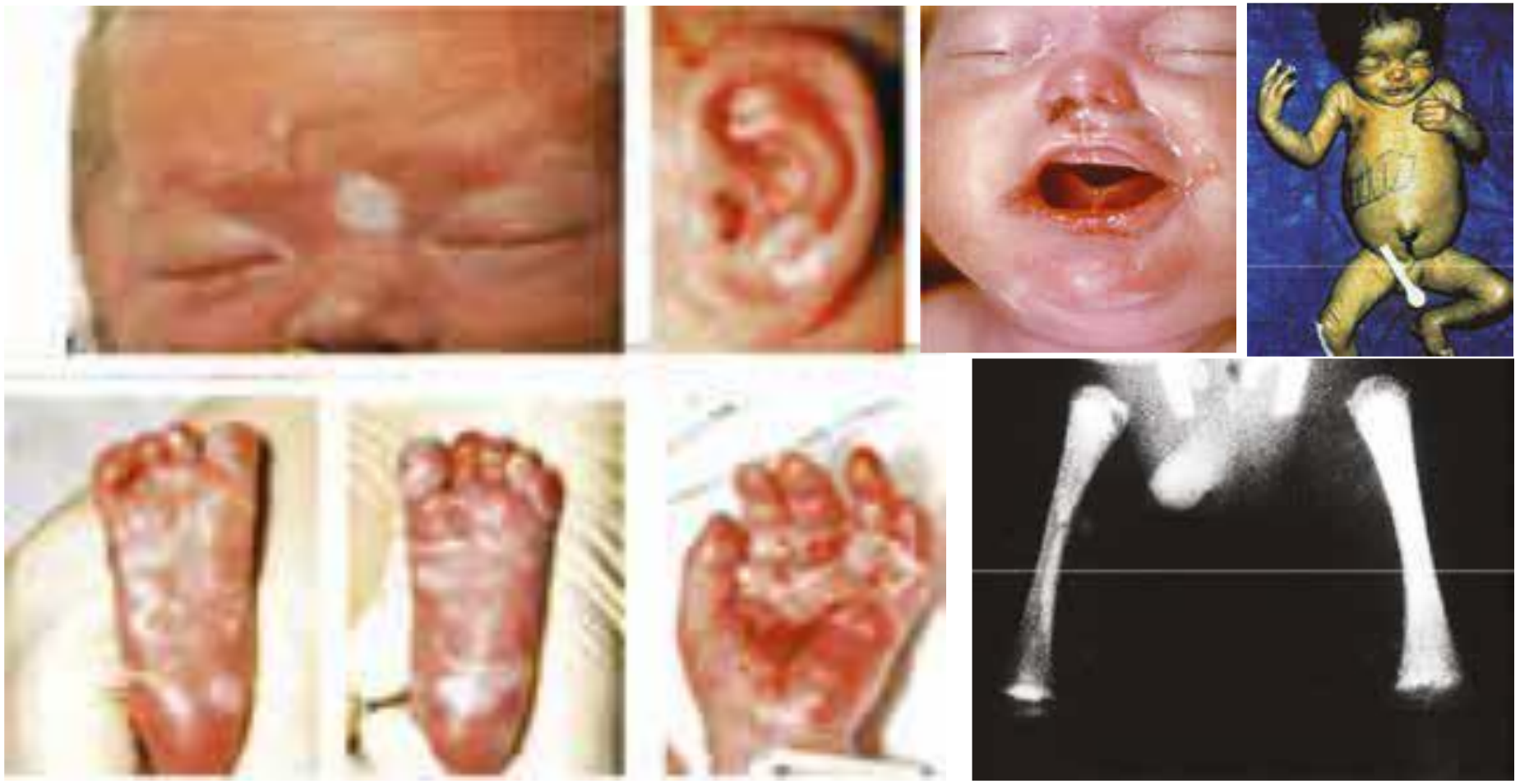
- P50.9 Congenital syphilis, unspecified
- Q12P35.0 Congenital rubella syndrome
- Q02 Microcephaly
- Q12.0 Congenital cataracts
- Q15.0 Congenital glaucoma
- Q25.0 Patent ductus arteriosus
- Q25.6 Stenosis of pulmonary artery

Photograph source: CDC public health image library;
X-ray source: Government of Canada webpage (www.canada.ca/en/public-health/services/diseases/rubella/information-health-professionals-rubella.html)

Congenital infectious syndromes: rubella

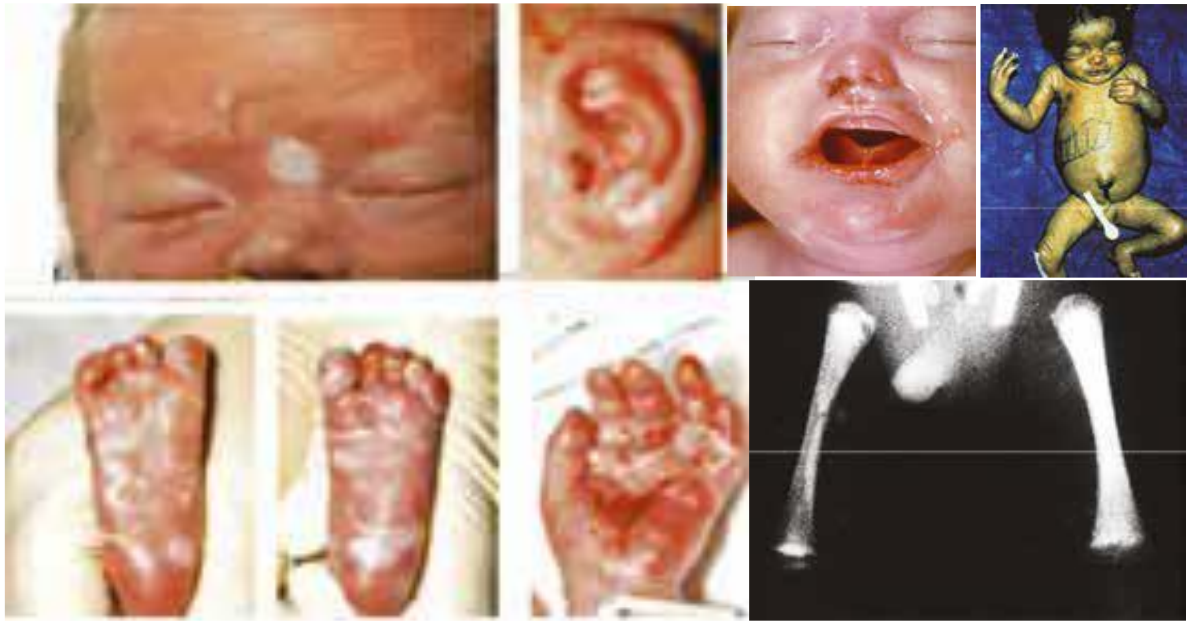
Description or documentation checklist for high-quality reporting	
	Congenital rubella syndrome
Describe in detail	•Eyes (cataracts or glaucoma or pigmentary retinopathy), skin (rash, jaundice), abdomen (splenomegaly), heart murmur •Maternal history of rubella in pregnancy
Take or report photographs	• Show head and face (front and side) •Full body (front and back)
Describe evaluations	•Consultations (e.g., genetics, cardiology, ophthalmology) •Examinations (e.g., echocardiogram, radiology, hearing) •Surgery •Rubella antibody testing
Autopsy findings	Report if available

Congenital infectious syndromes: syphilis



Photograph and X-ray source: Dr Ronald Ballard

Congenital infectious syndromes: syphilis



Infants may have:

- Skin rash (desquamation, blistering, pustules)
- Jaundice
- Rhinitis
- Bone disease (metaphysitis, osteochondritis, diaphyseal osteomyelitis, periostitis)
- Eye abnormalities (chorioretinitis, keratitis, glaucoma, cataracts, optic neuritis)
- Hearing impairment
- Hepatosplenomegaly

Relevant ICD-10 codes

- P50.9 Congenital syphilis, unspecified
- Q12.0 Congenital cataracts
- Q15.0 Congenital glaucoma

Photograph and X-ray source: Dr Ronald Ballard

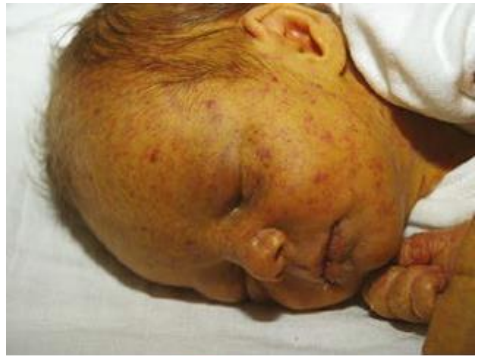
Congenital infectious syndromes: syphilis

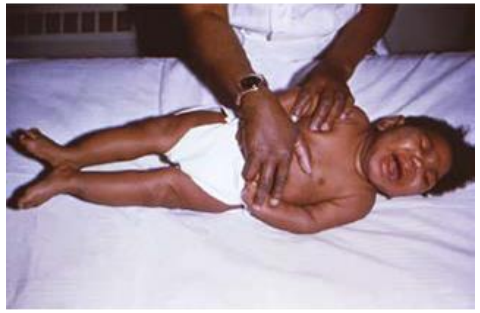
Description or documentation checklist for high-quality reporting	
	Congenital syphilis
Describe in detail	•Eyes (cataracts, glaucoma, other findings), skin (rash, jaundice, pustules), rhinitis, and abdomen (hepatosplenomegaly) •Maternal syphilis infection
Take or report photographs	•Show head and face (front and side) •Full body (front and back) •Extremities
Describe evaluations	•Consultations (e.g., genetics, cardiology, ophthalmology) •Examinations (e.g., echocardiogram, radiology, hearing) •Antibody testing, nucleic acid amplification tests
Autopsy findings	Report if available

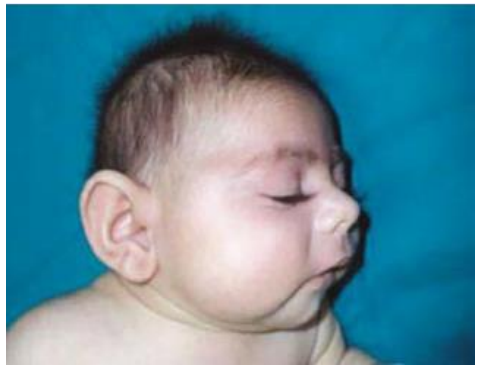
Congenital infectious syndromes: cytomegalovirus (CMV)

Photograph sources: Jacob Johan; CDC Public Health Image Library; Isikay S, Yilmaz K. Congenital cytomegalovirus infection and finger anomaly. BMJ Case Rep. 2013 Jun 10;2013:bcr2013009486.

Congenital infectious syndromes: cytomegalovirus (CMV)







Infants with congenital cytomegalovirus may have:

- Skin rash (petechiae/blueberry muffin)
- Jaundice
- Hepatosplenomegaly
- Microcephaly
- Ascites/hydrops
- Growth restriction
- Hepatitis
- Thrombocytopenia
- Anaemia
- Seizures
- Chorioretinitis
- Hearing impairment

Relevant ICD-10 code

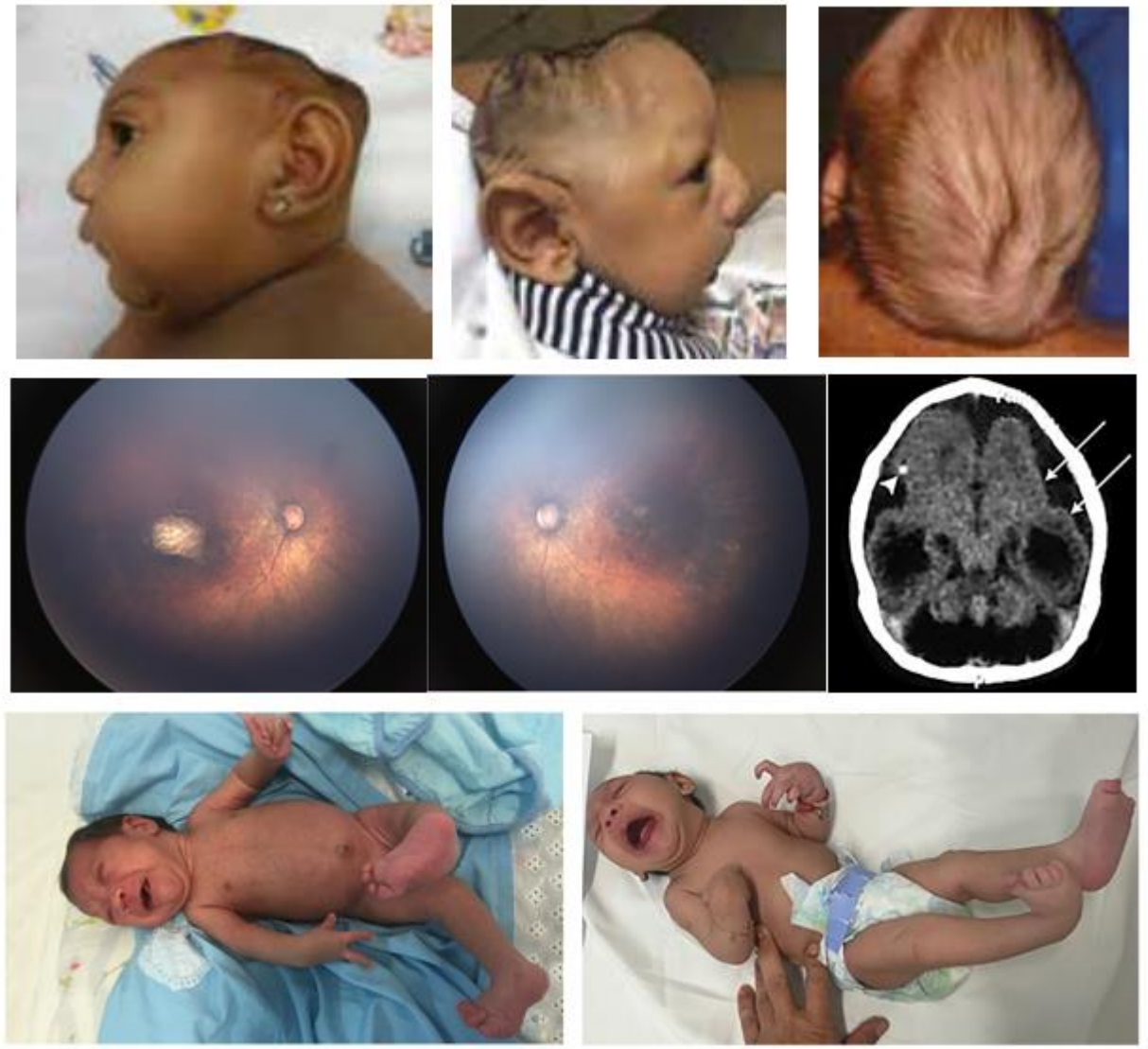
- P35.1 Congenital cytomegalovirus infection (CMV)

Photograph sources: Jacob Johan; CDC Public Health Image Library; Isikay S, Yilmaz K. Congenital cytomegalovirus infection and finger anomaly. BMJ Case Rep. 2013 Jun 10;2013:bcr2013009486.

Congenital infectious syndromes: cytomegalovirus (CMV)

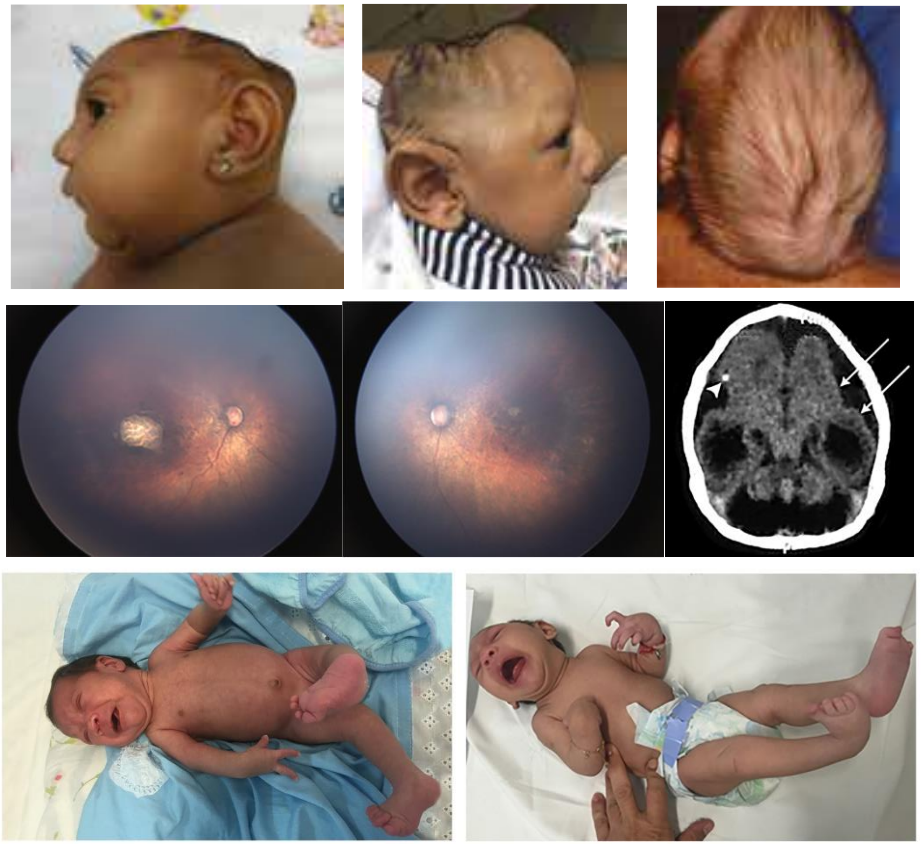
Description or documentation checklist for high-quality reporting	
	Congenital Cytomegalovirus (cCMV)
Describe in detail	• Head (circumference) size and if brain calcifications are present, eyes (cataracts, glaucoma, chorioretinitis), skin (petechial rash, jaundice), and abdomen (hepatosplenomegaly) • Maternal infection if available
Take or report photographs	• Show head and face (front and side) • Full body (front and back) • Extremities
Describe evaluations	• Consultations (e.g., genetics, ophthalmology, neurology) • Examinations (e.g., brain, hearing) • Antibody testing, nucleic acid amplification tests, PCR
Autopsy findings	Report if available

Congenital infectious syndromes: Zika



Photographs and imaging source: Moore CA, Staples JE, Dobyns WB, et al. Characterizing the Pattern of Anomalies in Congenital Zika Syndrome for Pediatric Clinicians. JAMA Pediatr. 2017;171(3):288–295. doi:10.1001/jamapediatrics.2016.3982.

Congenital infectious syndromes: Zika



Infants with congenital Zika syndrome have five unique features:

1. Severe microcephaly
2. Thin cerebral cortices with subcortical calcifications
3. Macular scarring and focal pigmentary retinal mottling
4. Congenital contractures of major joints (arthrogryposis)
5. Marked early hypertonia or spasticity and symptoms of extrapyramidal involvement in those with brain anomalies

Relevant ICD-10 codes

- P35.4 Congenital Zika disease
- Q02 Microcephaly

Photographs and imaging source: Moore CA, Staples JE, Dobyns WB, et al. Characterizing the Pattern of Anomalies in Congenital Zika Syndrome for Pediatric Clinicians. JAMA Pediatr. 2017;171(3):288–295. doi:10.1001/jamapediatrics.2016.3982.

Congenital infectious syndromes: Zika

Description or documentation checklist for high-quality reporting	
	Congenital Zika syndrome
Describe in detail	• Head size, circumference (microcephaly) • Any other anomalies • Contractures • Eyes (optic nerve hypoplasia, pigmentary mottling, chorioretinal scarring) • Maternal zika infection
Take or report photographs	• Show head and face (front and side) • Full body (front and back) • Extremities
Describe evaluations	• Consultations (e.g., genetics, ophthalmology, neurology) • Examinations (e.g., brain imaging, hearing) • Maternal: NAAT, PCR, antibody test • Infant: RNA test for virus
Autopsy findings	Report if available

Facilitator's Guide

Module 5: Coding

World Health Organization

CDC

International Clearinghouse for Birth Defects Surveillance and Research

Objectives

By the end of this module, participants will be able to:

- Understand the importance of coding
- Understand the importance of good clinical description and documentation for accurate coding
- Describe advantages and disadvantages of the ICD-10 and ICD-10 RCPCH extension
- Identify critical issues for coding

Overview: International Statistical Classification of Diseases and Related Health Problems, 10th Revision (ICD-10)

- Developed and maintained by WHO
- Considered the international standard diagnostic classification system
- Recent version of the ICD-10 is available on the WHO website (https://icd.who.int/browse10/2019/en) or the 2008 version used by EUROCAT (https://eu-rd-platform.jrc.ec.europa.eu/sites/default/files/EUROCAT-Q-Chapter-2008.pdf)
- Widely used in many countries as a classification system for diseases
- Codes are listed in alpha-numeric order and are grouped into chapters by topic areas such as diseases, organ systems, causes, health services encounters
  - Classification of structural congenital anomalies is found in Chapter XVII: Congenital malformations, deformations and chromosomal abnormalities (Q00–Q99)
  - Two other relevant chapters (IV and XVI) include codes for non-structural anomalies that may be used:
    1. Classification of congenital adrenal hyperplasia (E25.0) and albinism (E70.3) found in Chapter IV: Endocrine, Nutritional and Metabolic Diseases
    2. Classification of congenital infections (P35.0 Congenital Rubella syndrome, P35.1 Congenital cytomegalovirus infection) found in Chapter XVI: Certain Conditions Originating in the Perinatal Period
- Useful for analysis and assessment of the health situation of population groups and monitoring the incidence and prevalence of diseases and other health conditions

Coding: specificity

- ICD-10 codes lack specificity for some congenital anomalies and most genetic syndromes
- Surveillance programmes may use their own local modification of the ICD-10:
  - Includes additional codes for congenital anomalies not found in the ICD-10 (see (https://eu-rd-platform.jrc.ec.europa.eu/sites/default/files/EUROCAT-Q-Chapter-2008.pdf)
  - Add an extra digit to allow for more detailed coding of some anomalies and specificity of diagnoses
- The Royal College of Paediatrics and Child Health (RCPCH) developed an adaptation of the ICD-10
  - Commonly used by programmes for public health surveillance of congenital anomalies
- Example of Coding Specificity between ICD-10 and RCPCH:
  - Look at Figure 5. Encephalocele from the QRH, page 10
  - When the ICD-10 code is not specific enough, for example, “Q01.8 Encephalocele of other sites,” then using the classification developed by the RCPCH could be beneficial. For this example, relevant RCPCH codes include: “Q01.80 Parietal encephalocele”, “Q01.81 Orbital encephalocele”, “Q01.82 Nasal encephalocele”, “Q01.83 Nasopharyngeal encephalocele”
- Be as specific as information permits to code each congenital anomaly

Coding: certainty of diagnosis

- Diagnosis may vary for live births vs. stillbirths and for prenatal vs. postnatal assessments
- Prenatal diagnosis in pregnancy terminations may not be verified:
  - Method of termination
  - Condition of the specimen
  - No post-termination examination or autopsy
- More information on inclusion of prenatal diagnosis is available from the National Birth Defects Prevention Network (USA) guidelines (https://www.nbdpn.org/docs/NBDPN_Prenatal_Chapter_Final%200412_noApp.pdf)
- Neonates who die shortly after birth may not have a confirmed diagnosis if certain examinations (X-rays, autopsy) are not done
- It is beneficial to code possible diagnoses differently from confirmed diagnoses (for analytic purposes)
  - Use a separate field on the congenital anomalies abstraction form to include this information
- An extra digit can be added to the ICD-10-RCPCH codes

Coding: responsible personnel

- If coding is done at the hospital, someone knowledgeable about congenital anomalies should review and confirm diagnoses and assign codes
- Codes should be reviewed and verified at the central registry level
- Final code will always be at the central registry where final review and verification of all codes reported by participating sites occurs
- Important to train hospital staff responsible for diagnosis and coding of congenital anomalies
- Not every site will have hospital personnel who are knowledgeable about congenital anomalies; if no knowledgeable staff are available, it is suggested that coding be done at the central registry level

Coding: multiple congenital anomalies

- Approximately 75% of babies with major congenital anomalies present as isolated anomalies
- The remaining 25% have one or more major anomalies and may also have one or more minor anomalies
- Detailed description of each major anomaly should always be recorded when more than one congenital anomaly is present
- Most congenital anomalies surveillance programmes allow for coding at least 10 anomalies
- Major anomalies should always be coded on the data-collection form before minor anomalies, when filling the available coding spaces; coding major anomalies in cranio-caudal order can be helpful, especially when a review is necessary
- A thorough description of the observed anomaly is important for accurate diagnosis and coding

More details about the types of congenital anomalies according to clinical presentation is available in Appendix C of the MPM2

Coding: considerations

Coding of recorded diagnostic information needs to be accurate:

- Central to process of generating valid and reliable information within surveillance system
- Achieved by following a standardized coding system (ICD-10 or ICD-10-RCPCH)
- Important to obtain the best possible clinical descriptions
  - Careful review and an accurate classification will result in assignment of the correct code(s) for the congenital anomaly
  - Precise clinical descriptions can improve the accuracy of disease classification and coding
- Clinical descriptions are recorded on the data-collection tool or abstraction form during data collection
- Photographs of external congenital anomalies supplement clinical description and help with proper and accurate coding
- Information should be coded and entered into an electronic system whenever possible, to allow for easy retrieval and analysis when needed for reporting purposes

More details about the types of congenital anomalies according to clinical presentation is available in Appendix C of the MPM2

Clinical scenario 1: baby Adaeze

Spina bifida and selected related topics

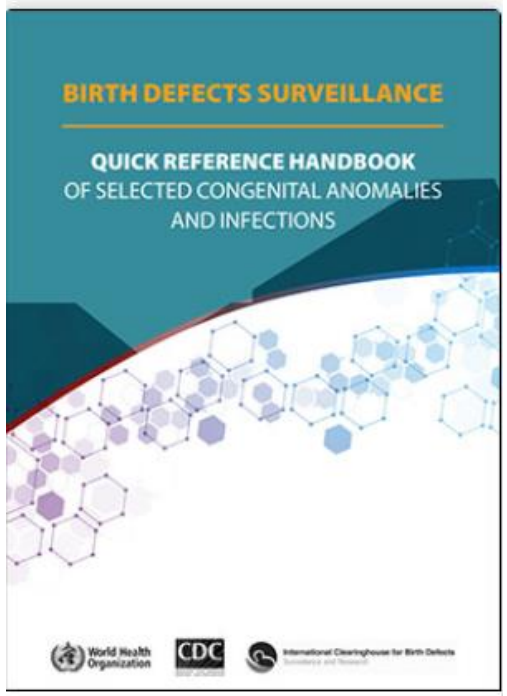
World Health Organization

CDC

International Clearinghouse for Birth Defects Surveillance and Research

Clinical scenario: baby Adaeze

Topics: spina bifida, spina bifida sequence, trisomy 18, folic acid



Learning objectives – by the end of the case study, you will be able to:

- Recognize spina bifida
- Use the QRH to describe specific findings in spina bifida (location, skin covering, etc.) and possible presence of other anomalies
- Relate spina bifida to the anomalies of the spina bifida sequence
- Code cases of spina bifida accurately
- Understand the relation between folic acid insufficiency and risk for spina bifida

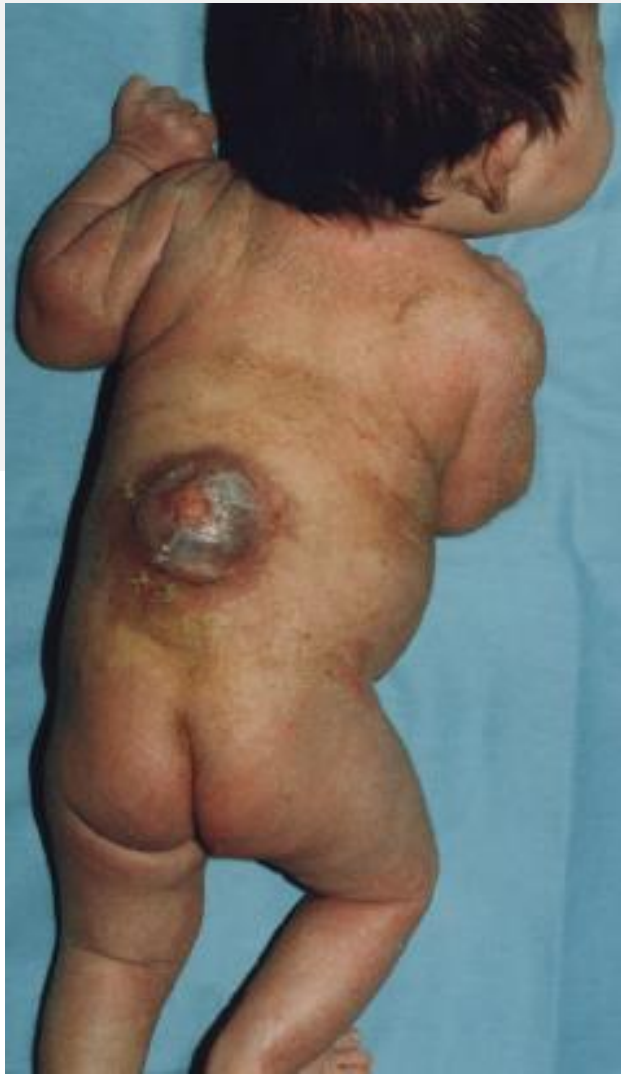
More information is available in the QRH:
https://apps.who.int/iris/handle/10665/338485

World Health Organization
CDC
International Clearinghouse for Birth Defects Surveillance and Research

Part 1: Clinical Scenario

Clinical scenario: baby Adaeze

Baby girl Adaeze is born at term to a 36-year-old mother after an apparently uneventful pregnancy. At the initial exam right after birth, *the midwife sees an unusual area on the baby's back*. She is concerned that this could be clinically significant and calls you to the bedside.



Photograph source: © CDC–Beijing Medical University collaborative project

You now examine the baby.

Q1. Describe in detail what you see, then use the visual aids and description checklist in the QRH to check and revise your description.

Note: see the QRH

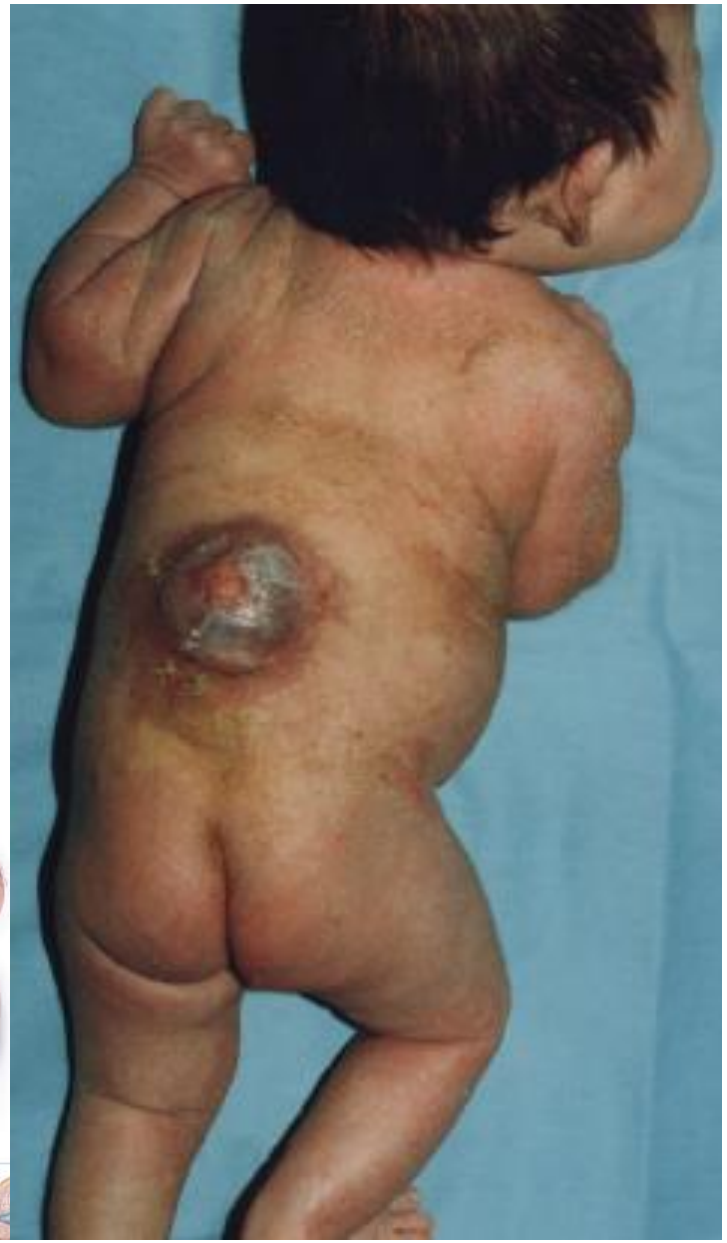
Checklist for high-quality reporting

Spina Bifida – Documentation Checklist

- ☐ **Describe defect in detail:**
  - Location – specify level (e.g. cervical, thoracic, thoraco-lumbar, lumbar, lumbosacral, sacral, etc.).
  - Size of lesion.
  - Covering – covered by skin or not covered by skin.
  - Content – only meninges (meningocele) or also spinal cord (myelomeningocele – spinal cord visible).
  - Anomalies – document sequence defects (hydrocephalus, talipes) and other anomalies.
- ☐ **Take and report photographs:** *Show clearly* the level of spina bifida (back and side if possible); can be crucial for review.
- ☐ **Describe evaluations to find or rule out related and associated anomalies:**
  - Sequence – hydrocephalus, talipes, other.
  - Other unrelated anomalies – describe procedures to assess other anomalies.
  - Genetic or chromosomal conditions.
  - Specialty consultations, imaging and surgery.
- ☐ **Report whether autopsy (pathology) findings are available and if so, report the results.**

Checklist

Anatomy



Photograph source: © CDC–Beijing Medical University collaborative project

Checklist for high-quality reporting

Spina Bifida – Documentation Checklist

- ☐ **Describe defect in detail:**
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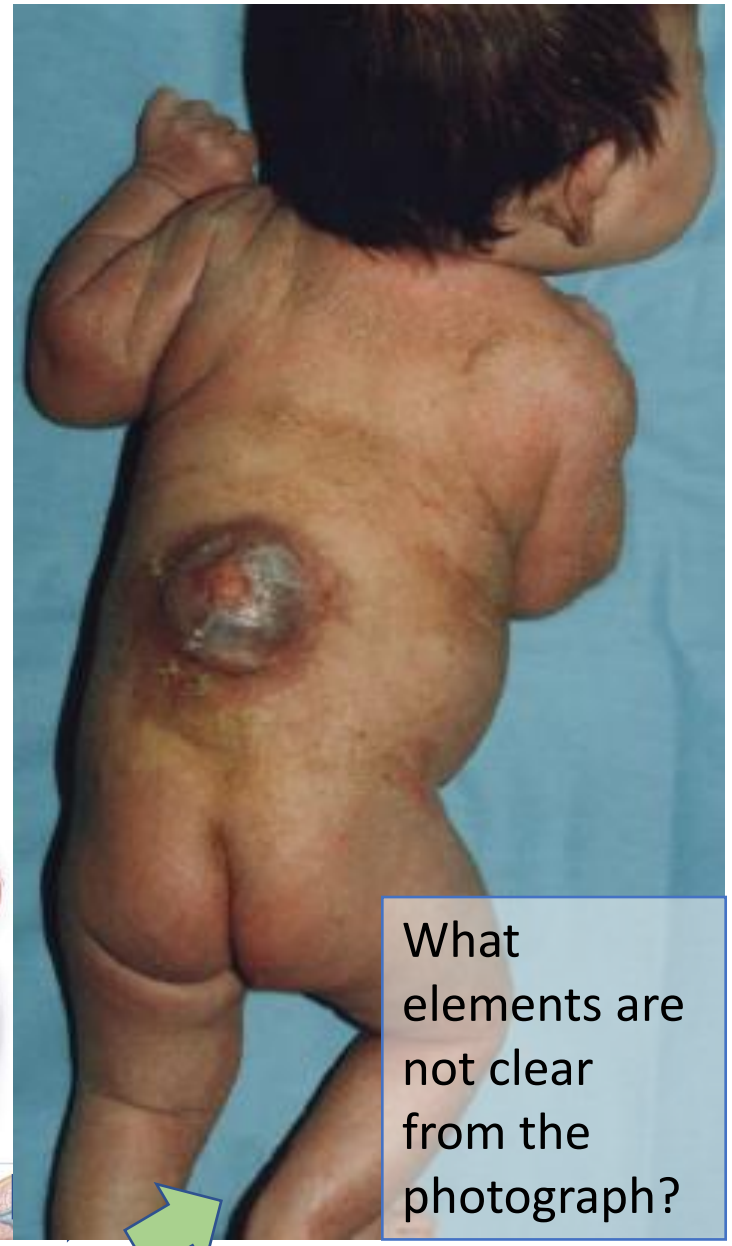
  - Specialty consultations, imaging and surgery.
- ☐ **Report whether autopsy (pathology) findings are available and if so, report the results.**



What elements are not clear from the photograph?

- **Location**: thoracic (lower); in a baby the iliac crest is approximately at the levels of L2
- **Covering**: not skin covered
- **Size**: can use a tape measure (not in photo) – visually ~ 5 cm in diameter
- **Content**: difficult to tell from photograph
- **Anomalies**: see later

Photograph source: © CDC–Beijing Medical University collaborative project

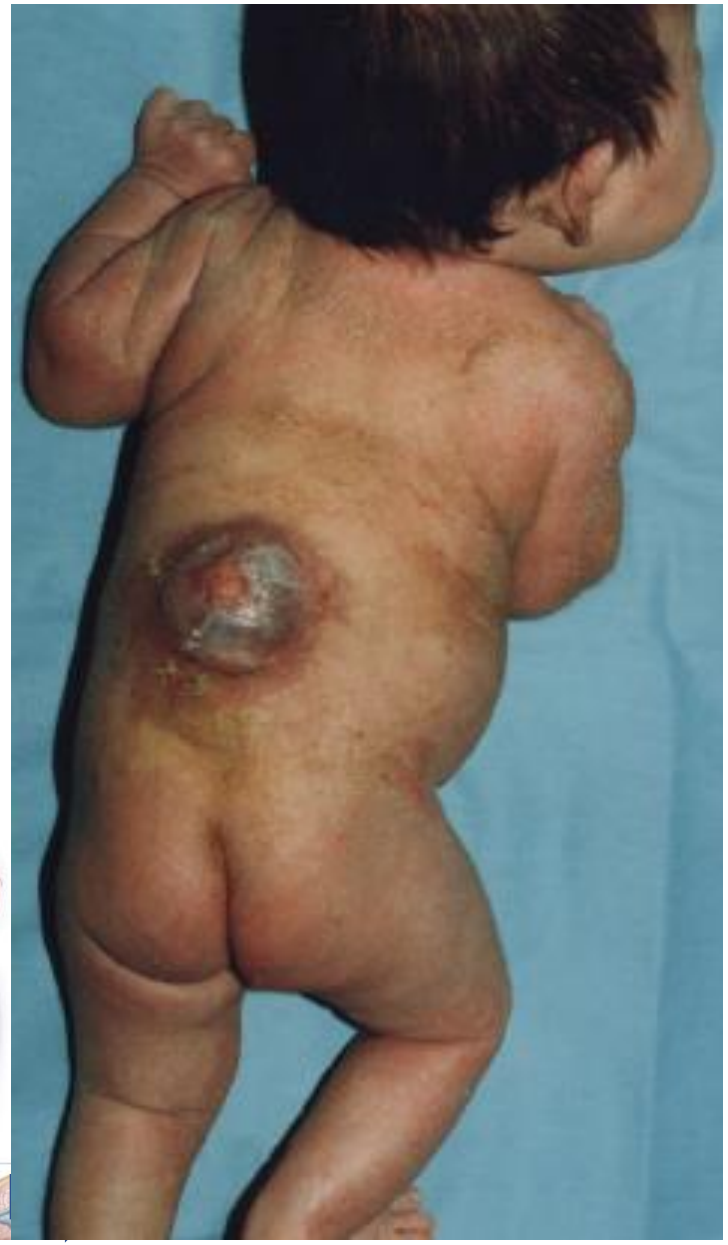
Checklist for high-quality reporting

Spina Bifida – Documentation Checklist

- ☐ **Describe defect in detail:**
  - Location – specify level (e.g. cervical, thoracic, thoraco-lumbar, lumbar, lumbosacral, sacral, etc.).
  - Size of lesion.
  - Covering – covered by skin or not covered by skin.
  - Content – only meninges (meningocele) or also spinal cord (myelomeningocele – spinal cord visible).
  - Anomalies – document sequence defects (hydrocephalus, talipes) and other anomalies.
- ☐ **Take and report photographs:** *Show clearly* the level of spina bifida (back and side if possible); can be crucial for review.
- ☐ **Describe evaluations to find or rule out related and associated anomalies:**
  - Sequence – hydrocephalus, talipes, other.
  - Other unrelated anomalies – describe procedures to assess other anomalies.
  - Genetic or chromosomal conditions.
  - Specialty consultations, imaging and surgery.
- ☐ **Report whether autopsy (pathology) findings are available and if so, report the results.**

You now go back to the QRH and review the clinical notes and the checklist for high-quality reporting.

Based on your review, what are some of the other anomalies that you would expect to see more frequently with spina bifida? Why are these important clinically?



Photograph source: © CDC–Beijing Medical University collaborative projec

Checklist for high-quality reporting

Spina Bifida – Documentation Checklist

- ☐ **Describe defect in detail:**
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  - Covering – covered by skin or not covered by skin.
  - Content – only meninges (meningocele) or also spinal cord (myelomeningocele – spinal cord visible).
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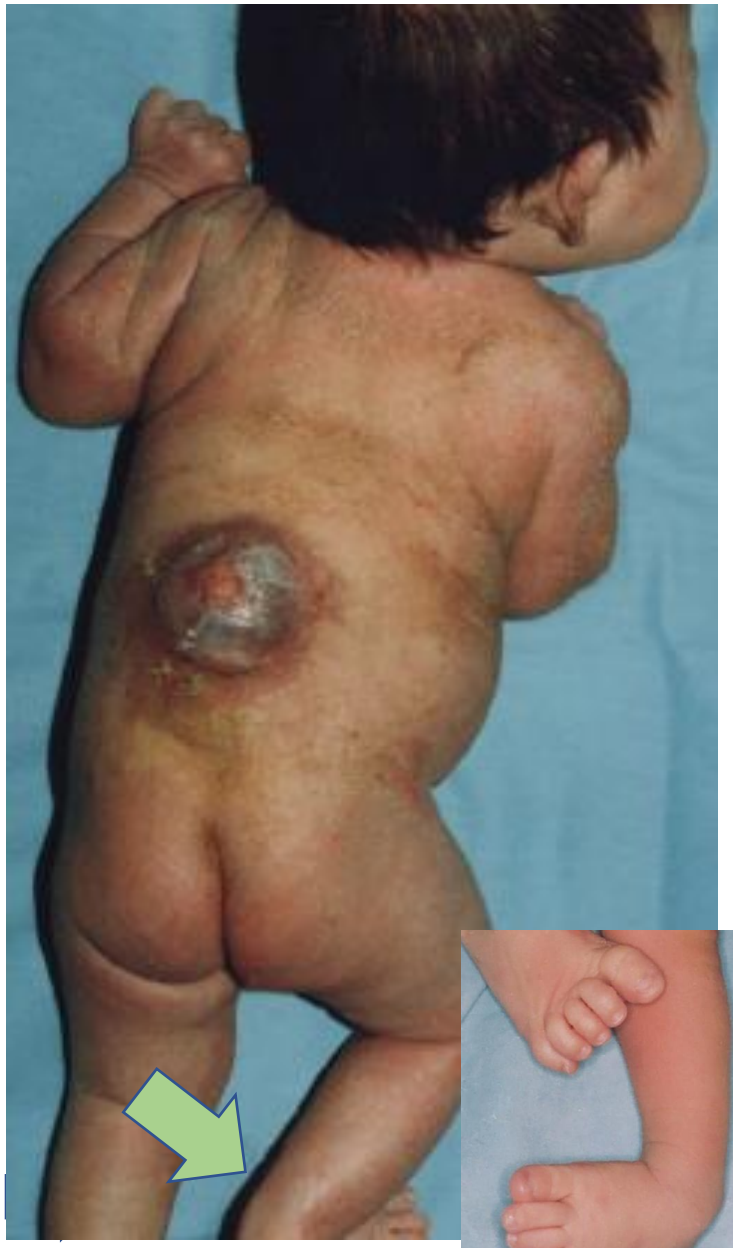
- ☐ **Describe evaluations to find or rule out related and associated anomalies:**
  - Sequence – hydrocephalus, talipes, other.
  - Other unrelated anomalies – describe procedures to assess other anomalies.
  - Genetic or chromosomal conditions.
  - Specialty consultations, imaging and surgery.
- ☐ **Report whether autopsy (pathology) findings are available and if so, report the results.**

Clinical and epidemiologic notes

Spina bifida is often an isolated, non-syndromic (~80%) anomaly. Related findings include:

- Chiari II malformation and hydrocephalus.
- Hip dislocation, talipes, lower limb paralysis.
- Loss of sphincter control, including neurogenic bladder.



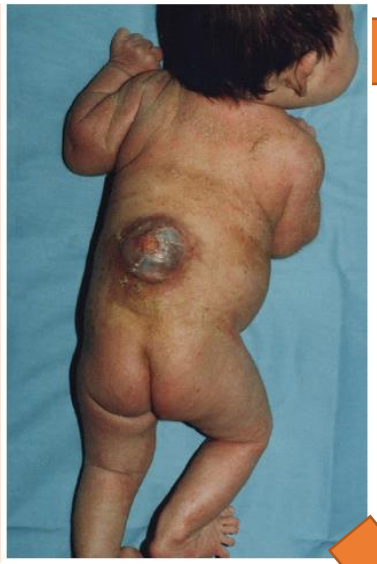
Look for more anomalies, especially the components of the spina bifida SEQUENCE

Photographs sources: © CDC–Beijing Medical University collaborative project; Dr. Idalina Montes and Dr. Rafael Longo

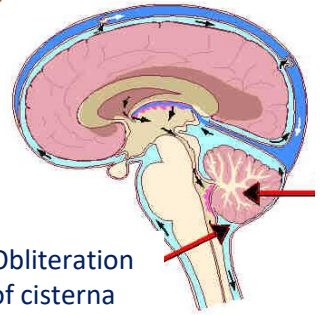
Spina bifida (myelomeningocele) sequence

focus on hydrocephalus, clubfoot, paresis, bladder dysfunction

The brain and spinal cord lesions in spina bifida (myelomeningocele) can generate a cascade or SEQUENCE of structural and functional changes in different parts of the body. It is important to look for these changes, as they are causes of morbidity and disability.



Type II Chiari Malformation



Downward displacement of both cerebellar and brain stem tissue into the foramen magnum

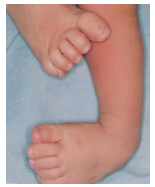
Obliteration of cisterna magna

Obstruction of CSF outflow (may be also due to aqueductal stenosis)

Hydrocephalus

in 90% of lumbo-sacral lesions

Note: only 10% apparent at birth, often develops later



Clubfoot

Hip dislocation

Hip and knee contractures

Spinal cord + nerve damage

Sensory and motor nerve deficits

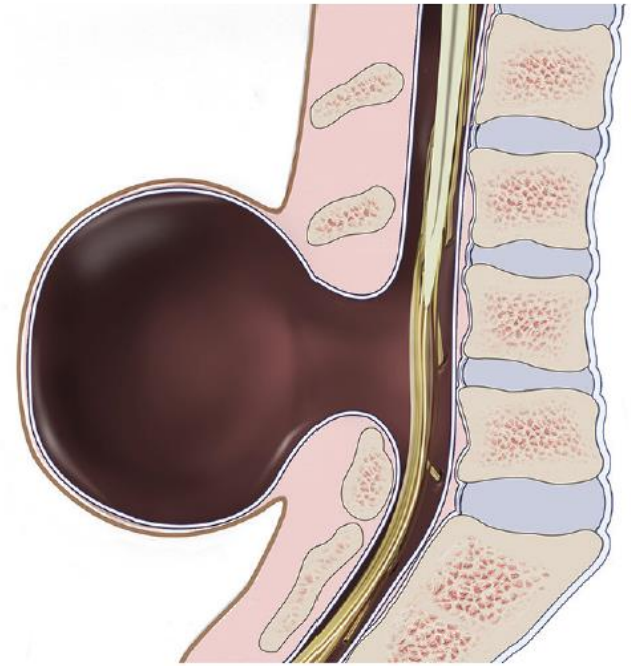
Lower limb paresis

Bladder and bowel dysfunction

Photograph source: © CDC–Beijing Medical University collaborative project

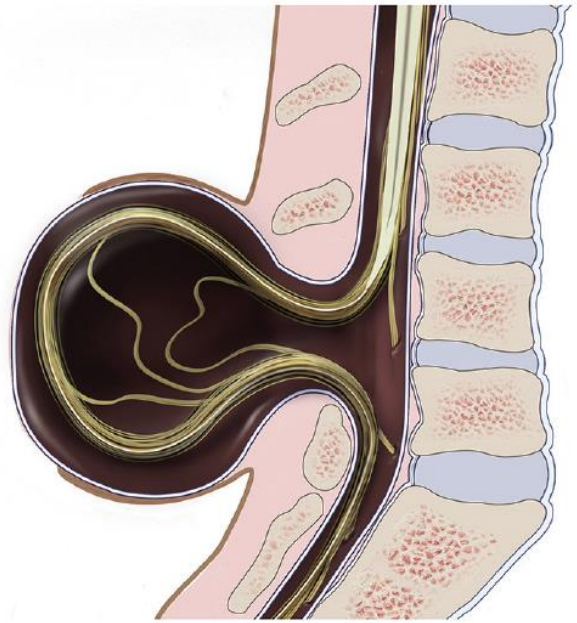
Spina bifida: myelomeningocele vs. meningocele

a. Meningocele (Q05)



The meninges herniate through a spinal defect, forming a cyst filled with cerebrospinal fluid but not containing spinal cord (might have some nerve elements)

b. Myelomeningocele (Q05)



Both meninges and spinal cord involved: typically, more severe clinically than meningocele because the spinal cord is injured (e.g., hydrocephalus and neurogenic deficits are very common with open myelomeningocele)

Coding Spina Bifida

- Spina bifida, thoracic
- Hydrocephalus
- Bilateral talipes equinovarus supinatus (clubfoot)

Relevant ICD-10 codes

Q05.0 Cervical spina bifida with hydrocephalus
Q05.1 Thoracic spina bifida with hydrocephalus
Q05.2 Lumbar spina bifida with hydrocephalus (includes lumbosacral spina bifida with hydrocephalus)
Q05.3 Sacral spina bifida with hydrocephalus
Q05.4 Unspecified spina bifida with hydrocephalus
Q05.5 Cervical spina bifida without hydrocephalus
Q05.6 Thoracic spina bifida without hydrocephalus
Q05.7 Lumbar spina bifida without hydrocephalus (includes lumbosacral spina bifida without hydrocephalus)
Q05.8 Sacral spina bifida with hydrocephalus
Q05.9 Spina bifida, unspecified

Relevant ICD-10 codes

Q66.0 Talipes equinovarus
Q66.8 Other congenital deformities of feet, clubfoot NOS (not otherwise specified)
Q66.1 Talipes calcaneovarus
Q66.4 Talipes calcaneovalgus

Source: © MPM2, Spina bifida, page 52; talipes equinovarus, page 112

Coding Spina Bifida

- Spina bifida, thoracic
- Hydrocephalus
- Bilateral talipes equinovarus supinatus (clubfoot)

Relevant ICD-10 codes

Q05.0	Cervical spina bifida with hydrocephalus
Q05.1	Thoracic spina bifida with hydrocephalus
Q05.2	Lumbar spina bifida with hydrocephalus (includes lumbosacral spina bifida with hydrocephalus)
Q05.3	Sacral spina bifida with hydrocephalus
Q05.4	Unspecified spina bifida with hydrocephalus
Q05.5	Cervical spina bifida without hydrocephalus
Q05.6	Thoracic spina bifida without hydrocephalus
Q05.7	Lumbar spina bifida without hydrocephalus (includes lumbosacral spina bifida without hydrocephalus)
Q05.8	Sacral spina bifida with hydrocephalus
Q05.9	Spina bifida, unspecified

Exclusions

D48	Neoplasm of uncertain behavior (sacrococcygeal teratoma)
Q06.8	Other specified congenital malformations of spinal cord
Q07	Other congenital malformations of nervous system
Q76.0	Spina bifida occulta

Source: © MPM2, Spina bifida, page 52

Part 2: Deeper dive into the clinical presentation

Adaeze is in her second day of life and is being treated for spina bifida**.** You now notice a few more findings:

- She is rather **small** for being a full-term baby (just over 2.2 kg).
- She has **delicate features**, with a jaw that appears small and a sloping forehead.
- She keeps her **fingers clenched** in **an unusual position**, with the 2nd finger overlapping the 3rd and the 5th overlapping the 4th.

You go back to the QRH and you notice the following clinical note:

Clinical and epidemiologic notes

Spina bifida is often an isolated, non-syndromic (~80%) anomaly. Related findings include:

- Chiari II malformation and hydrocephalus.
- Hip dislocation, talipes, lower limb paralysis.
- Loss of sphincter control, including neurogenic bladder.

Additional clinical tips:

- Always look for additional anomalies and syndromes (trisomy 18).
- Review examinations, procedures and imaging – rare conditions misdiagnosed as spina bifida include spina bifida occulta, sacrococcygeal teratoma, isolated scoliosis/kyphosis, and amniotic band syndrome.
- Lipomeningo(myelo)cele is a rare type of spina bifida with an overlying lipoma; many programmes do not include lipomeningo(myelo)celes as an NTD.

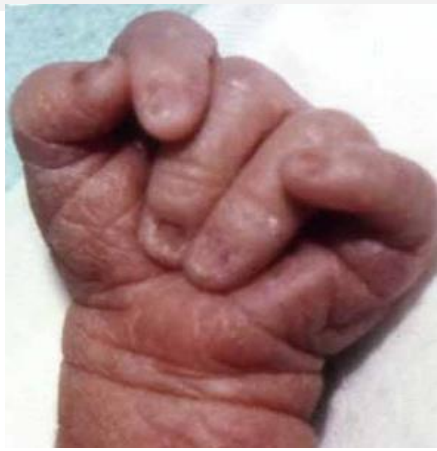


Photo source: © https://medlineplus.gov/images/PX0002AU_PRESENTATION.jpeg

Q7. What does this presentation suggest to you? Why might it be important?

Learn to recognize trisomy 18

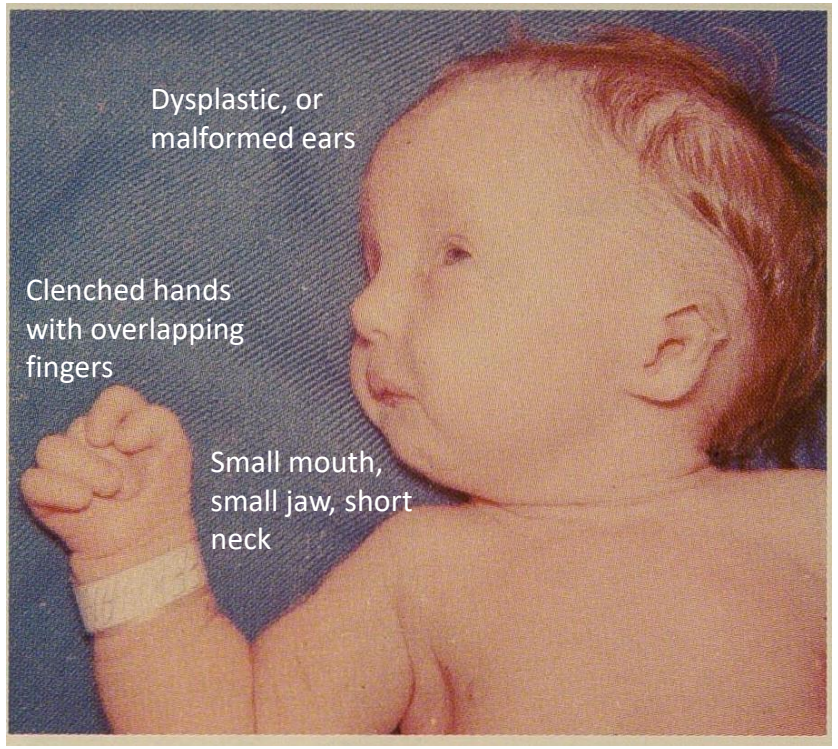
a syndrome that can be associated with myelomeningocele

Trisomy 18 syndrome (Edwards syndrome)

- Recognizable pattern of minor (see list) and major (see table) anomalies, with prenatal growth deficiency
- Due to presence of an extra chromosome 18 or, less commonly, to a partial trisomy of the long arm of chromosome 18; mosaic cases are rare

Organ system	Frequency	More common malformation
Heart	> 75%	Septal defects, PDA
Genitourinary	25–75%	Horseshoe kidney
Gastrointestinal	< 25%	Omphalocele
Craniofacial		Oral cleft
Eye		Corneal opacity
Limbs		Radial aplasia

- **Diagnosis** can be suspected clinically and is confirmed by chromosome studies
- **Overall prevalence** 4.0 per 10 000 pregnancies (Cereda A and Carey JC, 2012)
- Risk increases with maternal age

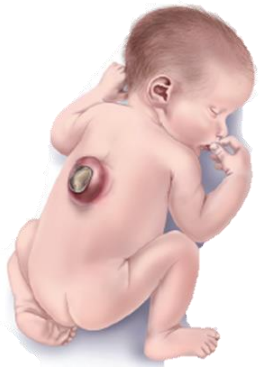


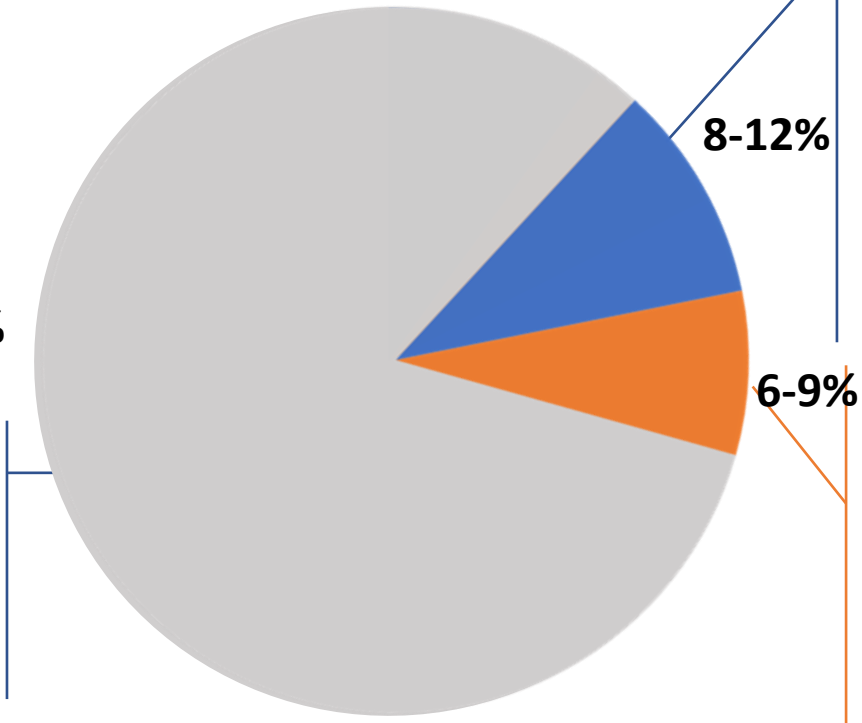
Short, prominent sternum; wide-set nipples

Source: Sydney S. Gellis and Murray Feingold - Atlas of mental retardation syndromes; visual diagnosis of facies and physical findings, Public Domain

Source: Cereda A, Carey JC. The trisomy 18 syndrome. Orphanet J Rare Dis. 2012 Oct 23;7:81. doi: 10.1186/1750-1172-7-81..

Myelomeningocele: clinical classification





Isolated
Include cases with related anomalies and sequences

Multiple congenital anomalies (MCA)

- Heart defects (VSD), kidney defects, oral clefts, genital anomalies, diaphragmatic hernia, omphalocele
- Note that true spina bifida is *not* a component of OEIS complex (omphalocele, exstrophy of the cloaca, imperforate anus, and spinal defects), just misdiagnosed as spina bifida

Syndromes

Chromosomal

- **Trisomy 18 (more frequent)**
- Trisomy 13
- Trisomy 9 mosaic
- Triploidy

Single gene (few)

- Cerebro-costo-mandibular syndrome

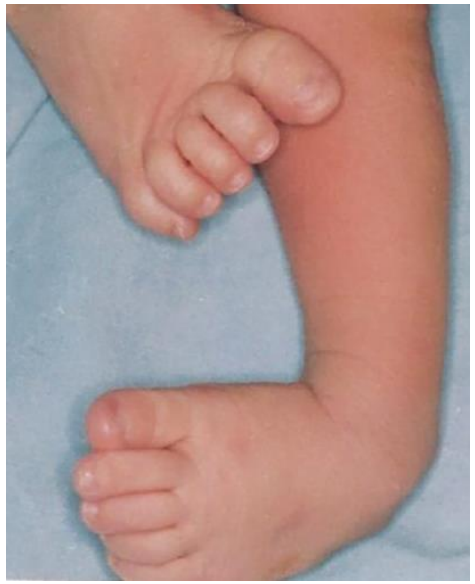
Teratogenic

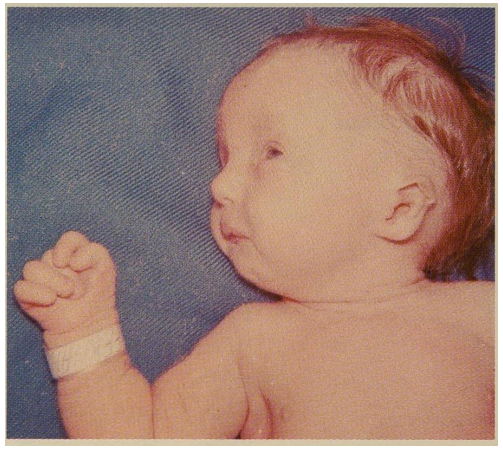
- Fetal valproate syndrome, diabetic embryopathy

Note: Percentages are approximate, compiled from different studies; definitions and methodology often vary in the studies.

Part 3: Putting everything together and review of key points

What is your final description, diagnosis, and coding?



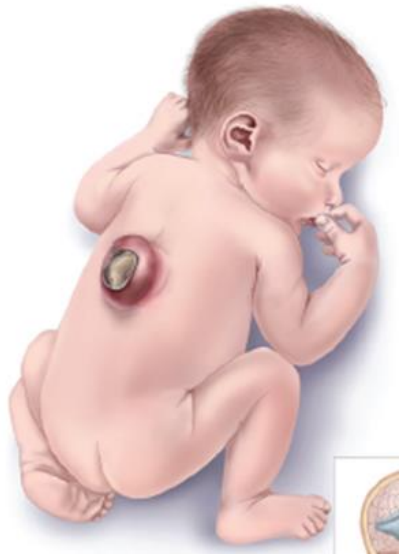


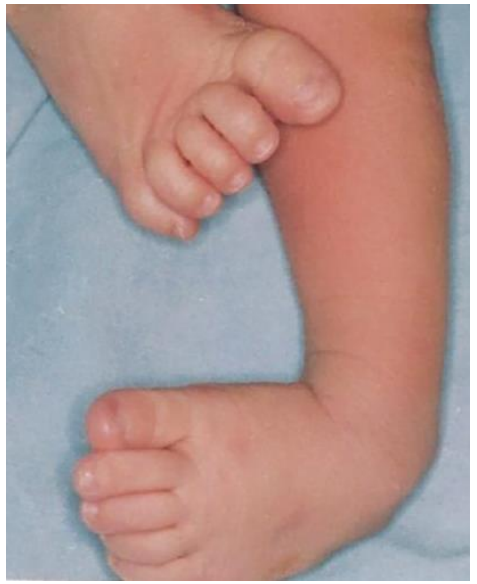
Photograph sources: Dr. Idalina Montes and Dr. Rafael Longo;
Sydney S. Gellis and Murray Feingold - Atlas of mental retardation syndromes; visual diagnosis of facies and physical findings, Public Domain

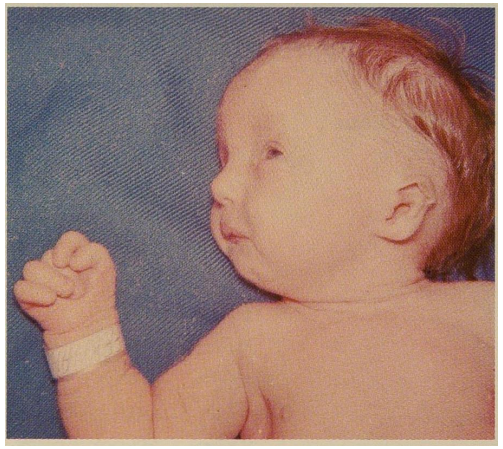
What is your final description, diagnosis, and coding?

- Spina bifida, thoracic, open (not skin covered), about 5 cm in diameter, with hydrocephalus
  - code: Q05.11
- Bilateral talipes equinovarus
  - code: Q66.0
- Trisomy 18, or Edwards syndrome
  - code: Q91.3

The overall clinical classification is ‘syndrome’







Photograph sources: Dr. Idalina Montes and Dr. Rafael Longo;
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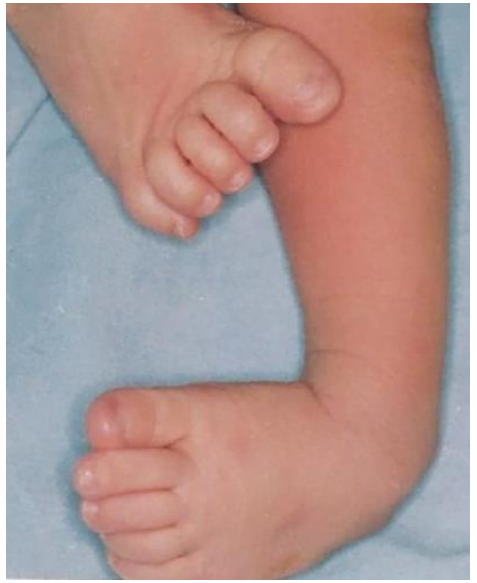
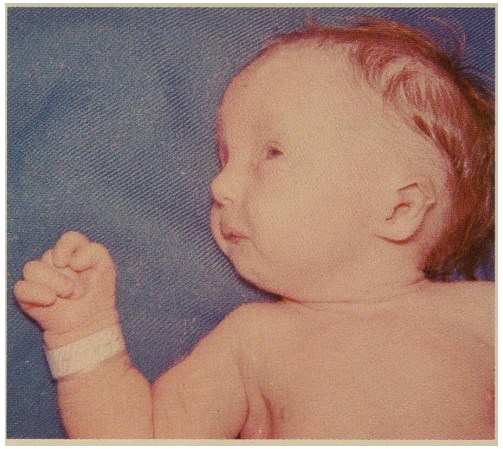
Some lessons learned from Adaeze's case: discuss

- Diagnosable at the initial newborn exam
- Describe location, skin covering, size
- Examine head to toe, front and back: check carefully for spina bifida sequence (e.g., hydrocephalus, clubfoot)
- Spina bifida usually an isolated anomaly but consider the possibility of additional anomalies and syndromes: precise and complete diagnosis helps improve care, counselling, and public health surveillance





Photograph sources: Dr. Idalina Montes and Dr. Rafael Longo;
Sydney S. Gellis and Murray Feingold - Atlas of mental retardation syndromes; visual diagnosis of facies and physical findings, Public Domain

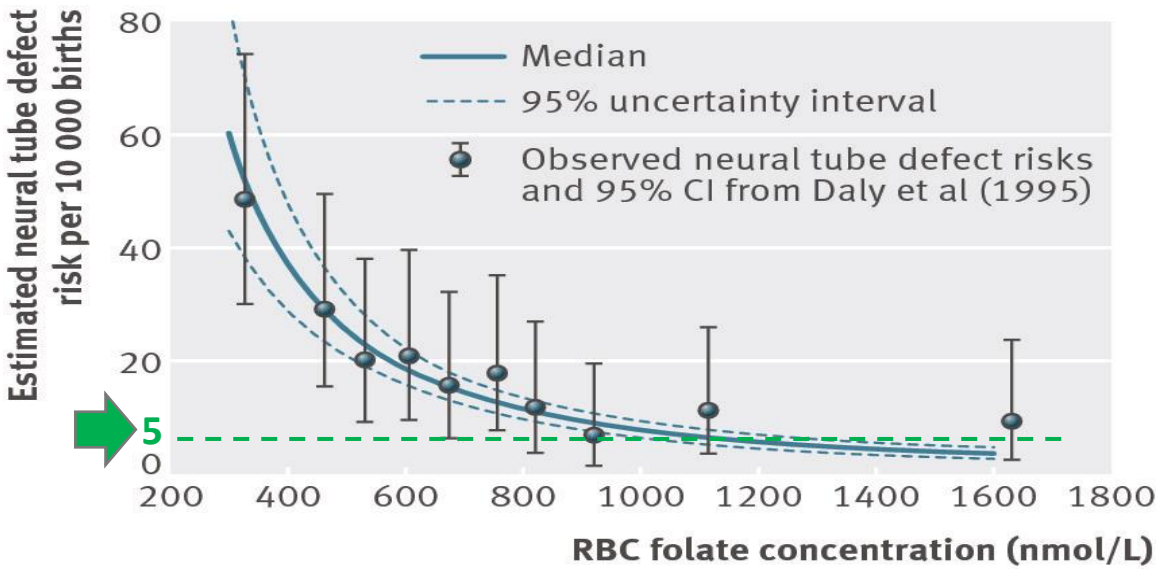
Part 4: Elements for public health prevention

Folic acid insufficiency and other selected risk factors

Folic acid and spina bifida: an opportunity for primary prevention not to be missed

Insufficient blood folate concentration at pregnancy outset is the most important modifiable risk factor for neural tube defects (NTDs)

The higher the folate concentration, the lower the rate of NTDs (down to ~ 5/10 000 or perhaps fewer)



- **Folate sufficiency** (RBC folate ≈ 1000 nmol/L) minimizes the risk for neural tube defects and should be achieved **before pregnancy**
- Better folate levels can be achieved through **diet, supplementation (pills), and fortified foods**
- Practically, from a **population perspective, food fortification is a proven strategy** ensuring widespread folate sufficiency for NTD prevention

Table source: Crider KS, Devine O, Hao L, Dowling NF, Li S, Molloy AM et al. Population red blood cell folate concentrations for prevention of neural tube defects: Bayesian model. BMJ. 2014 Jul 29;349:g4554. doi: 10.1136/bmj.g4554.

Some other established modifiable risk factors for neural tube defects, including spina bifida

Antiepileptic medications (monotherapy)

can also increase risk for other malformations

Occurrence (absolute risk) **of spina bifida**
estimated from 21 prospective studies combined

- **Valproic acid (dose dependent effect): 1.84%**
- Carbamazepine: 0.32%
- Barbiturates: 0.26%
- Lamotrigine: 0.12%
- Baseline reference risk (rate): approximately 0.05%

Source: Tomson T, Battino D. Teratogenic effects of antiepileptic drugs. Lancet Neurol. 2012 Sep;11(9):803-13. doi: 10.1016/S1474-4422(12)70103-5. Epub 2012 Jul 16.

Pregestational diabetes

increases risk for NTDs and several other malformations

- **Risk for NTDs** (anencephaly, spina bifida, encephalocele) with diabetes is increased up to fivefold
- **Risk is directly related to HbA1c** level at beginning of pregnancy

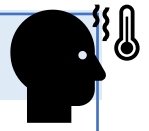
Obesity

Estimated relative risk from meta-analysis

- Anencephaly OR: 1.4 (95% CI: 1.0-1.9)
- Spina bifida OR: 2.2 (95% CI: 1.9-2.7)

Source: Stothard KJ, Tennant PW, Bell R, Rankin J. Maternal overweight and obesity and the risk of congenital anomalies: a systematic review and meta-analysis. JAMA. 2009 Feb 11;301(6):636-50. doi: 10.1001/jama.2009.113.

High fever



Estimated relative risk from meta-analysis

- Neural tube defects – OR: 2.9 (95% CI: 2.2-3.8)

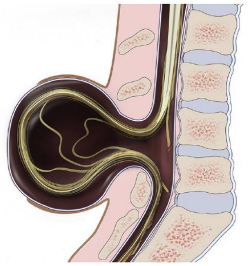
Source: Dreier JW, Andersen AM, Berg-Beckhoff G. Systematic review and meta-analyses: fever in pregnancy and health impacts in the offspring. Pediatrics. 2014 Mar;133(3):e674-88. DOI: 10.1542/peds.2013-3205.

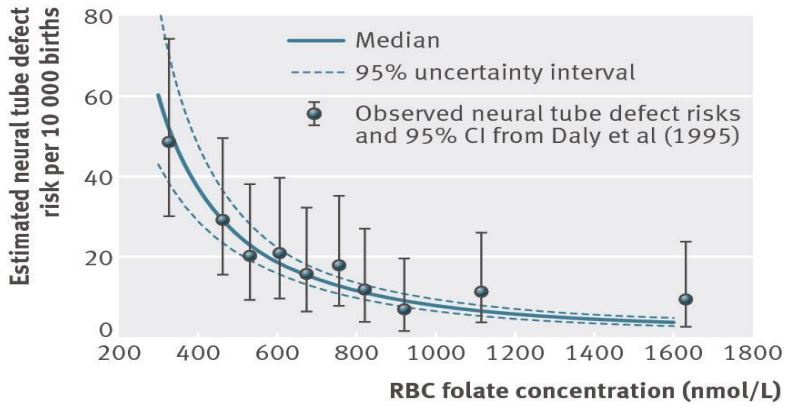
Adaeze's story: key points, lessons learned and comments….

b. Myelomeningocele (Q05)

This case study covers a lot of ground.

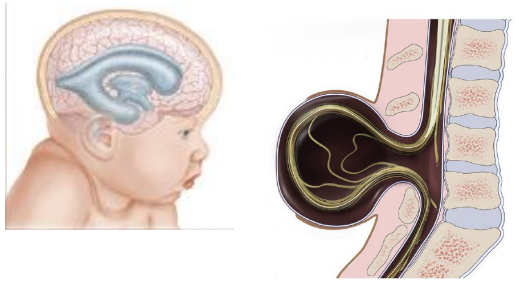
- How would you summarize and perhaps expand on the key points and lessons learned?
- Which points or issues are particularly relevant to your local setting?
- What types of data/information are available or missing in your setting to assess the key issues related to spina bifida?

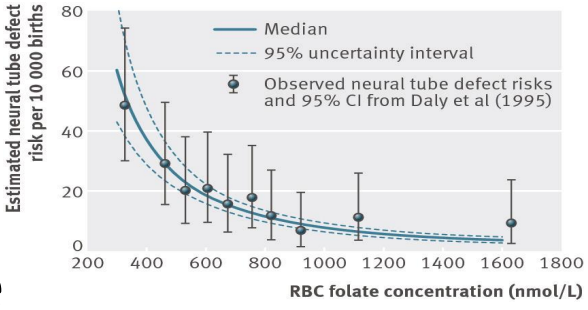




Adaeze's story: key points, lessons learned and comments….

- **Head-to-toe exam**: every time, every baby
- **Checklists**: use consistently
- **Spina bifida sequence**: use this knowledge to improve your exam and description
- **Multiples and syndromes**: can occur, and modify outcomes, treatment, and public health surveillance
- **Folic acid**: a precious opportunity for primary prevention
- **Making folic acid work**: supplementation, but especially fortification, are safe and effective
- **Data for action:** accurate, relevant and timely data that serve clinical care and population health (and prevention)





Thank you!
Questions?

Evaluation

1. When describing a case of spina bifida, **which elements of the birth defect itself should be reported, at a minimum, in the verbatim description** according to the QRH check list**?**

Choose <u>one or more</u> answers

a. Location (level of lesion, such as thoracic, lumbar, sacral, etc.)

b. Covering (whether it is skin covered or not, that is, open or closed)

c. Movement of lower limbs (presence or not of paralysis)

d. Size of lesion (approximate size in cm)

e. Pulsation of anterior fontanel (yes or no)

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d. **Size of lesion (approximate size in cm)**

e. Pulsation of anterior fontanel (yes or no)

2. You see a newborn baby with an open thoracic spina bifida, about 5 cm in diameter.

What related anomalies are commonly seen with this presentation?

Choose one or more answers

a. Polydactyly

b. Hydrocephalus

c. Renal anomalies

d. Clubfoot

e. Cleft palate

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c. Renal anomalies

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e. Cleft palate

3. You review the records of a baby born with spina bifida. You are trying to understand whether this is meningocele or myelomeningocele.

Match these conditions with the description of their contents

Choose one or more answers

Condition

a. Meningocele

b. Myelomeningocele

Description of contents

1. Sac containing meninges and cerebrospinal fluid, no spinal cord/nerves
2. Sac containing meninges, cerebrospinal fluid, and spinal cord/nerves
3. Sac containing cerebrospinal fluid, hair/teeth (more or less developed)

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Match these conditions with the description of their contents

Choose <u>one or more</u> answers

Condition	**Description of contents**
a. Meningocele	1. Sac containing meninges and cerebrospinal fluid, no spinal cord/nerves
b. Myelomeningocele	2. Sac containing meninges, cerebrospinal fluid, and spinal cord/nerves
	3. Sac containing cerebrospinal fluid, hair/teeth (more or less developed)

4. In reviewing hospital records for surveillance, you identify a baby with spina bifida (thoracolumbar, open), bilateral clubfoot (talipes equinovarus supinatus), Chiari 2 malformation, and hydrocephalus.

What is the most appropriate terminology to describe this combination of anomalies?

Choose the <u>one</u> best answer

a. Spina bifida association

b. Spina bifida sequence

c. Spina bifida syndrome

d. Spina bifida combination

4. In reviewing hospital records for surveillance, you identify a baby with spina bifida (thoracolumbar, open), bilateral clubfoot (talipes equinovarus supinatus), Chiari 2 malformation, and hydrocephalus.

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(Choose the <u>one</u> best answer)

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b. Spina bifida sequence

c. Spina bifida syndrome

d. Spina bifida combination

5. You review the history of a 25-year-old woman who recently had a baby with thoracic spina bifida. What are some factors that increase the risk of having a pregnancy affected by spina bifida?

Choose <u>one or more</u> answers

a. Maternal folic acid insufficiency (low level of blood folate)

b. Maternal diabetes mellitus, not well controlled

c. Rubella infection during pregnancy

d. Fetus affected by trisomy 18

e. Mother with epilepsy on valproic acid during the pregnancy

5. You review the history of a 25-year-old woman who recently had a baby with thoracic spina bifida. What are some factors that increase the risk of having a pregnancy affected by spina bifida?

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b. Maternal diabetes mellitus, not well controlled

c. Rubella infection during pregnancy

d. Fetus affected by trisomy 18

e. Mother with epilepsy on valproic acid during the pregnancy

Clinical scenario 2: Baby Esther

Cleft palate, tetralogy of Fallot, deletion 22q11

Epidemiology and risk factors for orofacial clefts

World Health Organization

CDC

International Clearinghouse for Birth Defects Surveillance and Research

Part 1: Clinical scenario

Clinical scenario – Baby Esther

- Baby girl Esther is born at term in apparent good health. She is now a few hours old and in the well-baby nursery.
- The plan is to discharge the baby home with her mother within 24–48 hours as soon as the neonatal checks are completed and the baby is feeding well.
- A first neonatal exam appears normal. Her mother has now started to breastfeed her.

Part 1. You are called to the crib by the mother, who tells you that she is a bit concerned for Esther: when she tries to breastfeed Esther, *she sees milk coming out of her nose*.

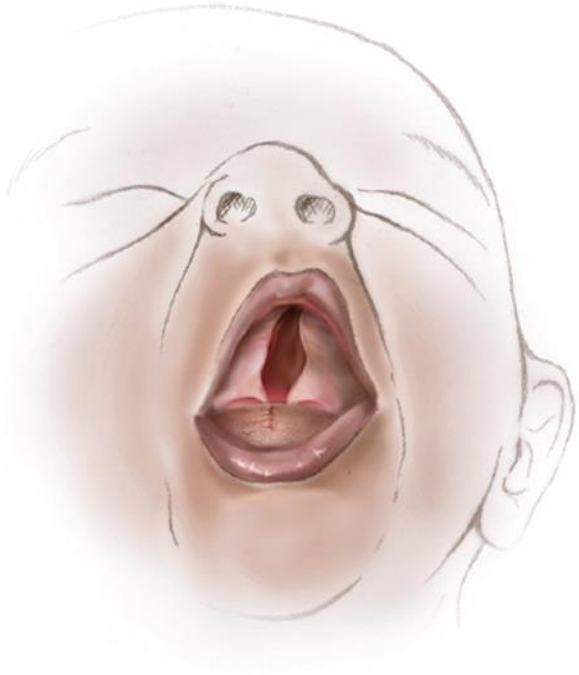
- You now re-examine the baby carefully and efficiently, from head to toe.

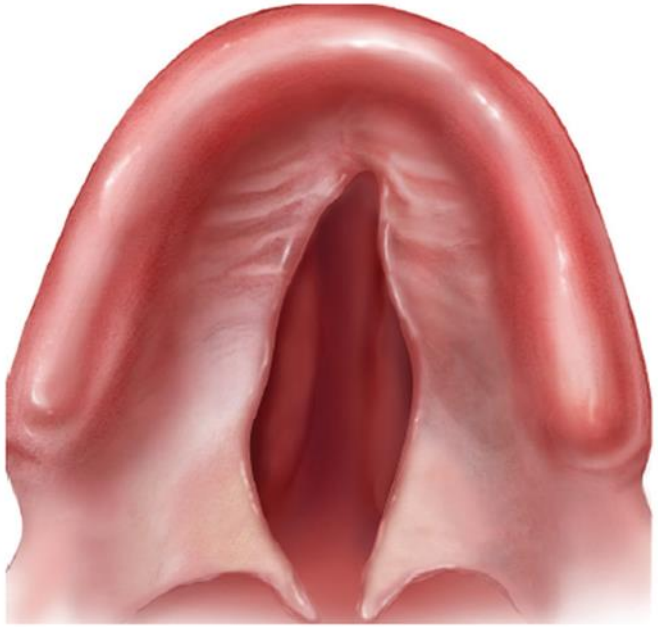
Q1. Based on the clinical history, what specifically should you look out for in your exam?

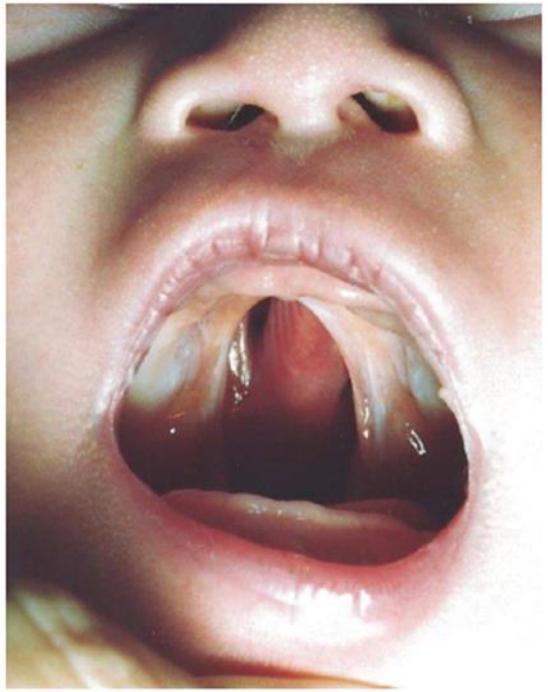
Photo source: stock image

Q2. What do you do next?

- **A2**. You open the mouth and examine the palate carefully, using a light and a tongue depressor. It takes a bit of time, but with patience and some help from a colleague, you are able to visualize the palate all the way to the back of the mouth.
- **This is what you see**

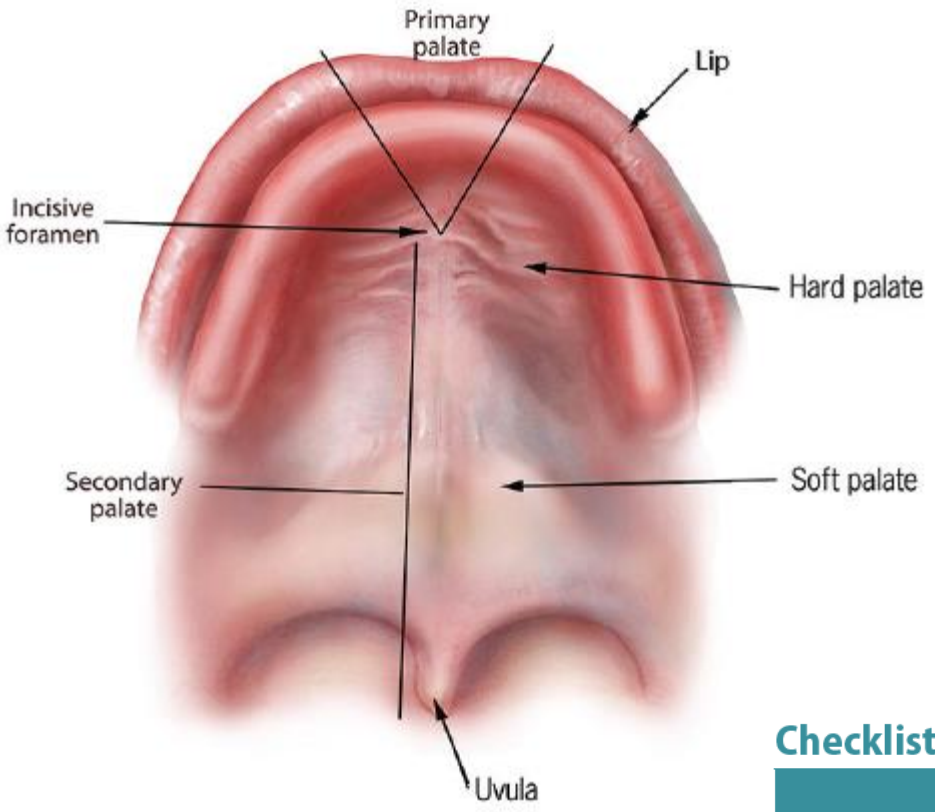


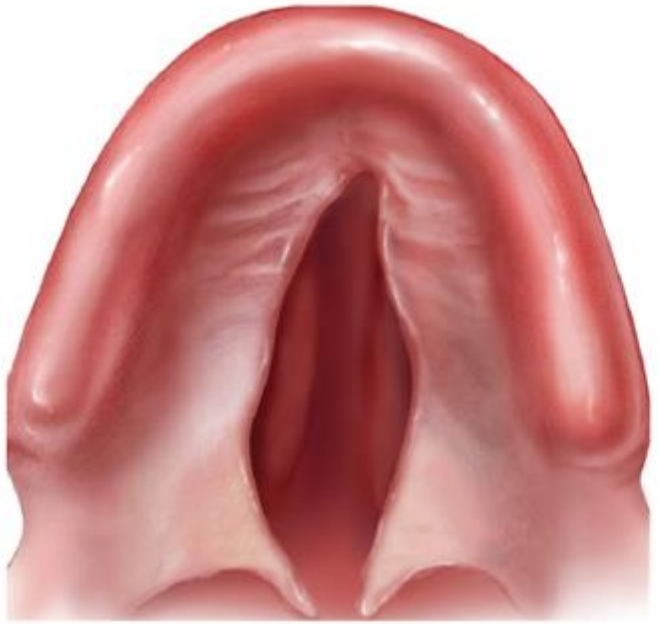


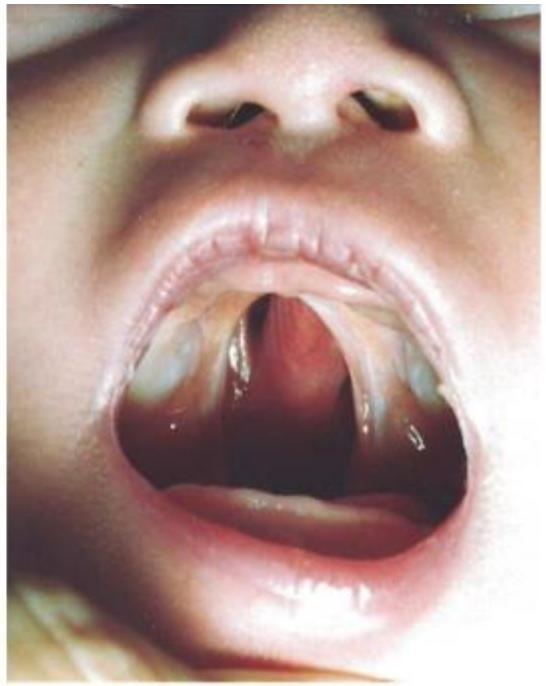


Photograph source: © CDC–Beijing Medical University collaborative project

Q3. Using the figures and the description checklist in the QRH (pages 37 and following) to describe the elements of the palate, and then the cleft palate.







Photograph source: © CDC–Beijing Medical University collaborative project

Anatomy

Checklist

Checklist for high-quality reporting

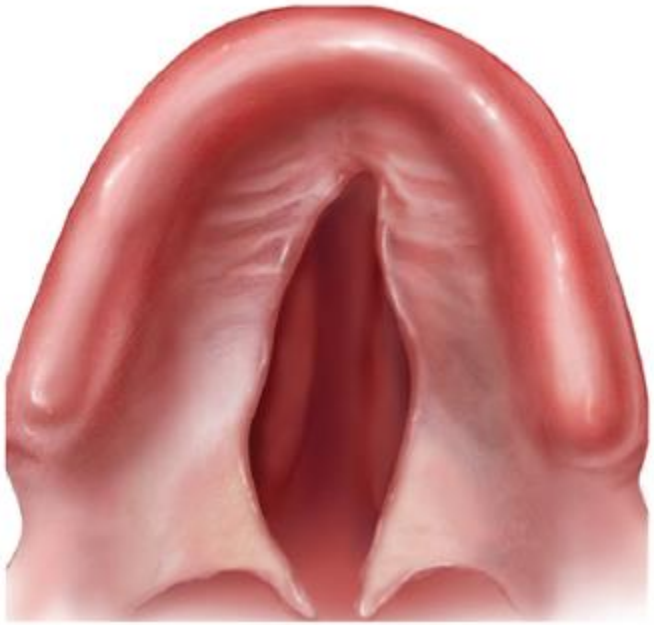
Cleft Palate – Documentation Checklist

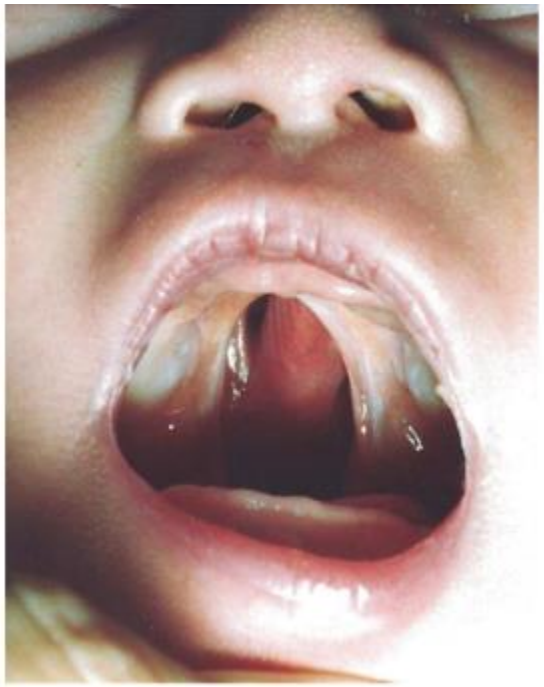
- ☐ **Describe in detail**, including:
  - Extension (cleft palate) – hard palate, soft palate.
  - Lower lip (see Fig. 23) – pits present or absent (when present, the van der Woude syndrome should be strongly suspected).
  - Presence of components of the Pierre Robin sequence – microretrognathia (small recessed jaw), glossoptosis (posterior displacement of the tongue) and respiratory obstruction.
- ☐ **Describe procedures to assess further additional malformations and if present, describe.**
- ☐ **Take and report photographs:** Very useful; can be crucial for review.
- ☐ **Report whether specialty consultation(s) were done and if so, report the results.** Plastic surgery and genetics consultation reports are useful.

Q3. Using the figures and the description checklist in the QRH (pages 37 and following) describe the elements of the palate, and then the cleft palate.

A3. Basic description of the cleft palate:

- The defect involves the secondary palate, from the foramen incisivum in front to the back of the palate, where the uvula should be
- The cleft palate is complete in that it involves both the bony hard palate (the anterior portion of the palate) and the soft palate
- The uvula is absent
- There are no lip pits

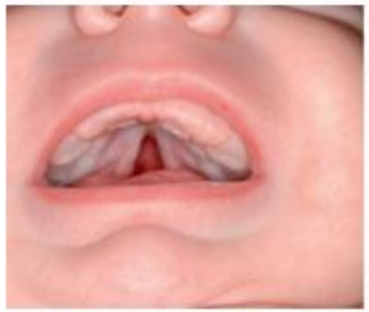




Photograph source: © CDC–Beijing Medical University collaborative project

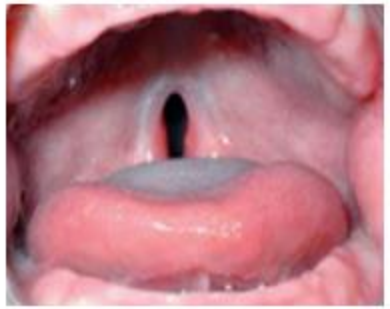
Cleft palate only (CPO): subtypes

Major Anomalies

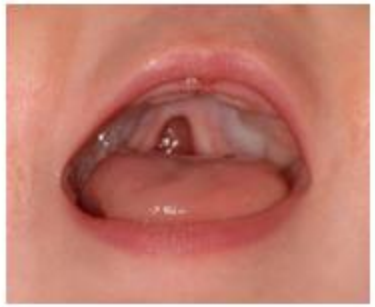


Cleft soft and hard palate
V-shaped

Cleft soft and hard palate
U-shaped



Cleft soft palate
V-shaped



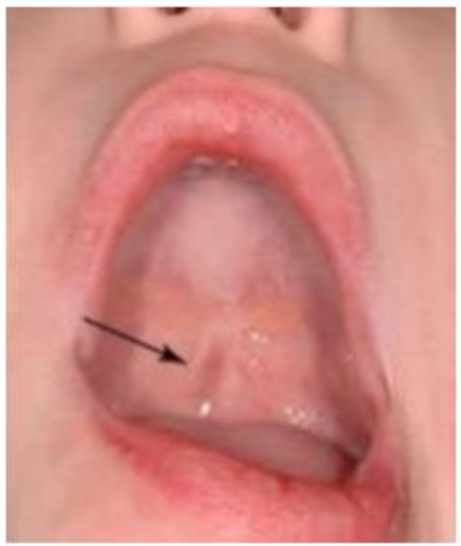
Cleft soft palate
U-shaped

http://www.slideshare.net/indiandentalacademy/syndromes-affecting-the-palate-dental-implant-courses
https://www.childrensmn.org/services/care-specialties-departments/ear-nose-throat-ent-facial-plastic-surgery/conditions-and-services/cleft-palate/

Submucous Cleft Palate

Often a late diagnosis

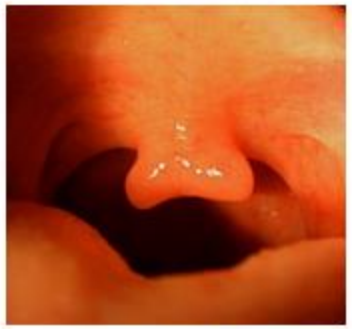
May be an eligible anomaly in programs with prolonged ascertainment period (years)



Submucous cleft palate

http://www.slideshare.net/joelslides/adenoidectomy-and-tonsillectomy

Minor anomaly



Cleft uvula

http://www.wikiwand.com/en/Palatine_uvula

High reported prevalence of cleft uvula in school children :

- Florida (US): 2.3%
- Navajos (US): 11%

Wharton P, Mowrer DE. 1992

Part 2: Deeper dive into the clinical presentation

Part 2. The mother also tells you that when she tries putting the baby down on her back to rest, Esther seems to struggle to breathe, but breathes better when placed on her belly, head to one side.

Q4. What could this be? Could it be related to the cleft palate? You check the QRH and review the description, clinical tips and checklist.

What might you check for now that could have been missed before? What do you do next?

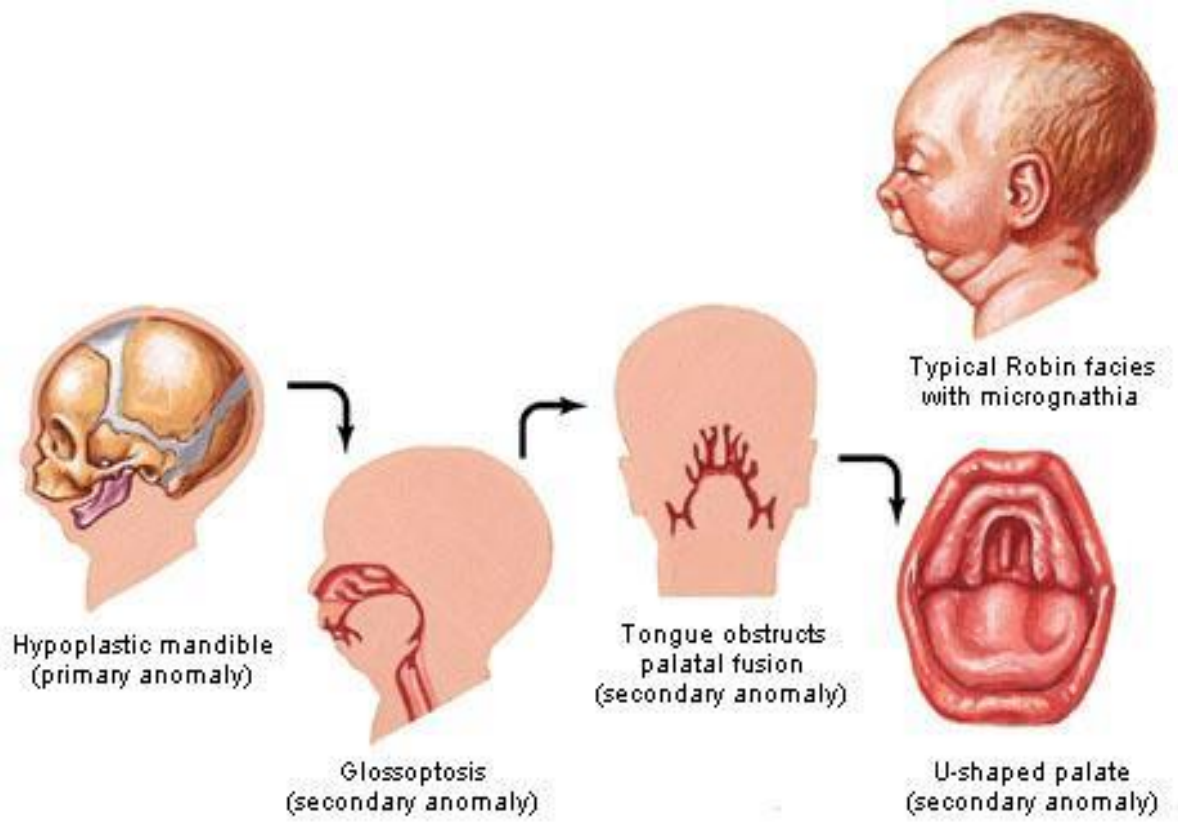
Clinical and epidemiologic notes

In cleft palate, a complete evaluation and physical examination is crucial as it is more commonly associated with additional anomalies and syndromes compared to other types of clefts (e.g. cleft lip).

Additional clinical tips:

- Check for lip pits in the lower lip (see Fig. 23), in the child ***and*** in the parents – it is a sign of a genetic condition (van der Woude syndrome) with high recurrence risk (a parent may have the pits but not the cleft).
- Check for additional anomalies, especially of the heart (e.g. in deletion 22q11 syndrome) and eye (e.g. Stickler syndrome).
- Check for components of the Pierre Robin sequence, including microretrognathia (small recessed jaw), glossoptosis (posterior displacement of the tongue) and respiratory obstruction.

Pierre Robin sequence



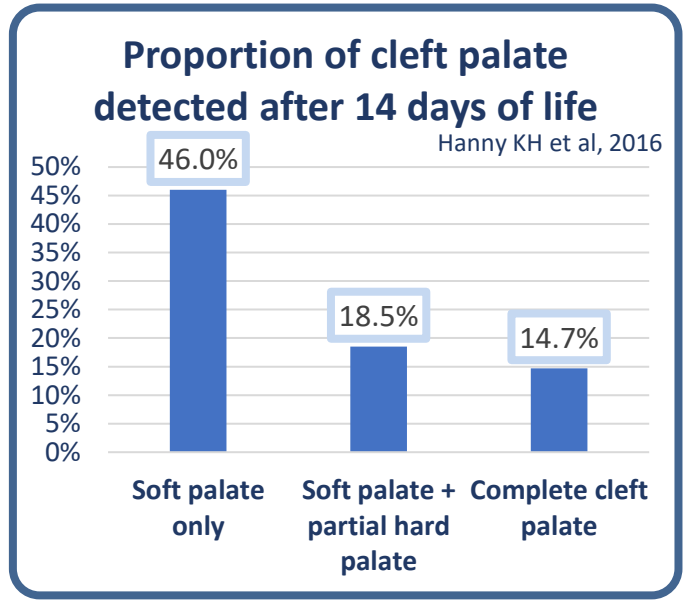
- Esther has cleft palate as part of the Pierre Robin sequence
- She has a cleft palate, but also her **jaw is small** (micrognathia), and the tongue falls back when placed on her back (glossoptosis), obstructing the airway and causing difficulty breathing (respiratory distress)
- Placing her prone moves the tongue forward and makes breathing easier

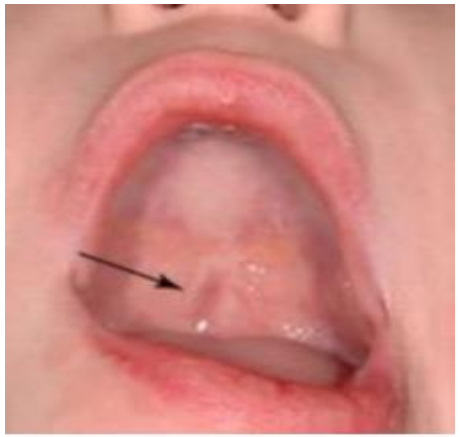
Esther has all three components of the Pierre Robin sequence.

Note: micrognathia and glossoptosis are the key components – a cleft palate is often present but *not required* for the sequence .
Clinical tip: Pierre Robin sequence can be a **clinical urgency**, so always look for it in a baby with a small jaw and/or cleft palate

Tips on neonatal diagnosis of CPO

- **Late diagnosis of CPO (after 14 days) is not uncommon,** particularly when only the soft palate is involved
- Visualizing the palate completely in a newborn is not easy (the mouth can be small and challenging to open wide); s**uggest systematically checking the palate in every newborn by feeling the inside of the mouth with your gloved finger**
- Submucous cleft palate: often missed for years
  - **The diagnosis of submucous cleft palate rests on the triad of** (a) **notching** of the posterior border of the hard palate, (b) **muscular diastasis** with mucosal integrity, and (c) **bifid uvula.** Note: these signs are not always all present.
  - **The diagnosis during the neonatal period or first year of life is very difficult.** For this reason, this condition is typically not included in surveillance programmes unless the ascertainment window extends for several years (e.g., 6–7 years).





http://www.slideshare.net/joelslides/adenoidectomy-and-tonsillectomy

Submucous cleft palate

Coding cleft palate

(from the MPM2)

Relevant ICD-10 codes

Suggest coding Q35.5 plus Q87.0

Q35.1	Cleft hard palate
Q35.3	Cleft soft palate
Q35.5	Cleft hard palate with cleft soft palate
Q35.59	Complete cleft palate
Q35.9	Cleft palate, unspecified
Q35	Cleft palate: Avoid using this generic code if more specific information is available

Related ICD-10 code

Q87.0 Robin sequence or defect, with core components including retro-micrognathia, posterior displacement of the tongue (glossoptosis) and respiratory obstruction. Cleft palate is a common though not required component of the Robin sequence. Whereas ICD-10 lists Robin sequence as an exclusion from the Q35 series, because it is such a common condition, the Q87.0 code is suggested.

Notes:

- Q35 is the generic code for cleft palate. However, avoid using this code if more specific information is available.
- A simple alternative approach to classification and coding is the LAHSAL method, used often by plastic surgeons to assess for treatment outcomes (see Fig. 4.23). In surveillance it is best used in addition to, rather than instead of, the ICD codes.

Coding cleft palate: note the exclusions

(from the MPM2)

Relevant ICD-10 codes

Q35.1 Cleft hard palate
Q35.3 Cleft soft palate
Q35.5 Cleft hard palate with cleft soft palate
Q35.59 Complete cleft palate
Q35.9 Cleft palate, unspecified
Q35 Cleft palate: Avoid using this generic code if more specific information is available

Related ICD-10 code

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Notes:

- Q35 is the generic code for cleft palate. However, avoid using this code if more specific information is available.
- A simple alternative approach to classification and coding is the LAHSAL method, used often by plastic surgeons to assess for treatment outcomes (see Fig. 4.23). In surveillance it is best used in addition to, rather than instead of, the ICD codes.

Inclusions

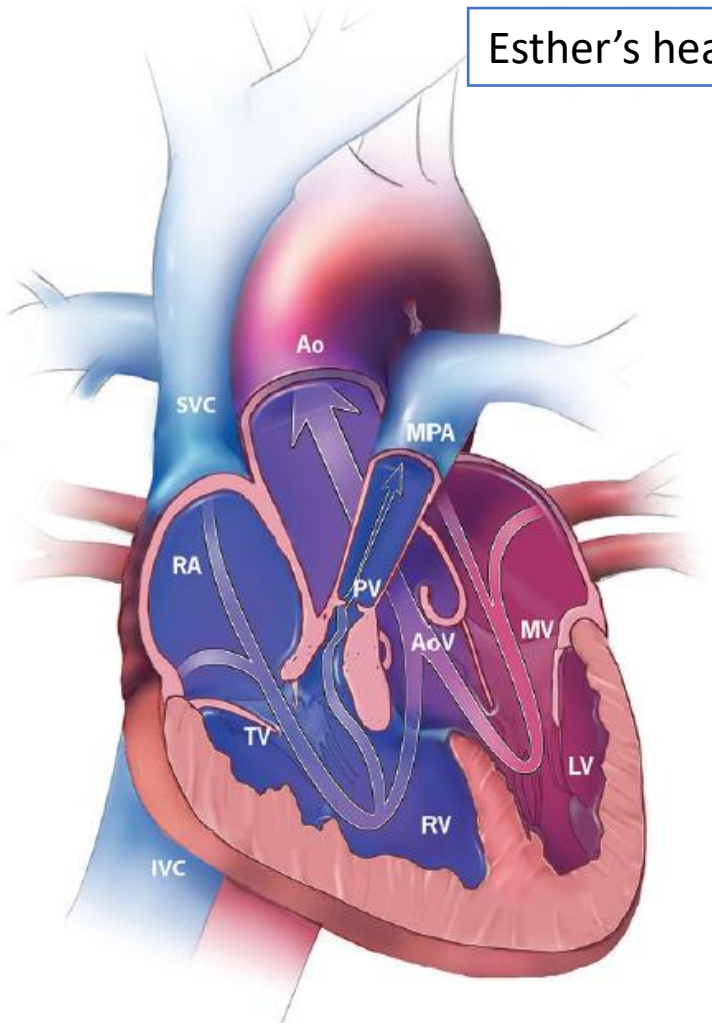
Q35 and subgroups except for Q35.7 (bifid uvula): Cleft palate (excluding bifid uvula and submucous cleft palate)

Exclusions

Q37 Cleft palate with cleft lip
Q35.7 Bifid (cleft) uvula
Q38.5 Absence of uvula

Submucous cleft palate: This defect is not a considered a major anomaly and should not be included in prevalence counts of cleft palate. Submucous cleft palate does not have a specific code.

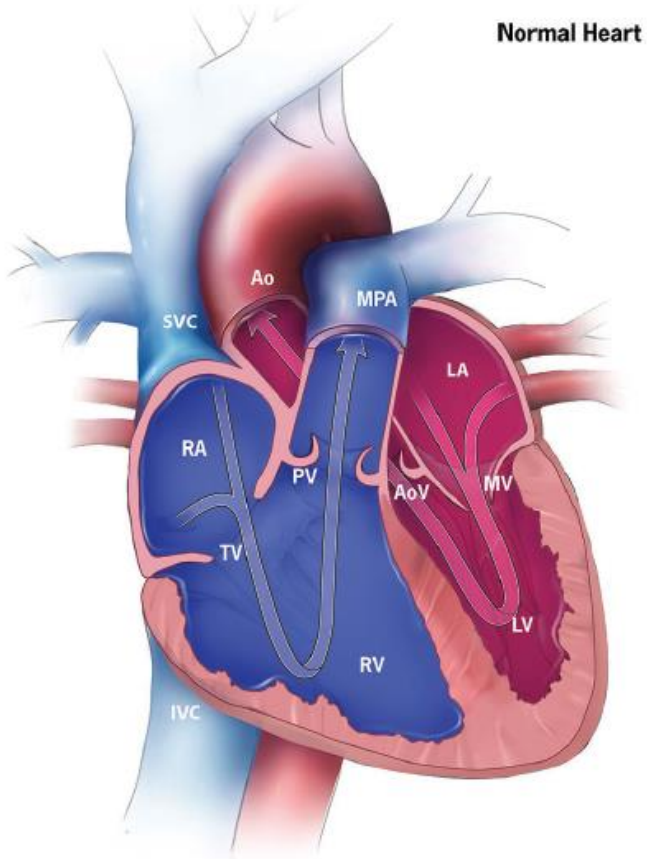
Esther's heart



RA. Right Atrium
RV. Right Ventricle
LA. Left Atrium
LV. Left Ventricle

SVC. Superior Vena Cava
IVC. Inferior Vena Cava
MPA. Main Pulmonary Artery
Ao. Aorta

TV. Tricuspid Valve
MV. Mitral Valve
PV. Pulmonary Valve
AoV. Aortic Valve



RA. Right Atrium
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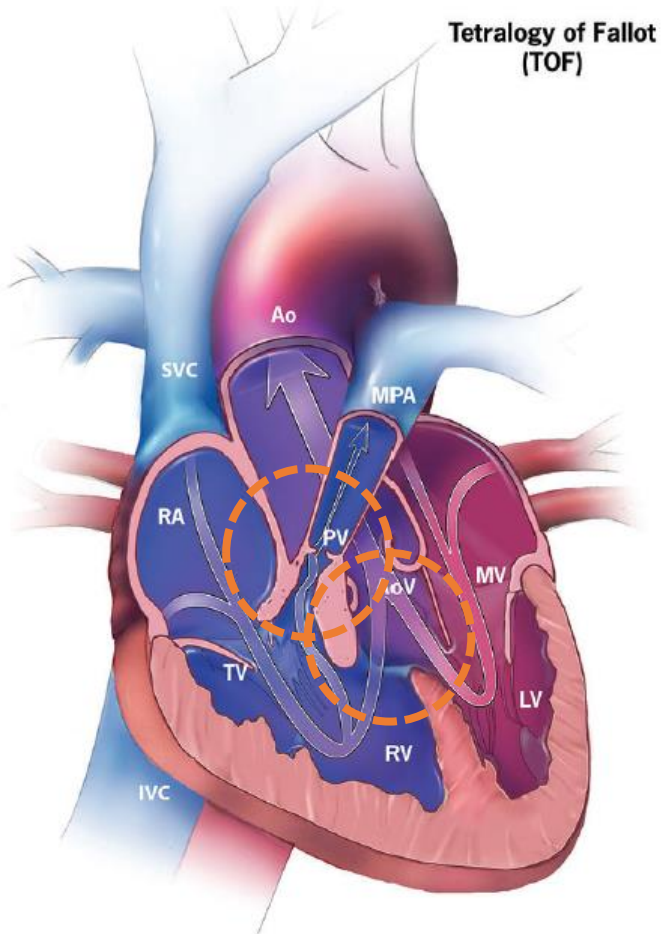
Tetralogy of Fallot (TOF)



RA. Right Atrium
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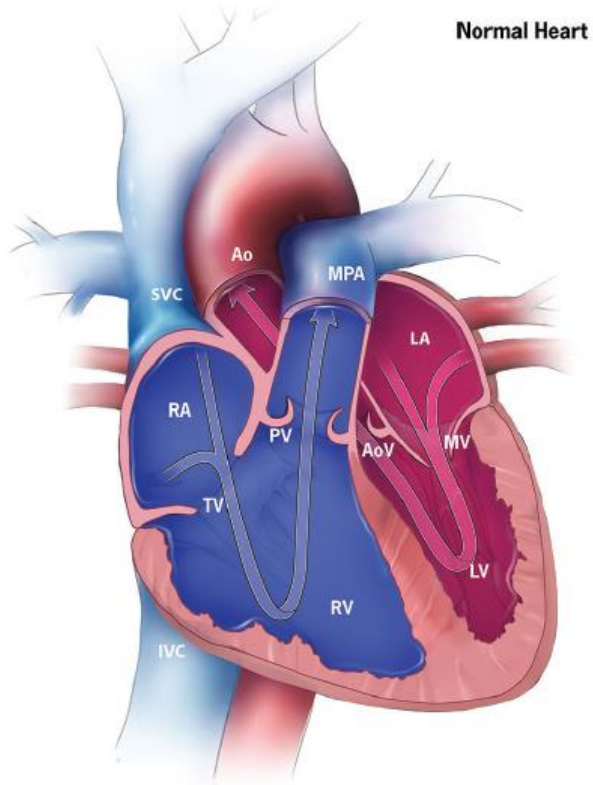
MPA. Main Pulmonary Artery
Ao. Aorta

TV. Tricuspid Valve
MV. Mitral Valve
PV. Pulmonary Valve
AoV. Aortic Valve

Normal Heart



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Newborn screening for critical congenital heart disease

- Can identify newborns with these conditions *before* signs or symptoms are evident and before the newborns are discharged from the birth hospital
- Uses a pulse oximeter to measure the percentage of hemoglobin in the blood that is saturated with oxygen

To improve efficiency and optimize results (high sensitivity, high specificity), it is crucial to follow an established protocol, train staff, and identify a rapid process to refer babies with a positive test to a clinical service able to provide a conclusive diagnosis

Source: https://www.cdc.gov/ncbddd/heartdefects/hcp.html

In cases of tetralogy of Fallot, always look for other birth defects and signs of genetic syndromes:

- A common genetic condition with tetralogy of Fallot (seen in about 15–20 % of cases) is deletion 22q11, a condition in which a small part of chromosome 22 is missing. This deletion leads to some CHDs and many types of birth defects, both visible externally (e.g. cleft palate, spina bifida) and internally (e.g. renal anomalies and many others) (~15–20%).
- Maternal pregestational diabetes is a modifiable risk factor for tetralogy of Fallot and other conotruncal conditions (e.g. truncus arteriosus).

Checklist for high-quality reporting

Tetralogy of Fallot – Documentation Checklist
☐ **Describe in detail the clinical and echocardiographic findings:** ▸ Anatomy – specify the type of right ventricular outflow tract obstruction (severity of stenosis, or presence of atresia) and the presence and type of ventricular septal defect (e.g. "subaortic", "perimembranous"). ▸ Procedure – specify whether the cardiac findings are from a prenatal or postnatal echocardiogram, or from other investigations (e.g. catheterization, MRI), surgery, or autopsy. ▸ Additional cardiac findings – specify any additional findings, including atrial septal defect, pulmonary collaterals, etc.
☐ **Look for and document extracardiac birth defects:** In deletion 22q11, the heart anomaly can be associated with several internal and external anomalies, including cleft palate, spina bifida, vertebral anomalies, or other defects.
☐ **Report whether specialty consultation(s) were done**, such as whether the diagnosis was made by a paediatric cardiologist, and whether the patient was seen by a geneticist.
☐ **Report any genetic testing and results** (e.g. chromosomal studies, genomic microarray, etc.).

Clinical and epidemiologic notes

In cleft palate, a complete evaluation and physical examination is crucial as it is more commonly associated with additional anomalies and syndromes compared to other types of clefts (e.g. cleft lip).

Additional clinical tips:

- Check for lip pits in the lower lip (see Fig. 23), in the child ***and*** in the parents – it is a sign of a genetic condition (van der Woude syndrome) with high recurrence risk (a parent may have the pits but not the cleft).
- Check for additional anomalies, especially of the heart (e.g. in deletion 22q11 syndrome) and eye (e.g. Stickler syndrome).
- Check for components of the Pierre Robin sequence, including microretrognathia (small recessed jaw), glossoptosis (posterior displacement of the tongue) and respiratory obstruction.

Checklist for high-quality reporting

Cleft Palate – Documentation Checklist

- ☐ **Describe in detail**, including:
  - Extension (cleft palate) – hard palate, soft palate.
  - Lower lip (see Fig. 23) – pits present or absent (when present, the van der Woude syndrome should be strongly suspected).
  - Presence of components of the Pierre Robin sequence – microretrognathia (small recessed jaw), glossoptosis (posterior displacement of the tongue) and respiratory obstruction.
- ☐ **Describe procedures to assess further additional malformations and if present, describe.**
- ☐ **Take and report photographs:** Very useful; can be crucial for review.
- ☐ **Report whether specialty consultation(s) were done and if so, report the results.** Plastic surgery and genetics consultation reports are useful.

Deletion 22q11.2

Synonyms, previously thought distinct syndromes: DiGeorge, Velocardiofacial s.

Etiology
Deletion (1.5–3 million base pairs in the proximal long arm of chromosome 22 (region 22q11.2)

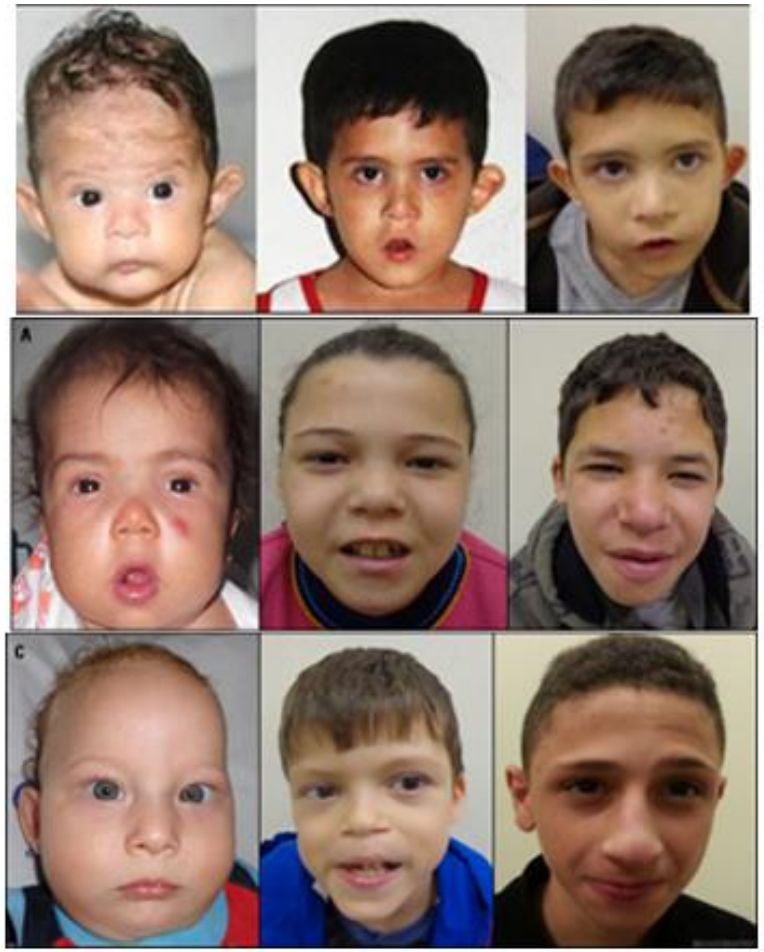
Estimated frequency
1 in 4 000 to 1 in 6 000 births
Botto LD et al, 2003

Variable presentation with many findings, of which these are quite common:

- Congenital heart defects (75%), mainly conotruncal anomalies
- Mild-to-moderate immune deficiency (75%)
- Palatal anomalies (75%), in particular **cleft palate (10%)**, submucous cleft palate (5–16%), velopharyngeal insufficiency
- Hypocalcemia during neonatal period (50%)
- Facial dysmorphism, with prominent nasal root, prominent ears, hooded eyelids
- Vertebral anomalies
- Speech and developmental delays
- Behavior issues, psychiatric disease in adolescents and adults

Kobrynski LJ and Sullivan KE, 2007

**Diagnosis in the neonatal period is challenging:
congenital heart disease and cleft palate can be early clues,
with diagnosis confirmed by genomic microarray**



Grassi MS et al, 2014

Facial traits at different ages

Note: conotruncal heart anomalies typically include ToF, pulmonary atresia with ventricular septal defects (as a variant of ToF), truncus arteriosus, transposition of the great arteries, and interrupted aortic arch type B.

Grassi MS, Jacob CM, Kulikowski LD, Pastorino AC, Dutra RL, Miura N, et. al. Congenital Heart Disease as a Warning Sign for the Diagnosis of the 22q11.2 Deletion. Arq Bras Cardiol. 2014 Nov;103(5):382-390. doi: 10.5935/abc.20140145. Epub 2014 Oct 10.

Coding the main findings

(from the MPM2)

What is your final description and diagnosis of Esther at this time? Describe the anomalies (using the checklists as guides), code, and try to provide an overall clinical classification.

Hint: for tetralogy of Fallot, the code is mentioned in the QRH and well described in the MPM2.

Coding Tetralogy of Fallot

(from the MPM2)

Relevant ICD-10 codes

Q21.3 Tetralogy of Fallot

Q21.82 Pentalogy of Fallot (do not use, see below)

Q22.0 Pulmonary valve atresia with Q21.0 (ventricular septal defect)

Note:

- Pulmonary valve atresia with ventricular septal defect is nearly always a form of tetralogy of Fallot (a very severe form), and for this reason it is grouped with classic tetralogy of Fallot.
- Pentalogy of Fallot – which comprises tetralogy of Fallot plus atrial septal defect – is an old term seldom used now. If tetralogy of Fallot occurs with an atrial septal defect, code separately the two defects; this approach also allows the atrial septal defect to be coded using a more specific code.

Inclusions

Q21.3 Tetralogy of Fallot

Q22.0 Pulmonary valve atresia with Q21.0 (ventricular septal defect)

Exclusions

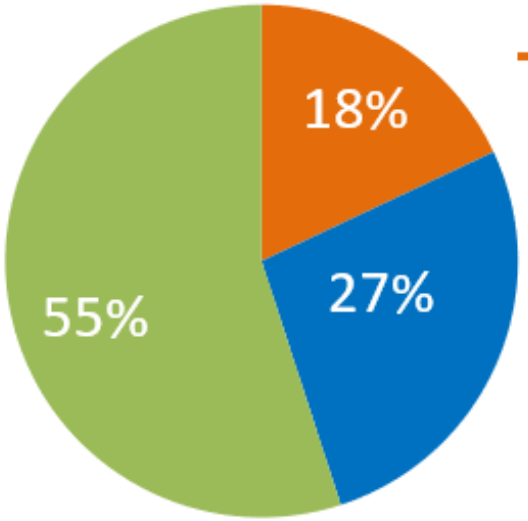
Q20.2 Double outlet right ventricle

Coding the main findings: summary

- Cleft palate, complete, involving hard and soft palate — code: Q35.5
  - No lip pits (if present, suspect Van der Woude syndrome)
- Micrognathia with Pierre Robin sequence — code: Q87.08
- Tetralogy of Fallot, with pulmonary stenosis and ventricular septal defect — code: Q21.3
- Chromosomal imbalance, specifically deletion 22q11.2 — code: D82.1

The overall clinical classification is “syndrome”: thus, when reporting cases of cleft palate, this case will be counted in the syndromic cleft palate column (and same for tetralogy of Fallot).

Clinical classification of cases with cleft palate



MCA Syndromes Isolated

> 400 syndromes have been described with oral clefts as occasional or common feature

Although individually rare, together these syndromes are not uncommon: look out for them

Percentage are approximate, compiled from different studies
Each study has its own definitions and methodology

Source: Mossey PA, Castilla EE. Global registry and database on craniofacial anomalies: Geneva, Switzerland: WHO. 2003. https://apps.who.int/iris/handle/10665/42840.

Commonly associated anomalies

- Micrognathia (Robin sequence)
- Heart defects
- Hydrocephaly
- Urinary tract anomalies
- Polydactyly
- Anomalies of cervical vertebrae*

Selected syndromes with CPO

Genetic syndromes

- Stickler syndrome (relatively frequent)
- 22q11.2 microdeletion (relatively frequent)
- Kabuki syndrome
- Orofaciodigital 1 syndrome
- Otopalatodigital syndrome
- Spondyloepiphyseal dysplasia congenita (SEDC)
- Treacher Collins syndrome
- Van der Woude syndrome

Teratogen-induced syndromes

- Retinoic acid embryopathy
- Valproate syndrome

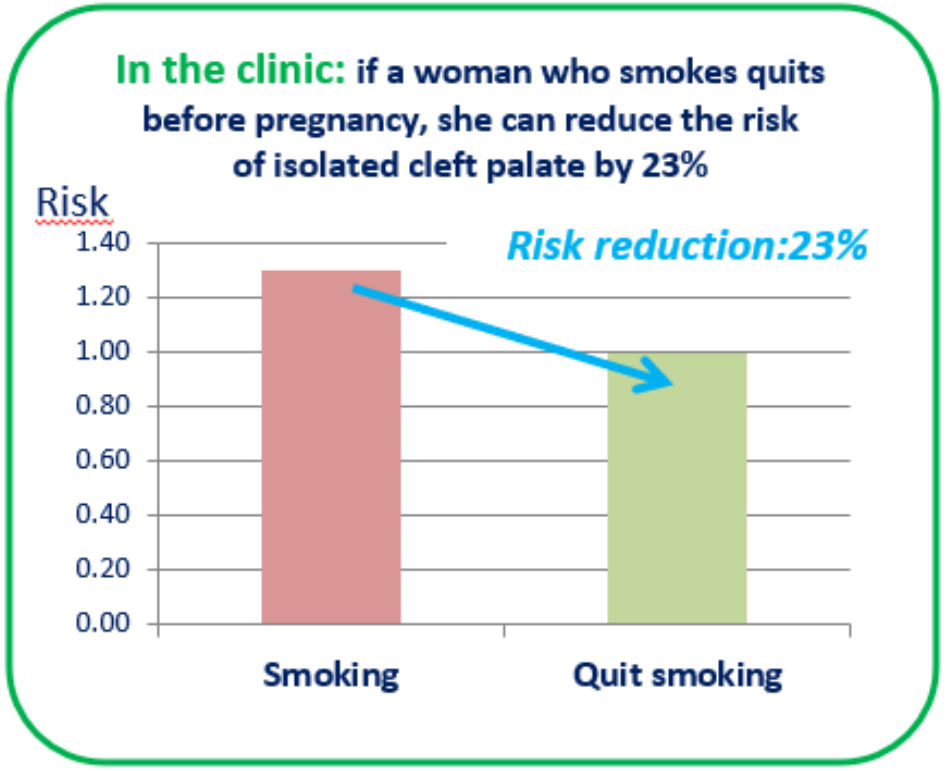
Source: Stevenson et al., 2016 and Jones et al., 2013

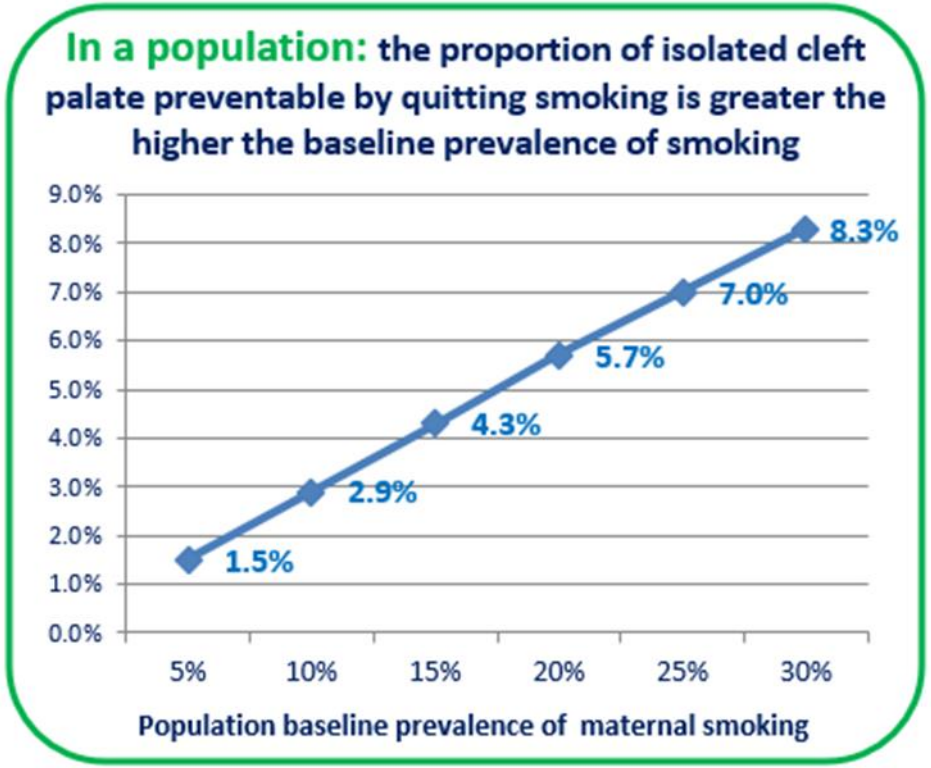
Part 3: Reviewing risk factors

Active smoking

Associated with a 30% increased risk in isolated cleft palate

- Based on a meta-analysis of 16 studies*
- Overall odds ratio of 1.30 (95% CI: 1.09-1.57)
- Some studies showed a dose-response effect
- Risk possibly also influenced by genetic variants (SNPs) in the fetus





*Xuan Z, Zhongpeng Y, Yanjun G, Jiaqi D, Yuchi Z, Bing S, Chenghao L. Maternal active smoking and risk of oral clefts: a meta-analysis. Oral Surg Oral Med Oral Pathol Oral Radiol 2016; 122: 680-690. pii: S2212440316302929.doi: 10.1016/j.oooo.2016.08.007.

Reproductive health risks associated with tobacco smoking

Pregnancy

- Infertility
- Delay in conception

- Ectopic pregnancy

- Placenta previa
- Placental abruption

- Premature rupture of membrane

Fetus – newborn – child

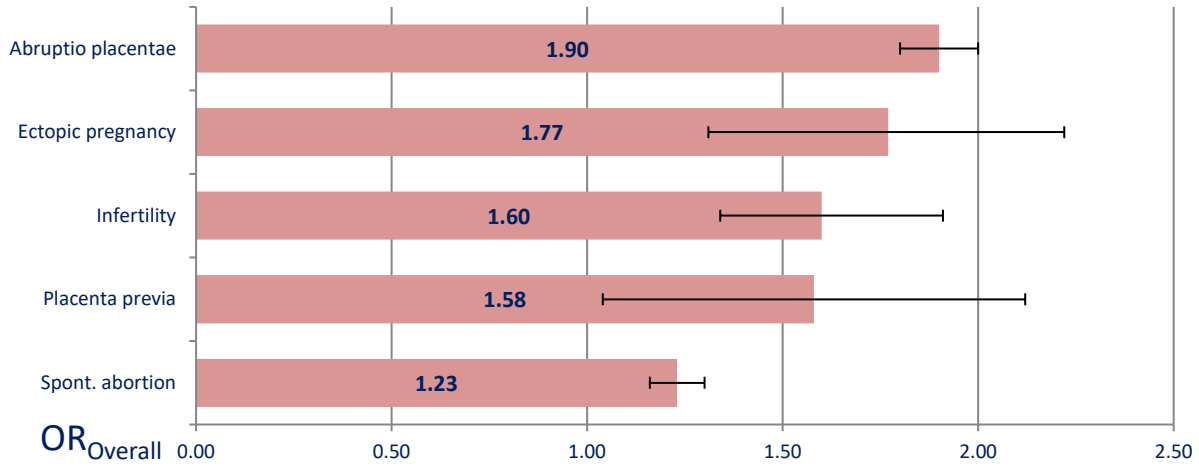
- Congenital anomalies
- Spontaneous abortion
- Stillbirth

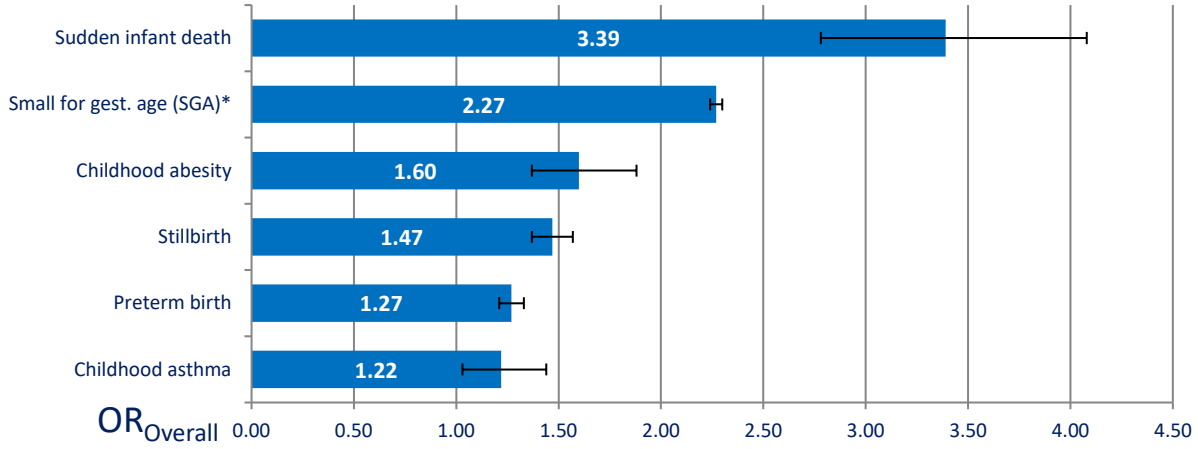
- Intrauterine growth restriction (IUGR)
- Preterm birth
- Sudden infant death (SIDS)

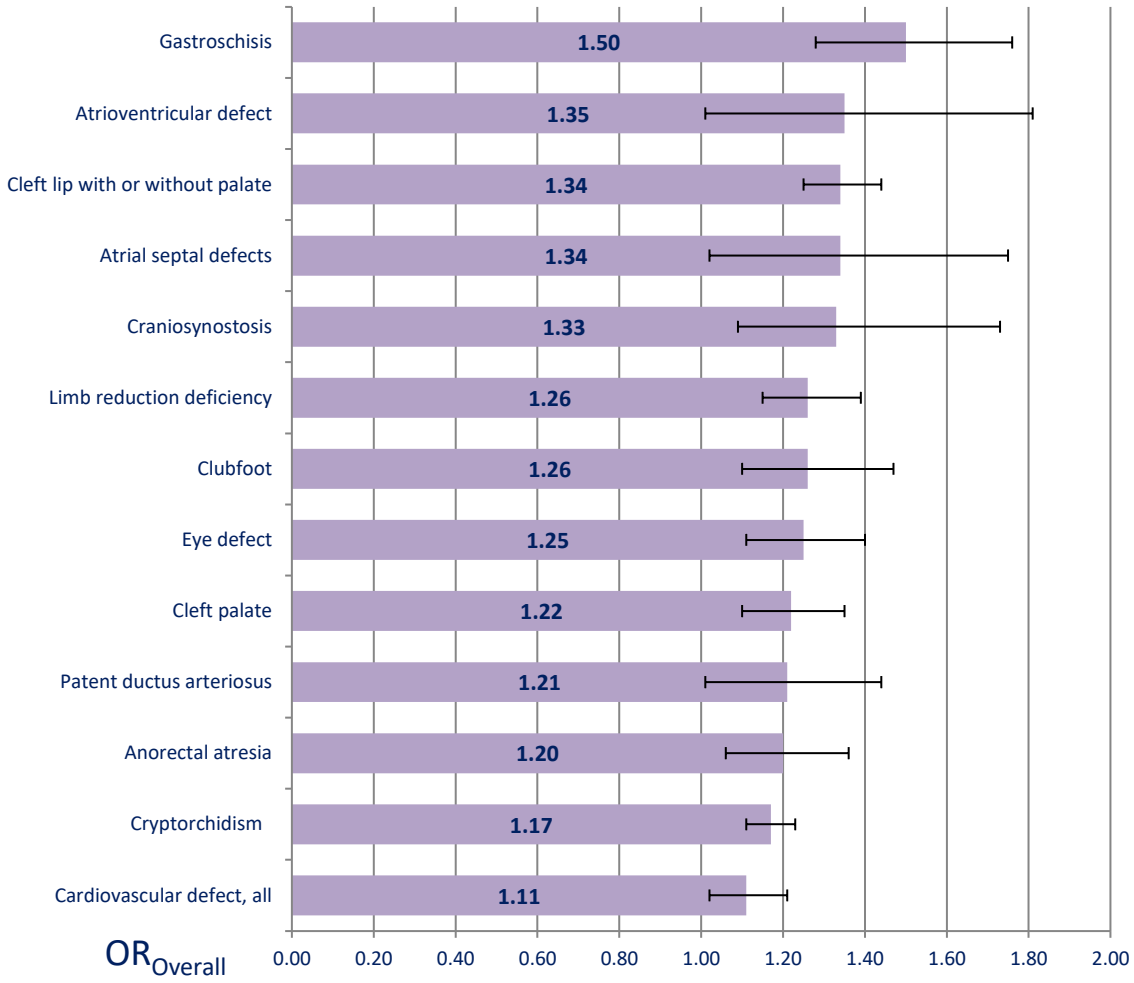
- Asthma/wheezing
- Childhood and adolescent overweight
- Acute lymphoblastic leukemia
- Non-Hodgkin lymphoma

Increased risk (odds ratio) associated with smoking

selected findings from meta-analyses





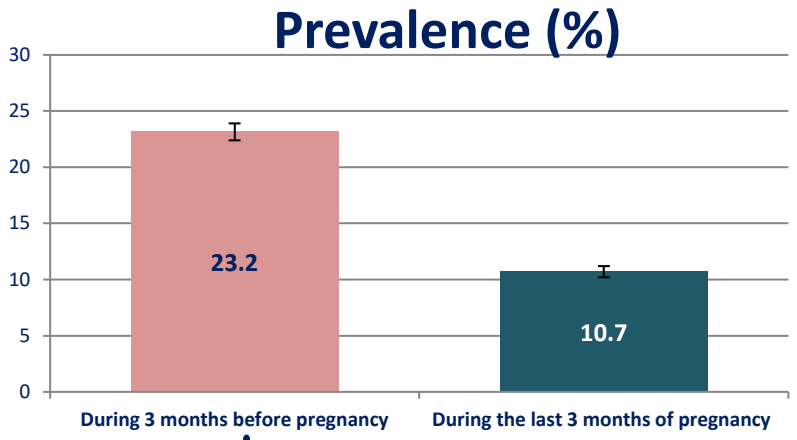


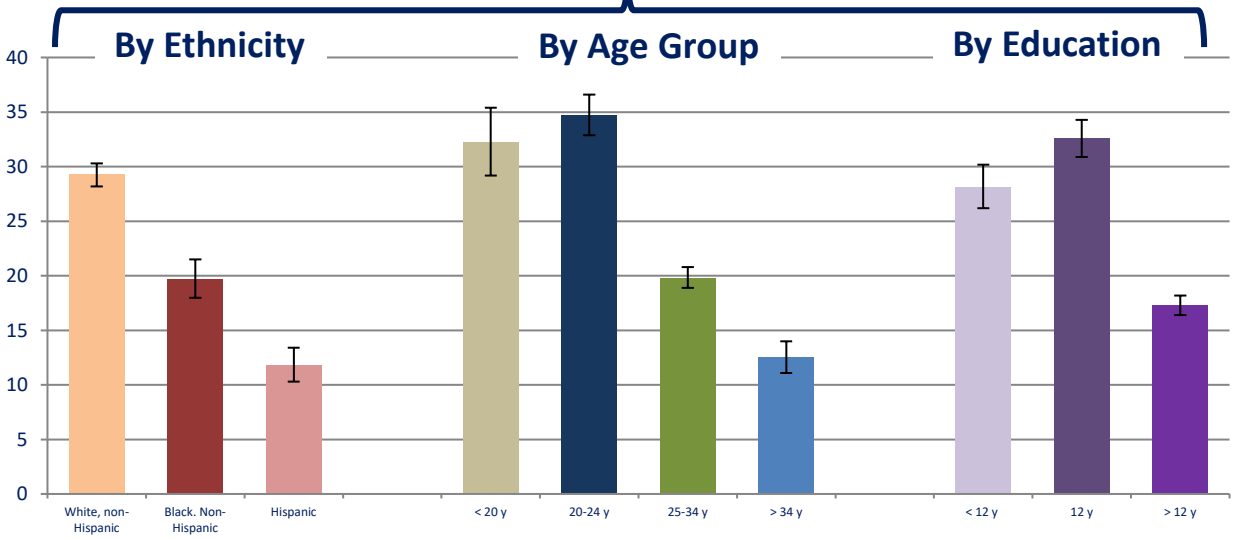
Source: Large cohort study, Aliyu HM et al, 2011

Prevalence of smoking before and during pregnancy

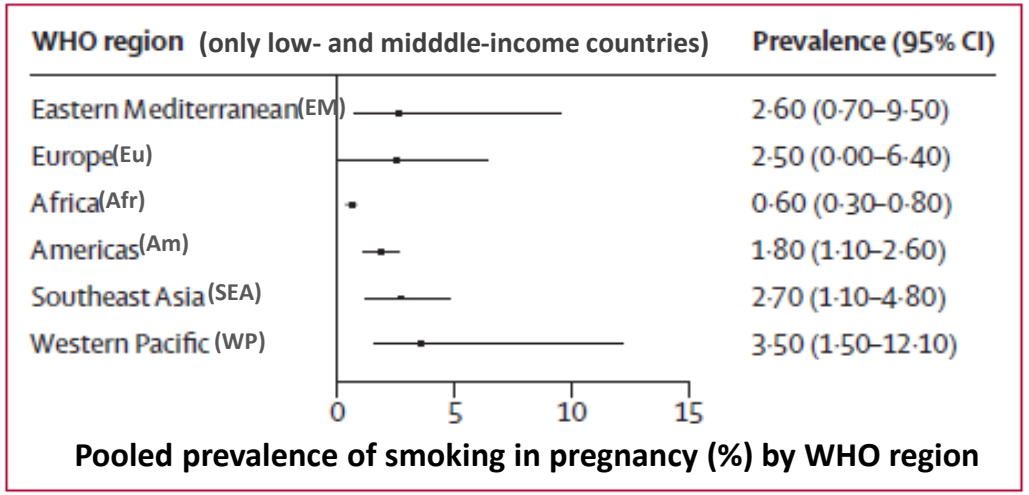
United States

Similar data needed everywhere

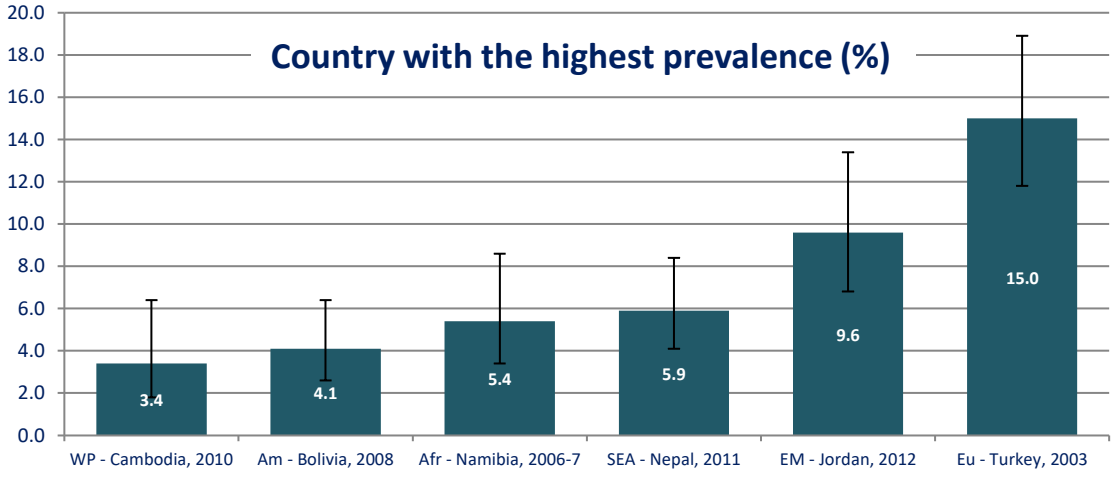




Source: Tong VT et al, MMWR 2013



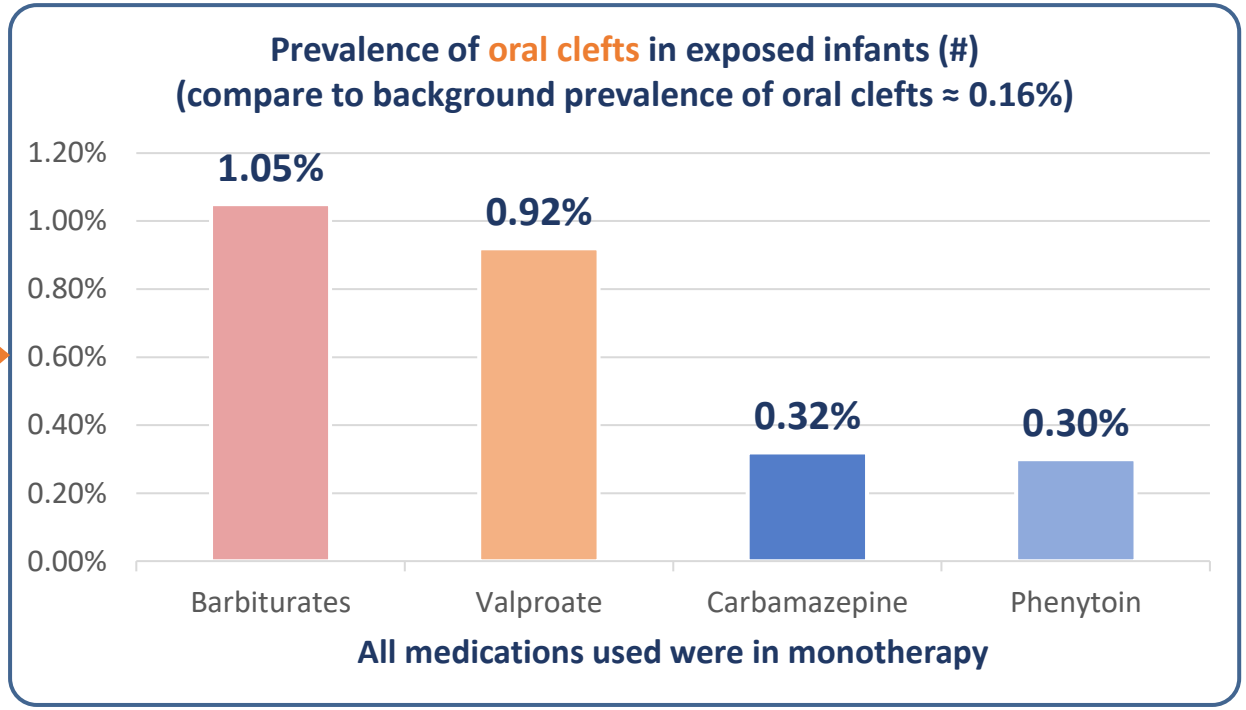
Pooled prevalence of smoking in pregnancy (%) by WHO region



Source: Caleyachetty R et al, Lancet 2014

Antiepileptics and oral clefts: a summary

- Antiepileptic medications increase the risk for frequent major anomalies, including **oral clefts**
  - Cleft lip with or without palate as well as cleft palate only
- Risk of **oral clefts** is higher for barbiturates and valporate, lower for carbamazepine and phenytoin



- The risk of **other antiepileptics** is generally less well known
- The exception is **topiramate**, a medication used for epilepsy, migraine, appetite suppression, and off label for conditions such as sleep and pyschiatric disorders
  - the associated prevalence of **oral clefts** [mainly CL(P)] in exposed infants of **1.0–1.3%** (de Jong J et al, 2016; Alsaad AM et al, 2015)

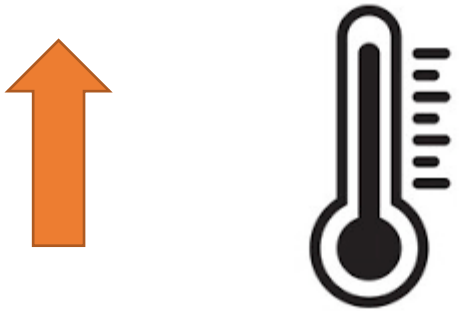
Fever and folic acid: risk and protective factors

High fever

First trimester exposure is associated with ~ two-fold increase of **any oral cleft**

- Based on a meta-analysis of 5 studies
- Overall odds ratio 1.94 (95% CI 1.35-2.79)

Dreier JW et al, 2014



Periconceptional folic acid supplements

Several studies (4 meta-analyses and a large cohort study in China) suggest periconceptional folic acid supplements **may reduce the risk of cleft lip with or without cleft palate, but are not conclusive for a similar effect for cleft palate only**

Butali A et al, 2013; Li S. et al, 2012; Johnson CY, Little J. 2008; Badovinac RL et al, 2007

Part 4: Concluding comments

Case study Esther: concluding comments and key points

This case study covers a lot of ground. With a firm grounding in the QRH, it should have been moderately challenging but quite doable.

What are some key points and take-home messages from this case study?

- Use a head-to-toe approach to examination in order not to miss organ systems/anomalies
- Use checklists consistently: quality increases, errors decrease, wasted time is minimized
- Cleft palate
  - Tricky condition: it may be missed if the baby is not examined systematically
  - Compared to cleft lip, more often associated with other anomalies, so be suspicious
  - Look out for related anomalies (Pierre Robin sequence), associated anomalies (e.g., heart anomalies) and syndromes (e.g., deletion 22q11, etc.)
- Critical congenital heart disease: can manifest at some distance from the time of birth, so important to examine baby carefully at birth and before discharge (including pulse oximetry if available)
- Smoking and other preventable causes of birth defects: learn about their frequency, support primary prevention

Useful resources/reading list

- Cleft palate and tetralogy of Fallot:
  - QRH
  - MPM2 (including evaluation questions).
- Deletion 22q11:
  - GeneReviews: www.ncbi.nlm.nih.gov/books/NBK1523/
  - a less technical overview: https://rarediseases.org/rare-diseases/chromosome-22q11-2-deletion-syndrome/
- Pierre Robin sequence:
  - NORD: https://rarediseases.org/rare-diseases/pierre-robin-sequence/
  - Orphanet: https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=718

- Smoking and pregnancy outcomes:
  - Simple overview (CDC) – www.cdc.gov/tobacco/basic_information/health_effects/pregnancy/index.htm

Evaluation

1. When describing a case of cleft palate, which elements of the birth defect itself should be reported, at a minimum, in the verbatim description according to the QRH check list? (**choose one or more answers)**

a. Extension (involvement of hard palate, soft palate, or both)
b. Presence (or not) of anomalies in the Pierre Robin sequence (e.g., glossoptosis, microretrognathia)
c. Anomalies of the lip (e.g., presence of lower lip pits)
d. Ear pits or tags

1. When describing a case of cleft palate, which elements of the birth defect itself should be reported at a minimum in the verbatim description according to the quick reference handbook check list? (**choose one or more answers)**

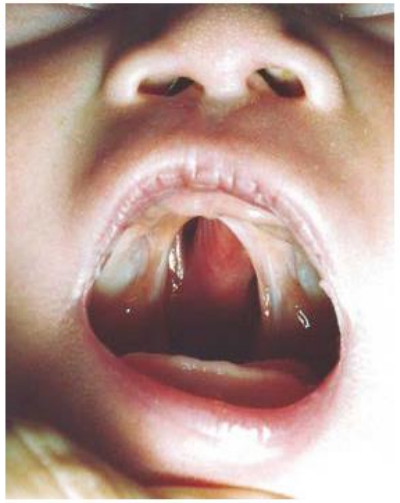
   a. **Extension (involvement of hard palate, soft palate, or both)**
   b. **Presence (or not) of anomalies in the Pierre Robin Sequence (e.g., glossoptosis, microretrognathia)**
   c. **Anomalies of the lip (e.g., presence of lower lip pits)**
   d. Ear pits or tags

Comment: extension of cleft palate, presence of anomalies of the Pierre Robin sequence, and presence or not of lip pits (and other lip anomalies) are key elements of a high-quality description, as described in the QRH checklist. The first two are associated with clinical severity and potential complications, which in turn drive morbidity and mortality. Pits in the lower lip are an indication that the cleft palate (or cleft lip with cleft palate) is part of an autosomal dominant condition, which has a different recurrence risk than the more common forms of cleft palate. All three elements of the description are easy and relatively quick to check on clinical exam.

Ear (preauricular) pits and tags are minor anomalies of the ear. While they can occur in some children with cleft palate, they are not strictly elements of the cleft palate. However, if they occur, we recommend reporting and describing them.

2. Match the images with the more appropriate description and definition

A



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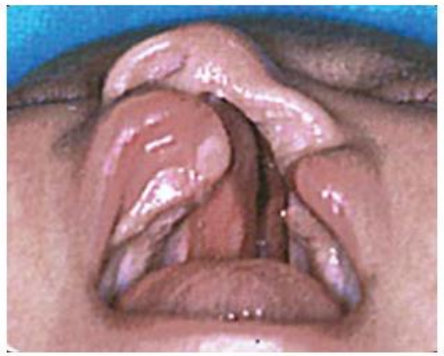


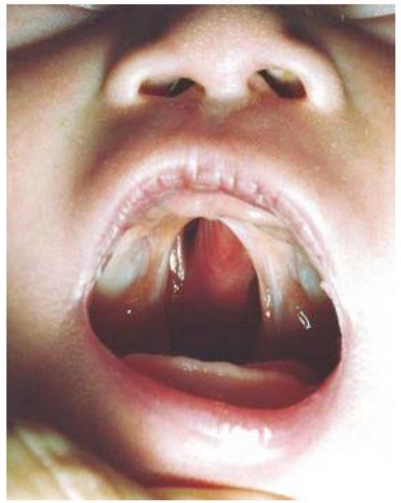
C

Photograph sources: © CDC–Beijing Medical University collaborative project; Dr. Pedro Santiago and Dr. Miguel Yanez; Dr. Jaime Frias

1. Cleft lip is characterized by a partial or complete fissure of the upper lip. It can be unilateral or bilateral. The cleft lip can extend through the gum, but not beyond the incisive foramen.
2. Cleft palate with cleft lip is characterized as a cleft of the upper lip extending through the hard palate (primary and secondary palate), and might also extend through the soft palate.
3. Cleft palate is characterized by a fissure (clefting) in the secondary palate (posterior to the incisive foramen) and can involve the soft palate only (the most posterior part of the palate), or both the hard palate and the soft palate.

2. Match the images with the more appropriate description and definition

A



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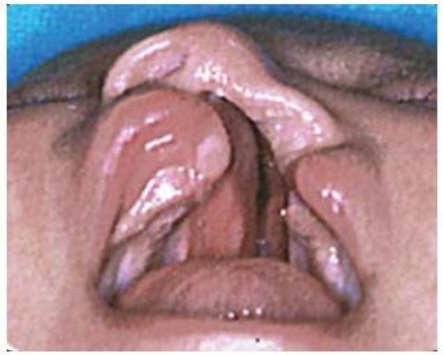

C

Photograph sources: © CDC–Beijing Medical University collaborative project; Dr. Pedro Santiago and Dr. Miguel Yanez; Dr. Jaime Frias

A3 (cleft palate), B2 (cleft lip and palate), C1 (cleft lip)

Key point: orofacial clefts have specific features that allow a clear distinction between the three main types, and it is important to describe these features.

Comment: refer to the QRH for additional images, drawings, and descriptions. Additional descriptors of these specific images may include the following:

A3. Cleft palate – involves both hard and soft palate, does not extend anteriorly of the foramen incisivum, does not involve gums; it is U-shaped (as opposed to V-shaped)

B2. Cleft lip and palate, unilateral left – appears to involve hard and soft palate

C1. Cleft lip, unilateral left – midline remnant of the philtrum is present (distinguishes the typical cleft left, which is slightly lateral of the median line from the atypical, median cleft lip, which is a different entity often associated with holoprosencephaly)

3. You see a newborn baby with complete cleft palate (involving both the hard and soft palate). The baby is not breathing well when placed supine (on her back). She is also small for gestational age and has microcephaly.
What anomalies should one look out for in such presentation?
(choose one or more answers)

a. Small jaw (micrognathia) and glossoptosis (small jaw) as part of the Pierre Robin sequence
b. Other internal and external anomalies, as cleft palate is isolated in only about half of cases
c. Genetic syndromes, if able (e.g., Stickler syndrome, deletion 22q11, many others)
d. Unlikely to find other anomalies, as cleft palate is nearly always an isolated malformation

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a (Pierre Robin sequence, small jaw), b (other anomalies), c (genetic syndromes)

3. You see a newborn baby with complete cleft palate (involving both the hard and soft palate). The baby is not breathing well when placed supine (on her back). She is also small for gestational age and has microcephaly. **What anomalies one should look out for in such presentation**?

Key point: cleft palate occurs with additional structural anomalies or genetic syndromes nearly half of the time, so a careful clinical exam is required in all cases and the additional anomalies should be carefully noted

Comment:

- **Nearly half of all cases of cleft palate may have something else** – additional major anomalies or syndromes; the implication is that finding a cleft palate calls for a careful head-to-toe examination of the baby and potentially the involvement of specialists

- Although many additional structural anomalies may be present, it is helpful to remember the **clinically most important and frequent** ones; these include micrognathia and Pierre Robin syndrome, congenital heart disease, and renal anomalies

- About **one quarter of all cases of cleft palate may have a genetic syndrome** – either a genomic imbalance or a single gene disorder; **deletion 22q11 and Stickler syndrome** are probably the two most common genetic conditions, worth remembering

- Finally, remember to **ask about pregnancy exposures**; retinoic acid and certain seizure medication, especially valproic acid, will increase the risk for clefting; these are preventable causes of clefting and should be sought for appropriate counseling and future prevention

4. The term **critical congenital heart disease** (CCHD) refers to a group of severe congenital heart anomalies that present typically around the time of birth or soon thereafter and require prompt diagnosis and intensive management.

Match the following type of CCHD with the appropriate features

a. Tetralogy of Fallot

b. Transposition of great arteries

1. Cyanosis, right ventricle connected to pulmonary artery, overriding aorta, can occur with deletion 22q1 and extracardiac anomalies

2. Cyanosis, right ventricle connected to aorta, tends to be isolated anomaly rather than associated with syndromes or extracardiac anomalies

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Correct: a1, b2

4. The term **critical congenital heart disease** (CCHD) refers to a group of severe congenital heart anomalies that present typically around the time of birth or soon thereafter and require prompt diagnosis and intensive management.

Key point: two common CCHD (tetralogy of Fallot and d-transposition of the great arteries) can cause cyanosis, but differ in anatomy and their associations with additional anomalies and syndromes

Tetralogy of Fallot has normally related great arteries whereas in D-transposition of the great arteries, the great arteries are ‘flipped’ compared to normal anatomy – the aorta originates from the right ventricle and the pulmonary arteries from the left ventricle

Although both are cyanotic conditions, they differ in epidemiologic features and clinical associations: **d-TGA is typically isolated and non-syndromic**, whereas **tetralogy of Fallot is more often associated with the presence of additional anomalies and genetic syndromes**

5. You are asked to discuss strategies to prevent cleft palate at a local meeting that includes public health professionals, primary care clinicians and ear-nose-throat specialists. You decide to focus on preventable causes.

What are well-established and preventable risk factors for cleft palate?

(choose one or more answers)

a. Smoking (30% increased risk for cleft palate)

b. Some seizure medications (e.g., valproate, barbiturates)

c. Rubella infection during pregnancy

d. Deletion 22q11 syndrome

e. High fever (about two-fold increased risk)

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e. **High fever (about two-fold increased risk)**

Answer: a, b, e

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What are well-established and preventable risk factors for cleft palate?

(choose one or more answers)

Key point: in some cases, cleft palate is a preventable anomaly.

- **Smoking is a well-established risk factor** for cleft palate, associated with a 30% increased risk (odds ratio in meta-analysis of about 1.3). Smoking is also a risk factor for many fetal and maternal health issues and complications, making it an important target for prevention.
- **Some seizure medications increase the risk for cleft palate**. Valproate increases the risk for both cleft palate and spina bifida, so its use in women should be avoided if at all possible.
- **High fever in early pregnancy has been associated with an increased risk** for cleft palate, although the evidence is not as strong as with smoking.

- Rubella infection in pregnancy is a well-established teratogen, causing rubella embryopathy (congenital rubella syndrome). However, cleft palate is <u>not</u> a common manifestation of congenital rubella syndrome (compared for example to congenital heart diseases or deafness).
- Deletion 22q11 is a cause of cleft palate, but currently deletion 22q11 is <u>not preventable</u>, except perhaps by appropriate preconception counseling of parents who carry this deletion.
- It is still debated whether or not folic acid supplementation or fortification can prevent orofacial clefts. The evidence is suggestive for cleft lip, less so for cleft palate.

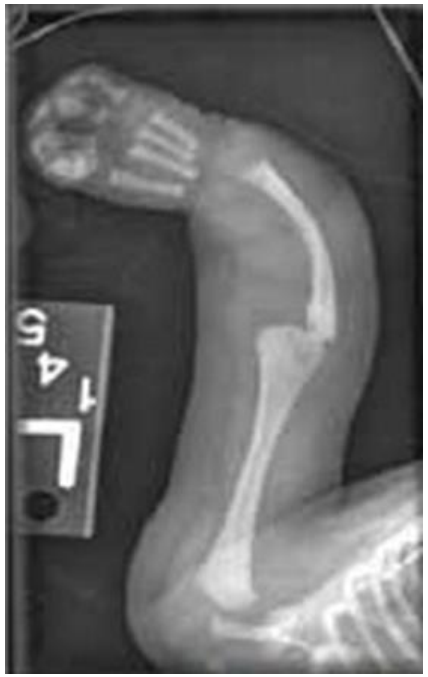
Clinical scenario 3: Baby Matias

Multiple congenital anomalies (VATER/VACTERL association, maternal diabetes)

Part 1: Clinical scenario

Clinical scenario: baby Matias

Part 1. Baby boy Matias is born at 38 weeks of gestational age, in apparent good health. At birth, however, you notice differences in in his left arm and hand.



Q1. Describe in detail what you see, using the visual aids and description checklist in the QRH.

Photograph and imaging source: © ECLAMC

Clinical scenario: baby Matias

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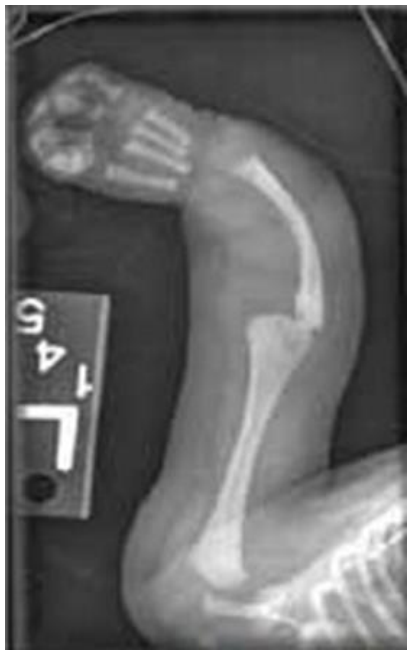

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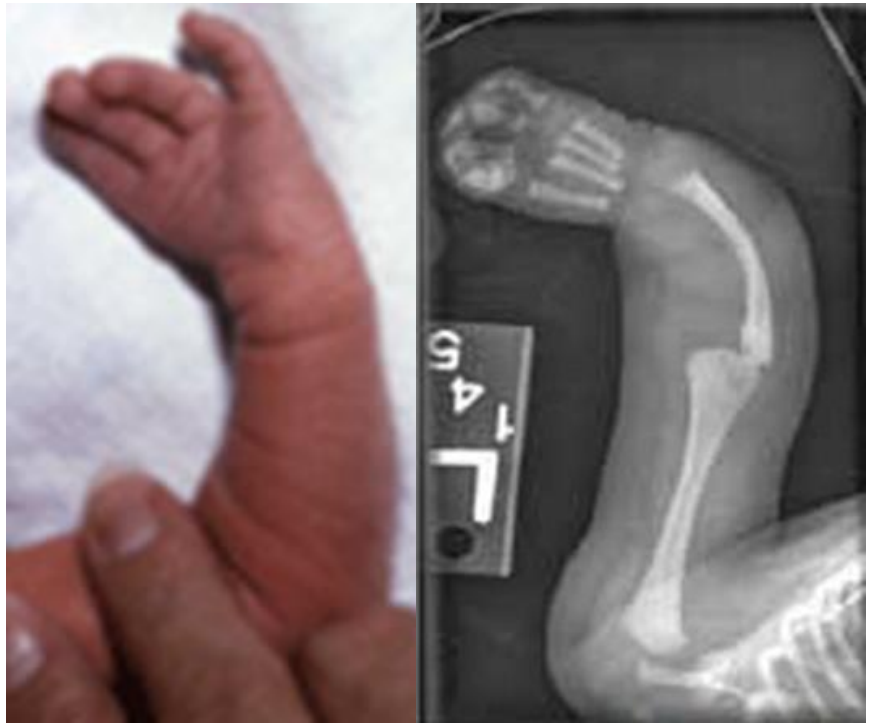
Checklist for high-quality reporting

Longitudinal Preaxial Defects – Documentation Checklist
☐ **Describe in detail**, including: ▸ Limbs involved. ▸ Note each segment involved for each limb affected – describe what is deficient or absent. Indicate involvement of radius, tibia, first–second finger, first–second toe, fibula, and others. ▸ Laterality – right, left, bilateral. ▸ Document specialty consultations (e.g. genetics, orthopaedics). ☐ **Use Fig. 35 to distinguish longitudinal preaxial defects from other subtypes of limb deficiencies.**

Photograph and imaging source: © ECLAMC

Clinical scenario: baby Matias

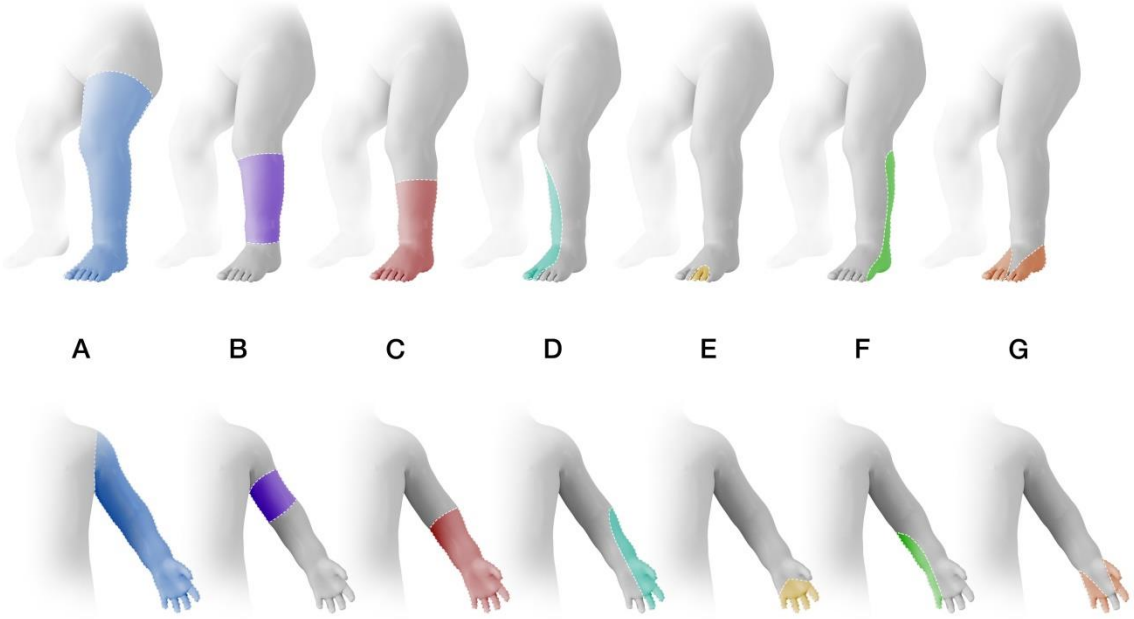
Q2. How would you classify this type of limb deficiency?



Photograph and imaging source: © ECLAMC

Types of limb deficiencies by axis and segment involved

(see text for details)

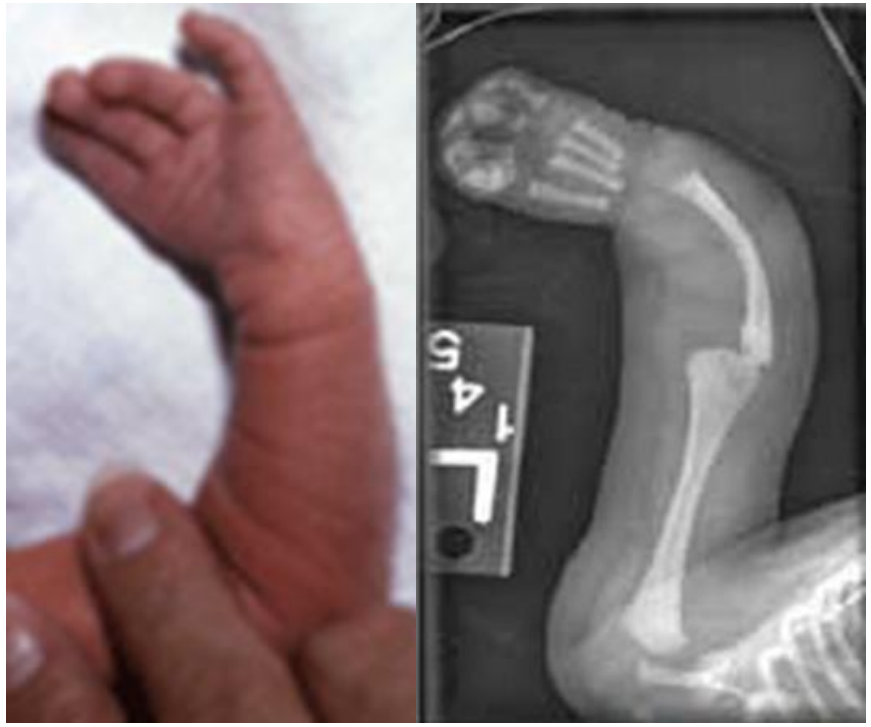


Absent or hypoplastic structures are shaded. (A): complete absence of limb (amelia); (B) intercalary defect; (C) terminal transverse defect; (D) longitudinal defect, preaxial; (E) longitudinal defect, central; (F) longitudinal defect, postaxial; (G) longitudinal, pre- and postaxial

Adapted from: Gold NB, Westgate MN, Holmes LB. Anatomic and etiological classification of congenital limb deficiencies. Am J Med Genet A 2011;155A(6):1225–35.

Clinical scenario: baby Matias

Q2. How would you classify this type of limb deficiency?



Photograph and imaging source: © ECLAMC

From the QRH

Preaxial limb deficiency is characterized by the absence or hypoplasia of the **'preaxial' segments** (those on the side of the thumb or big toes) of the upper or lower limb (see Fig. 42).

Preaxial limb deficiencies include:

- Hypoplasia/absence of the thumb (sometimes of the second finger)
- Hypoplasia/absence of the radius
- Hypoplasia/absence of the big toe (sometimes of the second toe)
- Hypoplasia/absence of the tibia

Part 2: Deeper dive into the clinical presentation

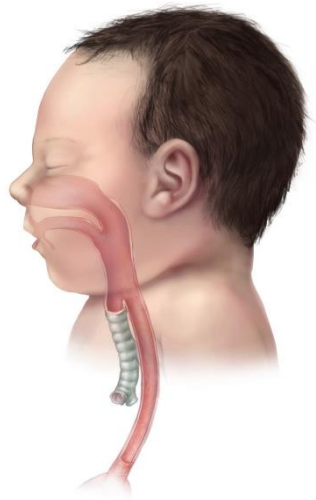
Clinical scenario: baby Matias

What might be going on – what is likely to be the congenital anomaly?

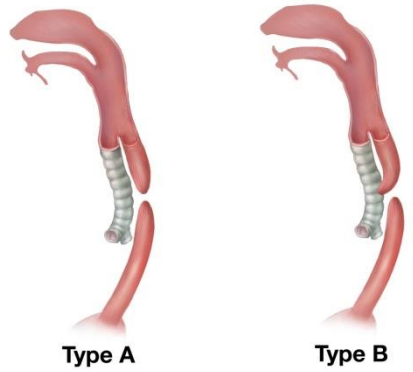
[...] As you are looking at the radiograph, the mother starts breastfeeding. However, though active and obviously hungry, Matias immediately starts choking and appears not to be able to swallow the breastmilk. Mom tries again, but the same happens.

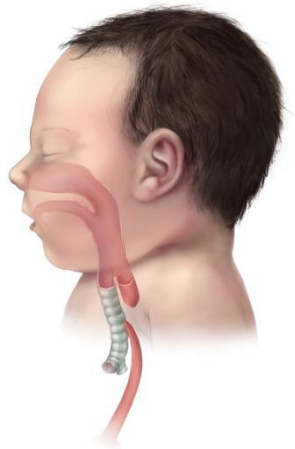
You try to pass a soft plastic feeding tube to the stomach but are unable.

Matias has a quick chest radiograph done and your radiologist colleague confirms that the radiopaque tube is not reaching the stomach but appears to be curled up more proximally.

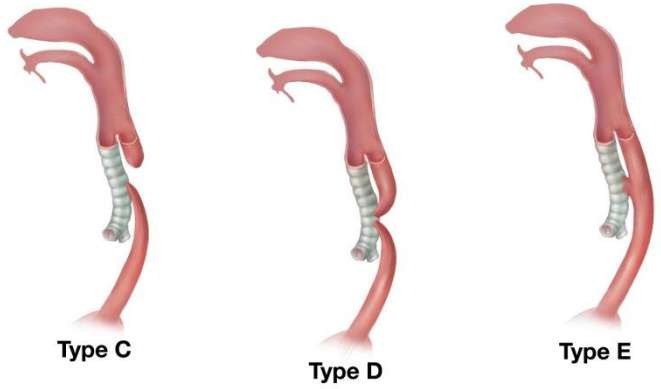


Normal oesophagus and trachea





Oesophageal atresia, type C



Clinical scenario: finding more...

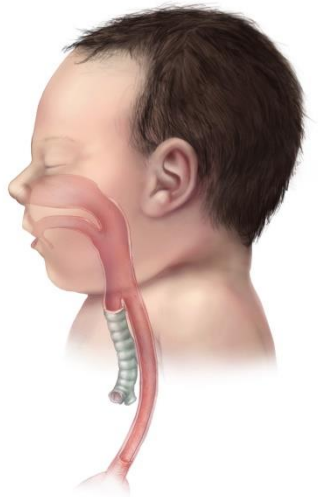
[...] In the following days Matias undergoes further evaluations. It is determined that he has **oesophageal atresia type C** – the type of oesophageal atresia in which the distal esophagus connects to the trachea. This is the most common type.

He is also noted to have two abnormally shaped thoracic vertebrae that the radiologist characterizes as '**hemivertebrae**'. Finally, he has an echocardiogram done, showing **normal cardiac anatomy**.

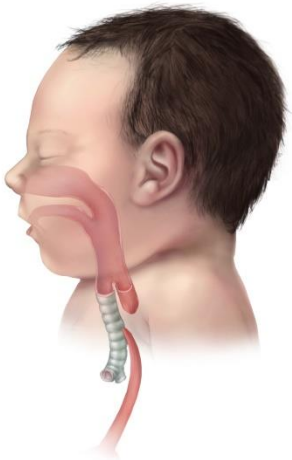
Now you take stock of Matias's examinations – he has a preaxial limb deficiency in his left arm (missing radius plus thumb), oesophageal atresia (type C), and vertebral anomalies (hemivertebrae).

With three major congenital anomalies, you are concerned that there might be more. You re-examine the baby from head to toe and notice that there is a **shallow dimple where the anus should be**. You cannot see any stool coming out, and mom tells you that Matias has not stooled yet. You ask about urine and she says he seems to urinate fine, but the urine seems as if it contains dark matter, like stool.

Now, having gone through the QRH for the prior anomalies, you come across the following **clinical note** […].



Normal oesophagus and trachea



Oesophageal atresia, type C

Thoracic hemivertebra

+ anorectal anomaly ?

Clinical scenario: baby Matias

Oesophageal atresia

Clinical and epidemiologic notes

Oesophageal atresia is frequently (55%) associated with additional birth defects that include:

- Other unrelated birth defects, particularly cardiac, anorectal, skeletal/vertebral and urogenital.
- Anomaly complexes (e.g. OAVS [**o**culo-**a**uriculo-**v**ertebral **s**pectrum], VATER or VACTERL association [**v**ertebral, **a**nus, **c**ardiac, **t**rachea, o**e**sophagus, **r**enal, **l**imb]).
- Genetic syndromes (e.g. trisomies 18 and 21, CHARGE [**c**oloboma, **h**eart defects, choanal **a**tresia, growth **r**etardation, **g**enital abnormalities, **e**ar abnormalities] syndrome, Feingold syndrome).

What are the associations shared by both oesophageal atresia and preaxial limb deficiencies?

What are the main *internal* anomalies to rule out upon seeing a preaxial anomaly?

Preaxial limb deficiency

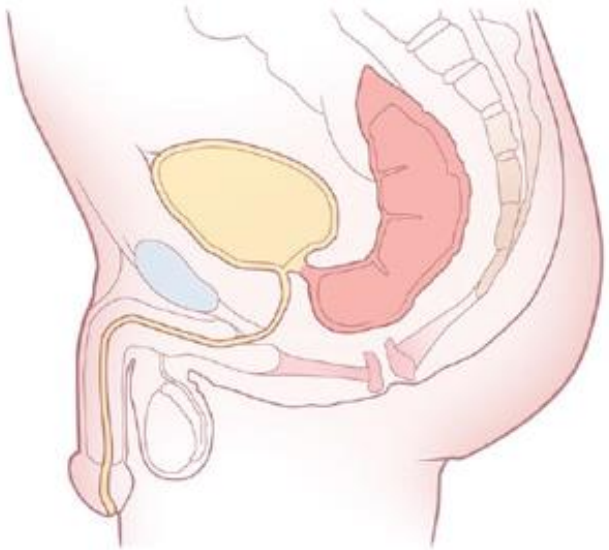
Clinical presentation:

- Radial deficiencies are commonly associated with other anomalies such as in the VATER/VACTERL (**v**ertebral, **a**nus, **c**ardiac, **t**rachea, o**e**sophagus, **r**enal, **l**imb) association as well as several genetic syndromes. Some possible genetic diagnoses include trisomy 18, Fanconi anaemia, Holt-Oram syndrome, thrombocytopenia absent radius syndrome (TAR). Of note, several genetic conditions with radial deficiency present also hematologic abnormalities (Diamond-Blackfan anaemia, Fanconi anaemia, TAR).

Given this presentation, you are suspicious that he might have an anorectal malformation (ARM)due to the unusual anal findings (indentation but not a clear anal passage, and no stools passed). You are suspicious about Matias possibly having a phenotype overlapping with VACTERL association, so you also want to look for renal anomalies even if his urine output looks fine.

You are now able to order a renal ultrasound. You also involve the pediatric surgeon to assess for anorectal anomalies.

The specialist comes to see the mother while you are there and shows her the following drawings.



Q5. Describe the anomalies

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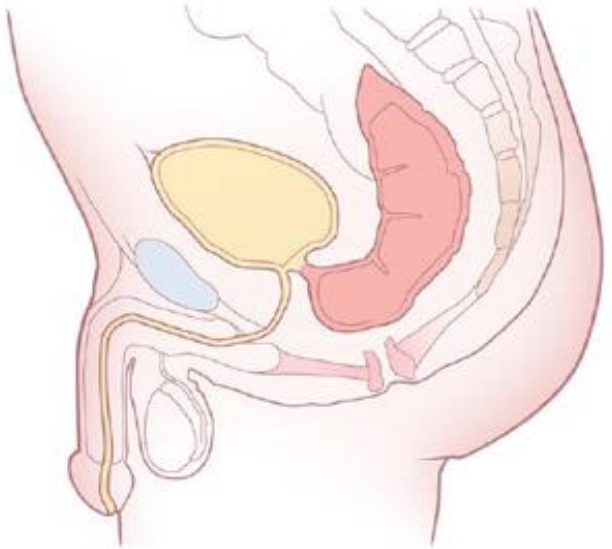


Q5. Describe the anomalies

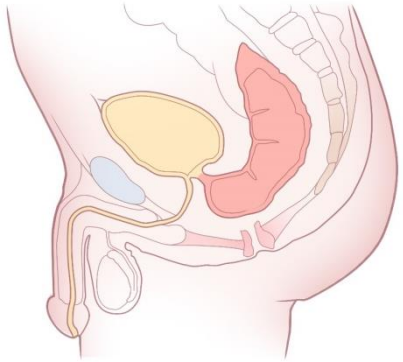
Matias has two additional anomalies:

- **unilateral left renal agenesis**: unilateral agenesis may be completely asymptomatic for many years
- **anorectal atresia, high** (rectum terminates above the pelvic floor muscles) **with a recto-vesical fistula** (rectum connects to bladder); the fistula explains the urine mixed with stool

Clinical scenario: baby Matias

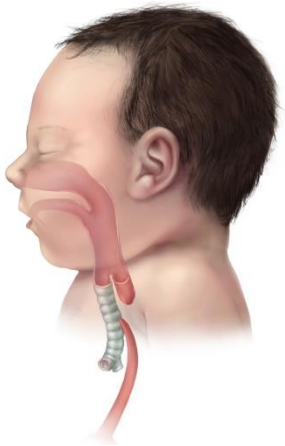
Thoracic hemivertebra

V



Anorectal atresia, high, with recto-vesical fistula

A



Oesophageal atresia with tracheoesophageal fistula, type C

TE

Renal agenesis, unilateral left

R

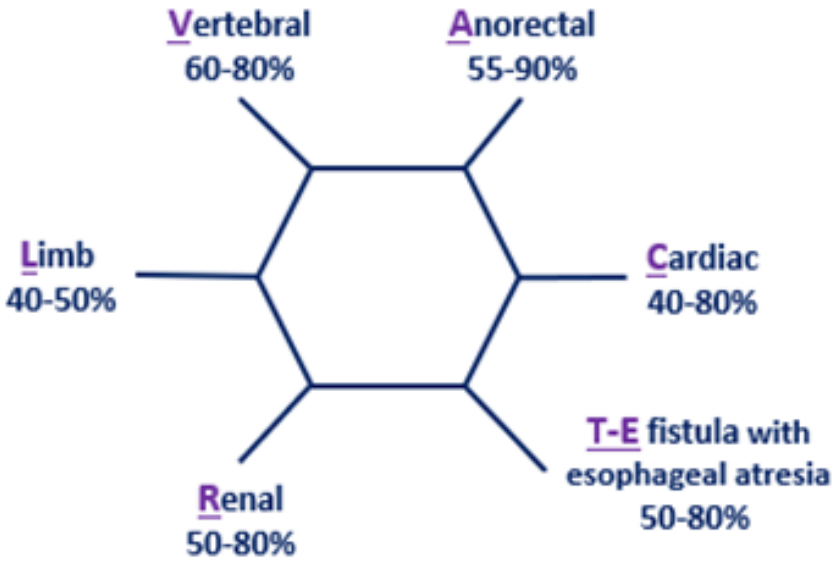
Preaxial limb deficiency
Unilateral left arm
Missing thumb and radius

L

Photograph source: © ECLAMC

VACTERL association

VACTERL association is commonly defined as the co-occurrence of at least three of the six anomalies included in the acryonym



Main anomalies described among VACTERL patients
Percentages are approximate and based on the review of Solomon DB, 2011

Limb deficiency is almost always a preaxial defect

VACTERL is a multiple congenital anomaly (MCA) association, because these anomalies occur together more frequently than expected by chance (non-random)

The differential diagnosis includes *diabetic embryopathy* and several genetic syndromes (e.g.,):

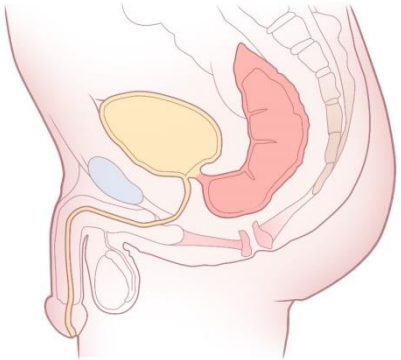
- Baller-Gerold
- CHARGE
- Currarino
- Deletion 22q11.2
- Fanconi anemia
- Feingold
- Holt-Oram
- Opitz G/BBB
- Pallister-Hall
- Townes-Brocks

Diagnosis is important because of the implications for care and counseling

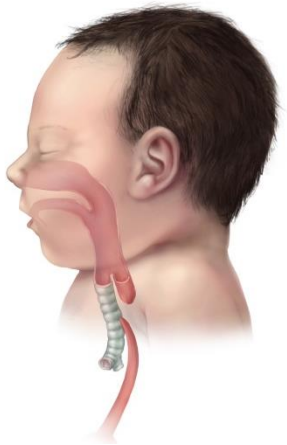
- **No specific cause** has been consistently found, although in some sporadic cases a genetic abnormality has been reported
- Infants in this highly heterogeneous and loosely defined group should be **classifed in the group of MCA** and have all **anomalies described in detail**
- **Prevalence:** 0.25 to 1.0 per 10 000 births; but with high variability due to variations in definitions and inclusion criteria

Clinical scenario: coding the findings

Thoracic hemivertebra



Anorectal atresia, high, with recto-vesical fistula



Oesophageal atresia with tracheo-oesophageal fistula, type C

Renal agenesis, unilateral left

Preaxial limb deficiency
Unilateral left arm
Missing thumb and radius

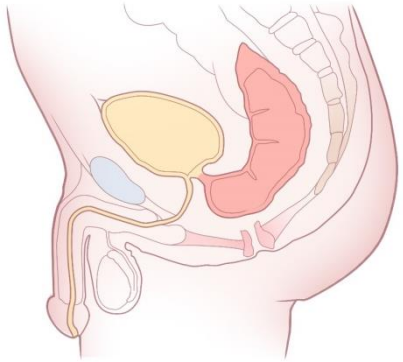
Photograph source: © ECLAMC

Clinical scenario: coding the findings

Thoracic hemivertebra

Q76.30

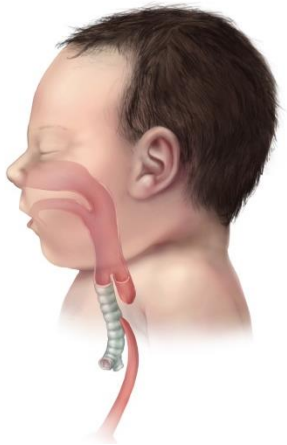
Note that some programmes may not include this among eligible anomalies



Anorectal atresia, high, with recto-vesical fistula

Q42.0

Note that presence but not type of fistula is captured



Oesophageal atresia with tracheo-oesophageal fistula, type C

Q39.1

Note that the specific type is lost in the coding

Renal agenesis, unilateral left

Q60.0

Note that laterality is lost in the coding

Preaxial limb deficiency
Unilateral left arm
Missing thumb and radius

Q71.4
(+/- Q71.31)

May be complicated to code; typically only Q71.4 is used even if both radius and thumb are missing

+ Q87.26 VACTERL association

Photograph source: © ECLAMC

Part 3: Risk factors

As you discuss the findings with parents, you review family history and **pregnancy history**. It turns out that the mother has **type 2 diabetes**, which was not controlled until the fourth month of pregnancy, with **HbA1c** values in the first trimester **between 8% and 12%** (normal reference range, ≤5.7%)

Q9. To what extent is maternal diabetes relevant to Matias's presentation?

What are some congenital anomalies associated with maternal diabetes?

How is it relevant to prevention?

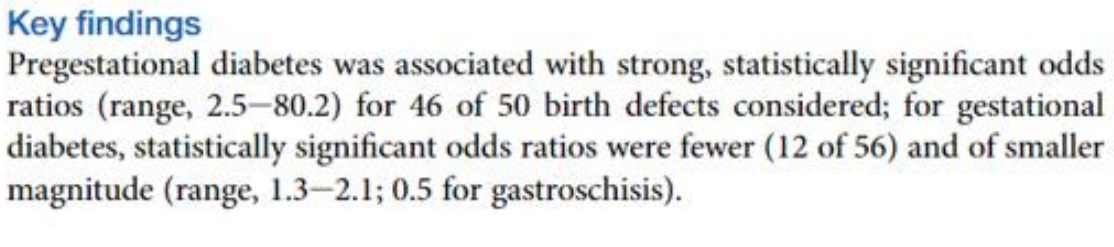
Maternal diabetes: a preventable teratogen for many congenital anomalies

OBSTETRICS

Specific birth defects in pregnancies of women with diabetes: National Birth Defects Prevention Study, 1997–2011

Sarah C. Tinker, PhD; Suzanne M. Gilboa, PhD; Cynthia A. Moore, MD, PhD; D. Kim Waller, PhD; Regina M. Simeone, MPH; Shin Y. Kim, MPH; Denise J. Jamieson, MD; Lorenzo D. Botto, MD; Jennita Reefhuis, PhD; and the National Birth Defects Prevention Study

https://pubmed.ncbi.nlm.nih.gov/35665489/



Key findings

Pregestational diabetes was associated with strong, statistically significant odds ratios (range, 2.5–80.2) for 46 of 50 birth defects considered; for gestational diabetes, statistically significant odds ratios were fewer (12 of 56) and of smaller magnitude (range, 1.3–2.1; 0.5 for gastroschisis).

Pre-existing diabetes substantially increased the risk for many types of major birth anomalies

- Risk increased for 46 out of 50 congenital anomalies studied
- Risk highest for sacral agenesis (a birth anomaly of the lower spine), holoprosencephaly (a birth defect of the brain), and limb deficiencies
- Risk also present for several types of severe congenital heart defects

Women with pre-existing diabetes can increase their chances of having a healthy baby by adopting health behaviours before and during pregnancy, including:

- Seeing a health-care professional regularly
- Keeping blood sugar levels under control, with the help of a doctor and dietician, before and during pregnancy
- Getting 400 micrograms (µg) of folic acid every day before and during pregnancy
- Striving to reach and maintain a healthy weight before pregnancy

Screening and treating before pregnancy prevents congenital anomalies and improves the health of mother and baby

Source: Marchincin SL, Howley MM, Van Zutphen AR, Fisher SC, Nestoridi E, Tinker SC, Browne ML; National Birth Defects Prevention Study. Risk of birth defects by pregestational type 1 or type 2 diabetes: National Birth Defects Prevention Study, 1997-2011. Birth Defects Res. 2023 Jan 1;115(1):56-66. doi: 10.1002/bdr2.2050.
Tinker SC, Gilboa SM, Moore CA, Waller DK, Simeone RM, Kim SY, Jamieson DJ, Botto LD, Reefhuis J; National Birth Defects Prevention Study. Specific birth defects in pregnancies of women with diabetes: National Birth Defects Prevention Study, 1997-2011. Am J Obstet Gynecol. 2020 Feb;222(2):176.e1-176.e11. doi: 10.1016/j.ajog.2019.08.028.

Part 4: Concluding comments

Matias's story: from newborn exam to prevention

key points, lessons learned and comments...

- Limb deficiencies are diagnosable at the initial newborn exam
- Examine head to toe, front and back, as well as top and bottom
- Describe all anomalies precisely and completely
- Maternal diabetes is a strong and preventable risk factor for many congenital anomalies

Evaluation

1. When describing a case of **limb deficiency**, which **elements should be reported,** at a minimum, in the verbatim description according to the QRH checklist?

Choose <u>one or more</u> answers

a. Limb involved (hand, foot, leg, etc.)
b. Segment and ideally bone(s) involved
   (e.g., absent tibia, 2nd and 3rd digit of the hand, etc.)
c. Laterality (left, right, bilateral)
d. Diagnostic label only (e.g., preaxial, postaxial, terminal deficiency

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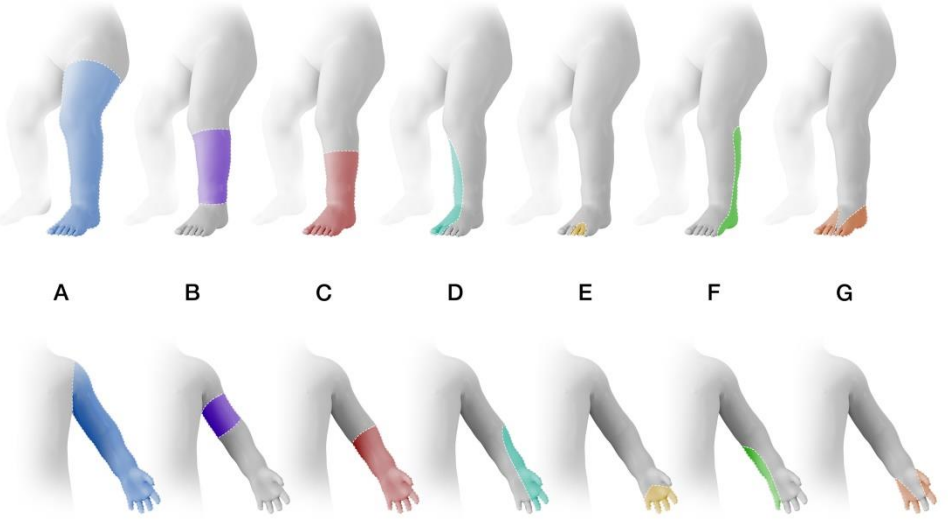
b. **Segment and ideally bone(s) involved**
(e.g., absent tibia, 2nd and 3rd digit of the hand, etc.)

c. **Laterality (left, right, bilateral)**

d. Diagnostic label only (e.g., preaxial, postaxial, terminal deficiency



Key point: limb deficiencies can be quite complex – describe anatomy systematically (limb, segment, laterality)

Comment: the anatomy of limb deficiencies is crucial for appropriate classification; anatomic details also help one understand clinical severity, health needs and risk for disability; just stating ‘preaxial limb deficiency of the upper limb’ is less than optimal – the label may be wrong, and does not provide crucial information on anatomy and severity

2. When describing a case of **anorectal anomaly**, which elements should be sought from the records and reported?

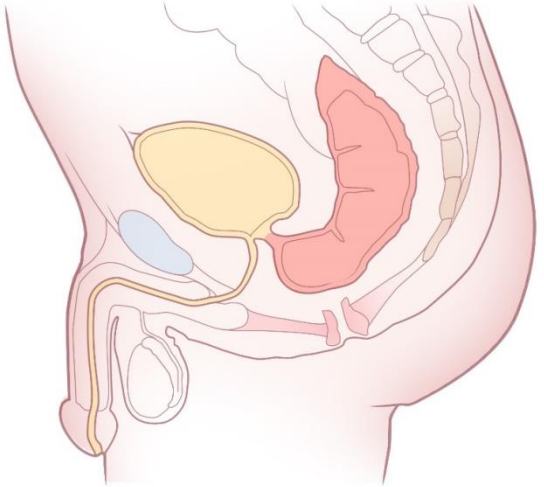
Choose <u>one or more</u> answers

a. Fistula – absent or present
b. Atresia limited to anus versus including rectum
c. Fistula type – involving which structure
d. Level of atresia – high or low

2. When describing a case of **anorectal anomaly**, which elements should be sought from the records and reported?

Choose <u>one or more</u> answers

a. **Fistula – absent or present**
b. **Atresia limited to anus versus including rectum**
c. **Fistula type – involving which structure**
d. **Level of atresia – high or low**



Key point: use the QRH checklist to describe anorectal anomalies, which can present in a variety of ways

Comment: two of the key traits in anorectal anomalies is whether the anomaly is high or low (i.e., the level of the atresia in relation to the pelvic floor) and what types of fistulas, if any, are present; these two traits are associated with clinical and surgical complexity and with the likelihood of additional anomalies; for these reasons, documenting the anomalies carefully and completely supports optimal clinical care and epidemiologic surveillance

3. A phenotype consisting of the co-occurrence of the following anomalies (vertebral anomalies, anorectal malformation, cardiac anomalies, oesophageal atresia/tracheoesophageal fistula, renal anomalies, and preaxial limb deficiencies) constitutes the … **(choose the best answer)**

a. VATER/VACTERL syndrome
b. VATER/VACTERL sequence
c. VATER/VACTERL association
d. VATER/VACTERL combination

3. A phenotype consisting of the co-occurrence of the following anomalies (vertebral anomalies, anorectal malformation, oesophageal atresia / tracheoesophageal fistula, renal anomalies, and preaxial limb deficiencies) constitutes the … **(choose the best answer)**

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c. VATER/VACTERL association
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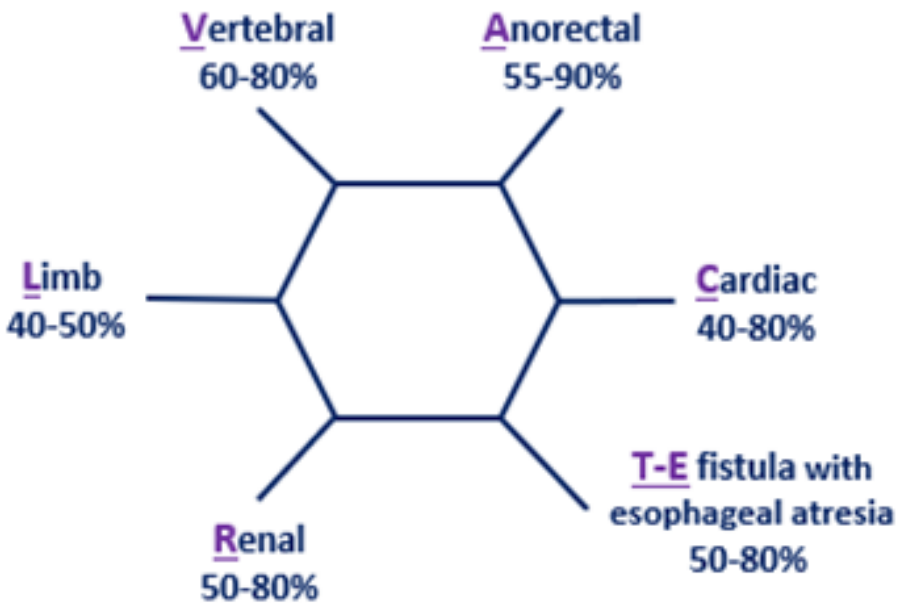


Question 3. Association vs. syndrome vs. sequence

Key point: association is a helpful concept for surveillance and clinical care, and the term has a specific meaning.

An **association** is a non-random occurrence of multiple congenital anomalies. Examples include the **VATER/VACTERL** association, the **OEIS** association (or complex) and the **MURCS** association (not covered here).

A **sequence** is a set of congenital anomalies in which **one of the congenital anomalies can be identified as the upstream cause of the other anomalies**. A classic example is the **spina bifida sequence**, in which the spina bifida itself is thought to be the primary cause of the other anomalies in the sequence – for example, the clubfoot (because of the involvement of the motor nerves of the lower limbs), the Chiari 2 malformation (an anomaly of the lower part of the cerebellum) and of the hydrocephalus (the obstruction of the normal circulation of cerebrospinal fluid). Another example of sequences is the **Pierre Robin sequence** (cleft palate, micrognathia, glossoptosis).

A **syndrome** has a recognized **etiology or cause** (regardless of number of major or minor anomalies) – many syndromes are genetic (e.g., **Down** syndrome due to trisomy 21), but some are environmental (e.g., **congenital rubella syndrome** due to fetal rubella infection).

Combination is **not** a term with a specific meaning in birth defect terminology – it may be used in a child with multiple congenital anomalies that do not qualify as a syndrome, association, or sequence (many children with multiple anomalies are in this category).

4. Renal anomalies can be identified during the following stages of life …
(choose one or more answers)

a. Fetal (e.g., via sonography)
b. Birth and neonatal period
c. Childhood
d. Adulthood

4. Renal anomalies can be identified during the following stages of life … **(choose one or more answers)**

a. fetal (e.g., via sonography)

b. birth and neonatal period

c. childhood

d. adulthood

Comment: renal anomalies can be as severe as bilateral renal agenesis, which is lethal postnatally and can cause fetal anomalies due to lack of amniotic fluid (Potter sequence) or may be asymptomatic for decades, such as unilateral renal agenesis or anomalies of the collecting ducts.
In situations in which the occurrence of renal anomalies is likely increased, systematic screening, such as with a renal ultrasound, is recommended, so that these anomalies can be documented (or excluded) and appropriate clinical management can be implemented. Situations like this may include the presence of other anomalies suggestive of the VATER/VACTERL association or the suspicion of syndromes.

5. In assessing potential **causes** of anomalies in the VATER/VACTERL association, you read about the fetal risks associated with uncontrolled **maternal diabetes**. What are some **adverse outcomes** associated with maternal diabetes?
(Choose one or more answers)

a. Spina bifida
b. Holoprosencephaly
c. Limb deficiencies
d. Sacral agenesis
d. Macrosomia and neonatal hypoglycemia

5. In assessing potential **causes** of anomalies in the VATER/VACTERL association you read about the fetal risks associated with uncontrolled **maternal diabetes**. What are some **adverse outcomes** associated with maternal diabetes? **(choose one or more answers)**

a. spina bifida
b. holoprosencephaly
c. limb deficiencies
d. sacral agenesis
d. macrosomia and neonatal hypoglycemia

5. In assessing potential **causes** of anomalies in the VATER/VACTERL association you read about the fetal risks associated with uncontrolled **maternal diabetes**. What are some **adverse outcomes** associated with maternal diabetes? (**choose one or more answers)**

Key point. Maternal diabetes is an important, common risk factors for many adverse fetal outcomes

Key point: maternal diabetes is an important, common risk factors for many adverse fetal outcomes

Comment: maternal diabetes increases the risk for many birth defects, many adverse fetal outcomes, macrosomia, and neonatal hypoglycemia, all of which increase fetal, neonatal, and childhood mortality.

There are indications that maternal diabetes may also increase the risk for adverse outcomes later in life. In addition, diabetes places the woman at increased risk for many health conditions.

Many professional organizations and public health groups strongly advocate for preventing diabetes. Women with pre-existing diabetes can increase their chances of having a healthy baby by adopting healthy behaviours before and during pregnancy, including:

- seeing a healthcare professional regularly
- keeping blood sugar levels under control, with the help of a doctor and dietician, before and during pregnancy
- getting 400 micrograms (μg) of folic acid every day before and during pregnancy
- striving to reach and maintain a healthy weight before pregnancy.

The main message is to screen and treat before pregnancy – these actions prevent congenital anomalies and improve health of mother and baby.

Facilitator's Guide

Module 7: Data Quality Exercises

World Health Organization

CDC

International Clearinghouse for Birth Defects Surveillance and Research

Objectives

By the end of this module, participants will be able to:

- Identify the key informative elements when presenting a table of prevalence of congenital anomalies, including time, place and definitions of numerators and denominators
- Understand the value of presenting meaningfully disaggregated groupings of congenital anomalies rather than large heterogenous groupings
- Describe the three major components of data quality – completeness, accuracy, timeliness – and how each may influence data quality
- Identify at least five potential methodologic causes of inaccurate birth anomaly prevalence estimates (inaccurate: measured prevalence different from true prevalence)
- Use a fishbone diagram to summarize and categorize potential factors that could influence data quality

Activity 7.1

You are the coordinator of the surveillance programme of the country of Loa. You receive this table of a report from the surveillance programme:

Congenital anomaly	**Number of cases**
Congenital malformations of the nervous system	211
Congenital malformations of the circulatory system	422
Facial clefts	259
Congenital malformations of the musculoskeletal system, not elsewhere classified	188

Q: What changes would you suggest?

Would you recommend adding more information?

Reporting: responses

- These are broad anatomic categories of congenital anomalies (the main headings of ICD-10). For surveillance (and clinical) purposes, you need more specific and disaggregated information. For example:
  - Congenital malformations of the nervous system -> Neural tube defects, spina bifida, anencephaly.
  - Congenital malformations of the circulatory system -> Critical congenital heart defects, tetralogy of Fallot, etc.
  - Facial clefts -> clef lip with or without cleft palate, cleft palate only.
  - Congenital malformations of the musculoskeletal system, not elsewhere classified -> gastroschisis, omphalocele.
- When presenting data on congenital anomalies, it is important to include the ICD-10 codes.
- The number of cases alone is not informative. You need to know the reference population (denominator). Note whether it is population-based vs. hospital-based.
- A good practice is to have a descriptive title for the table, including place and time period.

After you ask for more information, you receive a new table.

Table 1. Congenital anomaly cases in Loa, 2016–2020, among 245,070 births

Congenital anomaly	ICD-10 Code	Cases
Neural tube defects	Q00, Q01, Q05	189
Spina bifida	Q05	124
Anencephaly	Q00	55
Critical congenital heart defects	*	325
Tetralogy of Fallot	Q21.3	59
Transposition of the great arteries	Q20.3	54
Hypoplastic left heart syndrome	Q23.4	50
Abdominal wall defects	Q79.2, Q79.3	171
Gastroschisis	Q79.3	17
Omphalocele	Q79.2	154

*Persistent truncus arteriosus (Q20.0), double outlet right ventricle (Q20.1), D-transposition of great arteries (Q20.3), single ventricle (Q20.4), Tetralogy of Fallot (Q21.3), pulmonary valve atresia (Q22.0), tricuspid atresia (Q22.4), aortic valve stenosis (Q23.0), hypoplastic left heart syndrome (Q23.4), coarctation of the aorta (Q25.1), interrupted aortic arch (Q25.2), total anomalous pulmonary venous return (Q26.2).

Notes: case definitions can also be placed separately in a footnote, in a separate table, or in the methods section – regardless, definitions (e.g., ICD-10 codes with labels) with inclusions and exclusions need to be clearly documented.

Q: What changes would you suggest?

Using the given data, can you add more information?

Reporting: responses

- If you have number of cases and number of births (denominator) you can compute prevalence. For example, prevalence by 10,000 births.
  - Prevalence per 10,000 = cases * 10,000 / total births
  - Denominators also have to be explicitly defined – are these live births, stillbirths, terminations of pregnancy?

Table 1. Congenital anomaly cases in Loa, 2016–2020, among 245,070 births

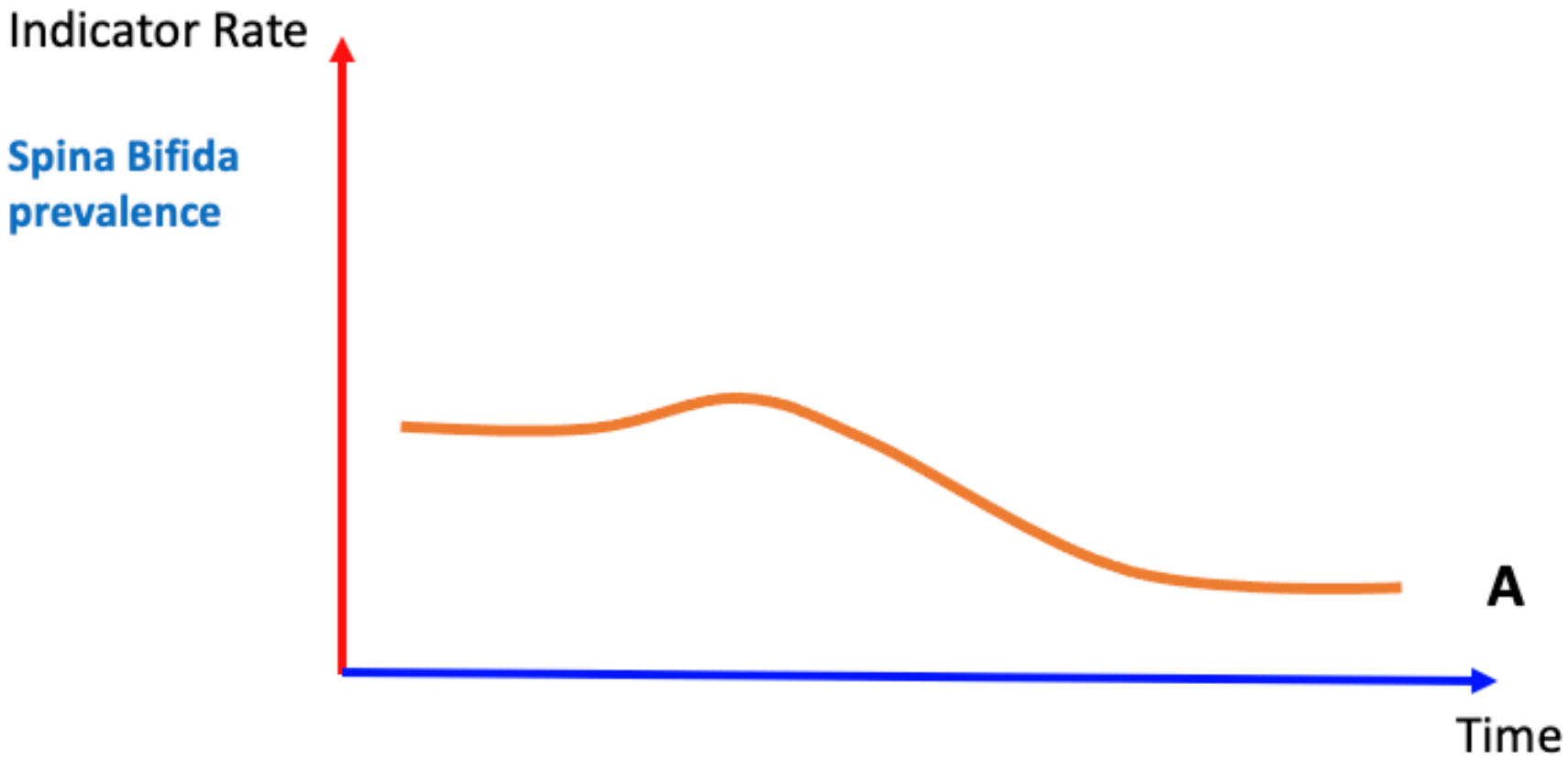
Congenital Anomaly	**ICD-10 Code**	**Cases**	**Prevalence per 10,000 births**
Neural tube defects	Q00, Q01, Q05	189	7.71
Spina bifida	Q05	124	5.06
Anencephaly	Q00	55	2.24
Critical congenital heart defects	*	325	13.26
Tetralogy of Fallot	Q21.3	59	2.41
Transposition of the great arteries	Q20.3	54	2.20
Hypoplastic left heart syndrome	Q23.4	50	2.04
Abdominal wall defects	Q79.2, Q79.3	171	6.98
Gastroschisis	Q79.3	17	0.69
Omphalocele	Q79.2	154	6.28

Prevalence comparisons

Now that you have prevalence figures, you can compare the data of your programme with other programmes. A good practice for comparing prevalence between programmes is to look at the 95% confidence intervals. You notice that the prevalence of spina bifida in Loa is lower than what is reported from a neighboring country, and in fact quite lower than the previous five-year period.

Discuss potential reasons for this low prevalence.

Q: This is what you see. What might be going on?



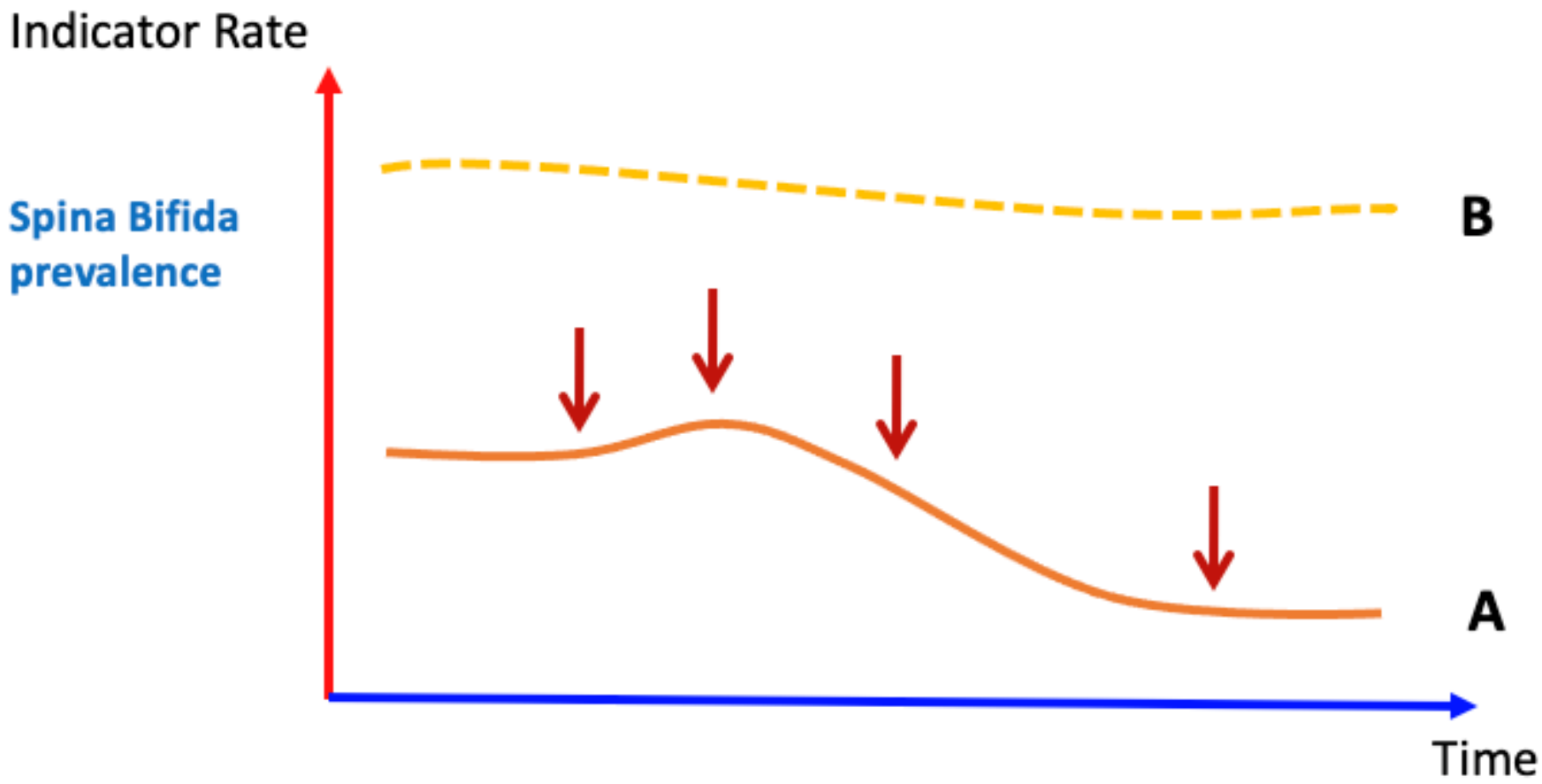
Internal comparison

1. First, think of program methodology: prevalence may be low as an artifact. Once methodology is ruled out, consider biology.

- Discuss considerations when doing an internal comparison of changes over time:
  - Methodology
  - Changes in staff - a trained doctor with experience retiring, lack of training of new staff
  - Changes in reporting sites (adding or dropping facilities)
  - Changes in SOPs
  - Changes in definitions

2. Biology: a new intervention implemented in Loa in the last 5 years: folic acid supplementation or fortification programs

Q: What might be going on in Loa, compared to B?



External comparison

1. First, think of program methodology: prevalence may be low as an artifact.

   Once methodology is ruled out, consider biology.

- Discuss considerations when doing an internal comparison of changes over time. Methodology:
  - How cases are defined; e.g., for spina bifida, consider if the other program includes spina bifida occulta or lipomeningomyelocele (usually excluded)
  - How data are collected, e.g., differences in the basic structure of the programs being compared: hospital- vs population-based, active vs passive, single source vs multiple sources, age of ascertainment
  - Diagnostic resources, e.g., ultrasounds: cardiac defects, renal anomalies
  - Pregnancy outcome inclusion, e.g., are stillbirths and/or pregnancy terminations included in the prevalence estimates?

2. Biology: if Loa has a folic acid fortification and supplementation program, and the neighboring country does not, that may be the reason for the difference.

You want more detailed information to understand data quality. You ask for more information on individual cases of spina bifida. You receive the following descriptions:

- Case 1: open spina bifida
- Case 2: spina bifida with hydrocephalus
- Case 3: myelomeningocele, lumbar, hydrocephalus, talipes equinovarus

Q: What information is missing from these case descriptions?

Accurate description of spina bifida

Checklist for high-quality reporting

Spina Bifida – Documentation Checklist

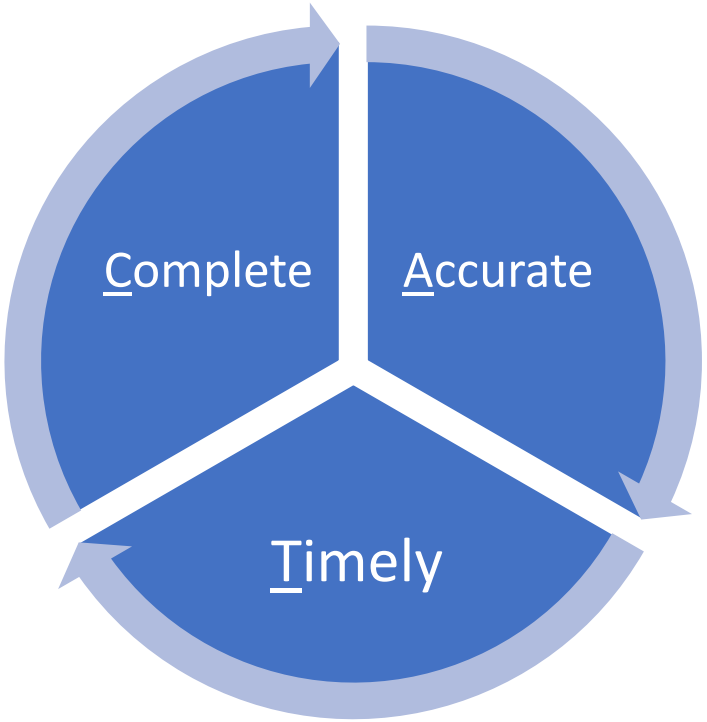
- ☐ **Describe defect in detail:**
  - Location – specify level (e.g. cervical, thoracic, thoraco-lumbar, lumbar, lumbosacral, sacral, etc.).
  - Size of lesion.
  - Covering – covered by skin or not covered by skin.
  - Content – only meninges (meningocele) or also spinal cord (myelomeningocele – spinal cord visible).
  - Anomalies – document sequence defects (hydrocephalus, talipes) and other anomalies.
- ☐ **Take and report photographs:** *Show clearly* the level of spina bifida (back and side if possible); can be crucial for review.
- ☐ **Describe evaluations to find or rule out related and associated anomalies:**
  - Sequence – hydrocephalus, talipes, other.
  - Other unrelated anomalies – describe procedures to assess other anomalies.
  - Genetic or chromosomal conditions.
  - Specialty consultations, imaging and surgery.
- ☐ **Report whether autopsy (pathology) findings are available and if so, report the results.**

Description	What is missing?
Open spina bifida	Information missing on location, size, involvement of meninges +/- spinal cord, related anomalies and other unrelated anomalies
Spina bifida with hydrocephalus	Information missing on location, size, skin covering, involvement of meninges +/- spinal cord, and unrelated anomalies
Myelomeningocele, lumbar; hydrocephalus, talipes equinovarus	This is the most accurate description; information is missing only on size of the lesion

Attributes of data quality

Q: It seems that data quality on spina bifida may be compromised. Please describe the three major components of data quality (completeness, accuracy, and timeliness) and how they may explain the low prevalence of spina bifida in Loa.

3 attributes of data quality



- **Complete:**
  - Inclusive of all cases, all variables
- **Accurate:**
  - reliable disease rates, comparable to others, reflecting standard of case definitions
- **Timely:**
  - timely prevention/linking cases to services, respond to investigations, monitor trends

Timely

Complete
Accurate

Completeness

- All cases are included and all data variables for cases are entered. Assessing completeness speaks directly to the sensitivity of the surveillance program.

- We see that prevalence of spina bifida in Loa is low. A reason for that may be that not all cases (e.g., terminated or stillborn) born are being included in the reports (completeness may be low).

Accuracy

- Information entered reflects the truth (e.g., case of spina bifida is indeed a case of spina bifida). Accuracy may be also affected if cases of spina bifida are reported as a different anomaly, resulting in lower prevalence. For example, cases of spina bifida misclassified as lipomeningomyelocele and excluded when counting spina bifida.

- This misclassification tends to happen more often with other anomalies (e.g., gastroschisis vs. omphalocele). In fact, in Loa we see a high prevalence of omphalocele (6.81 per 10,000 births) and a low prevalence of gastroschisis (1.92 per 10,000 births). The reason for this may be misclassification of gastroschisis cases as omphalocele.

Timeliness

- Data are available and disseminated at the time the program needs them. This is important in public health surveillance because of the focus on ongoing tracking of health events. Timeliness should be defined at the outset so it can be assessed and tracked as a key quality indicator.

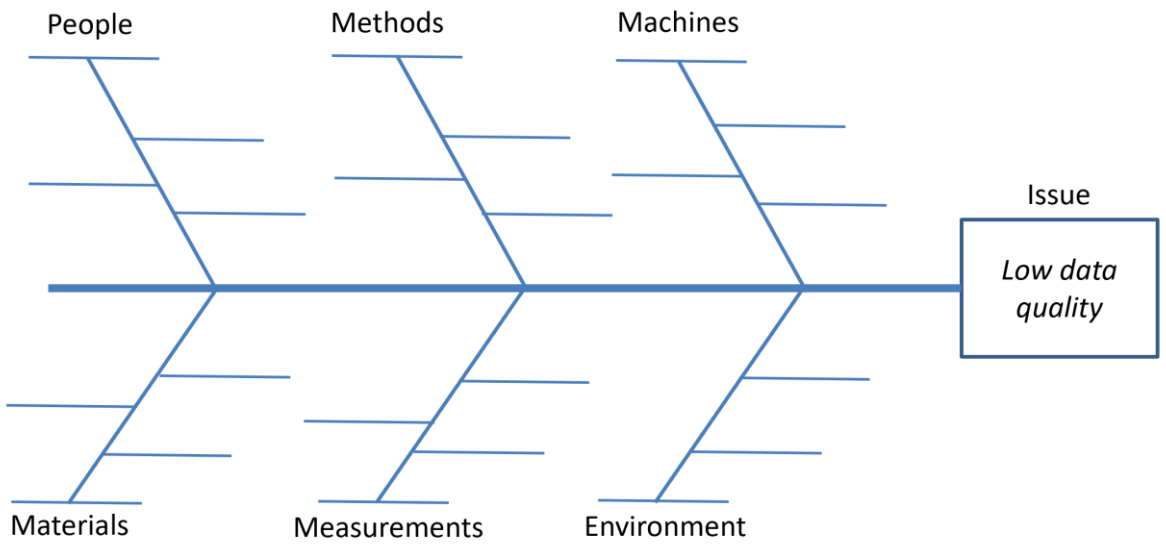
- If cases are not reported on time (for example, a 2020 case of spina bifida is reported in 2023, and therefore not included in the 2016-2020 report), that may also result in low prevalence.

Structured exercise to diagnose what might be causing low prevalence of spina bifida:

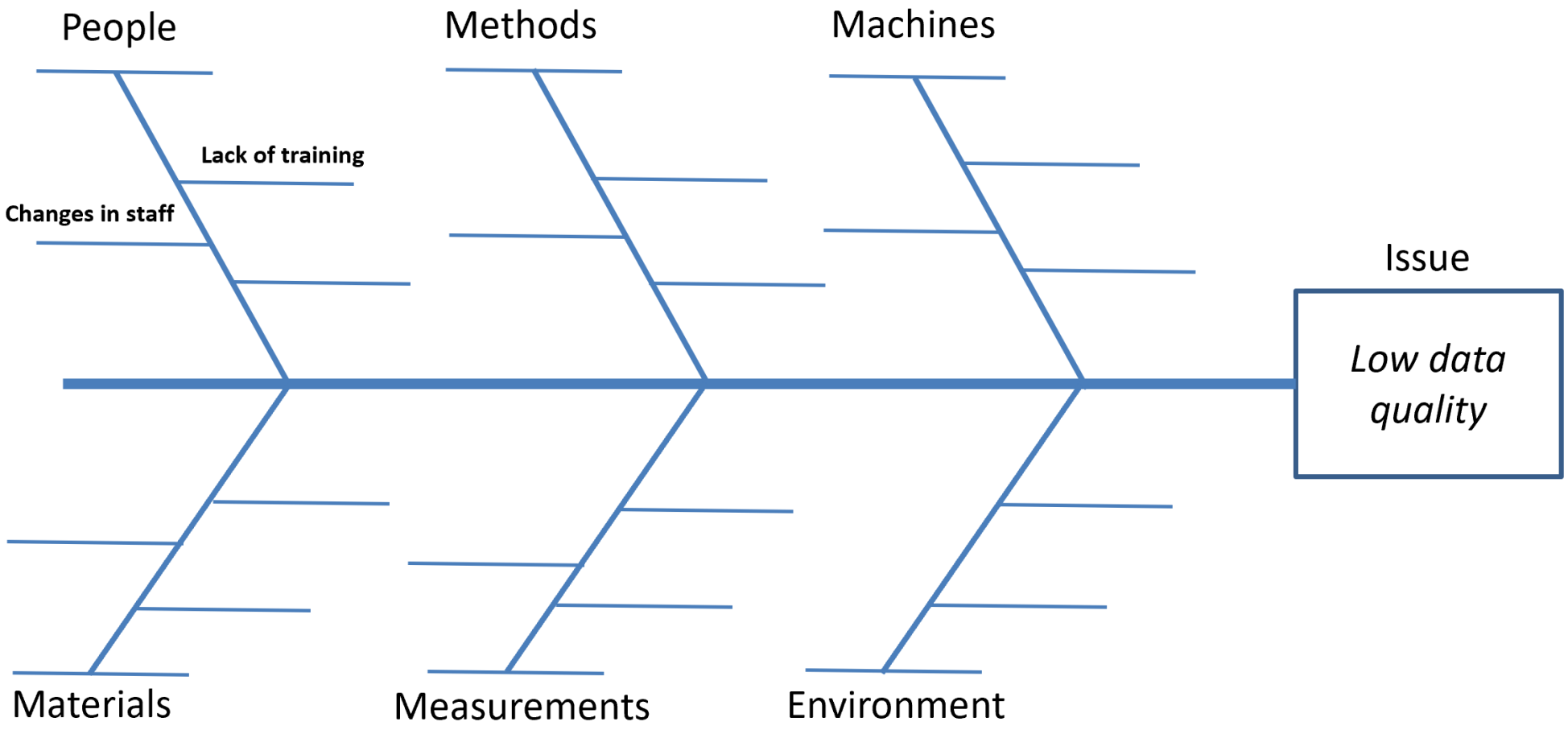
One way to do this is to create a Fishbone diagram that graphically displays an organized list of potential causes of a problem, e.g., low data quality.

To create a Fishbone diagram: draw a box at the right with topic summary (e.g., low data quality). Draw a large horizontal line to the left and add 6 branches. Each branch is a category of causes:

- **People**: involved with the process. Frontline workers, program coordination.
- **Methods**: procedures described in SOPs
- **Machines**: diagnostic equipment, computers, tools
- **Materials**: paper forms, measuring tape for head circumference
- **Measurements**: data analysis, calculation of prevalence, data quality indicators
- **Environment**: the conditions & culture of the reporting site, central coordination



Q: What are potential causes of low data quality (add responses within each category)?

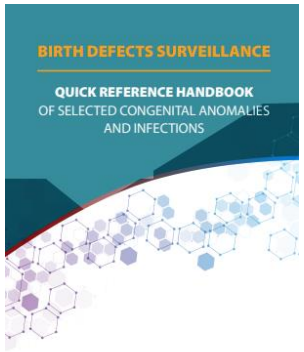


Q: Once you diagnose the potential causes of the issue, you can start with an intervention. How would you improve data quality?

Improving data quality

Response: these are some ideas to improve data quality:

- ✓ Use the QRH
- ✓ Use apps
- ✓ Use checklists
- ✓ Have a manual of Standard operating procedures (SOPs)
- ✓ Have direct contact between central and local teams, and with stakeholders
- ✓ Train the local teams
- ✓ Have champions



Q: Who are the champions of a surveillance system?
Why are they important?

Champions

- A champion is a member of a staff at the site, committed to the program, who helps to ascertain and report cases, improving quality.
- A champion is often a nurse or physician. This leader could train other personnel on how to identify cases and record the information.
- Also, the champion maintains an ongoing quality control of the information.

Champions are critical to the success of a program success, ensuring participation and data quality across units and services.

Take care of your champions, and the champions will take care of you

Quality in data collection

- It is the initial step of the process of birth defects surveillance.
- It is difficult to get results with bad data collection.

Garbage In…

… Garbage Out

“Your analysis is only as good as your data.”

Q: We found out that data quality was low after we looked at the reported data from 2016–2020. What would you do to prevent this from happening again?

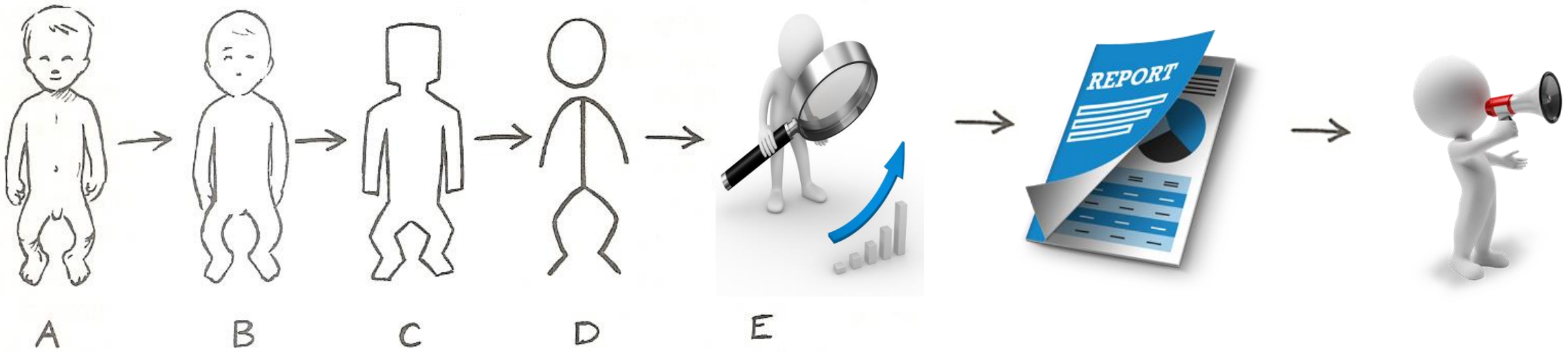
Discuss the difference between quality control and quality improvement.

In this exercise we looked at the data and found issues with data quality. This is a quality control approach: it´s important but retrospective and reactive. It focuses on discovery and detection at the level of the “end product” (e.g., the final data or report). A good quality control approach includes the use of data quality indicators (see http://www.icbdsr.org/data-quality-indicators-tool/).

Quality control finds the flaws in the end product but does not fix them: in fact, it may not necessarily identify where in the process the data quality issues arise. Quality control can trigger quality improvement.

Quality improvement proactively focuses on preventing data quality issues, typically by identifying the steps in the surveillance programme where quality issues are generated. Effective quality improvement builds (“bakes”) high quality into each process of the surveillance system, and often involves rethinking, tweaking and redesigning some step of the process. Every process in your surveillance system should add value, otherwise it is a waste.

Quality in every process of the system



Newborn	Description	Coding	Classification	Analysis	Report	Dissemination

Adapted from Bengt Kallen, Epidemiology of Human Reproduction, CRC 1988

Take-home message

- **Integrate surveillance activities into the way professionals work** in their institutions
- Assure quality, from data collection to dissemination
- **Use champions**: motivate those who report by providing clinical information
- Collect data and USE IT: the **use** of timely delivered **results** is an essential part of a good programme