Birth defects surveillance training

Facilitator's guide

Second edition

Participant workbook

World Health Organization

CDC
U.S. CENTERS FOR DISEASE CONTROL AND PREVENTION

International Clearinghouse for Birth Defects Surveillance and Research

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Birth defects surveillance training: facilitator's guide, second edition. Participant workbook

ISBN 978-92-4-009991-3 (electronic version)
ISBN 978-92-4-009992-0 (print version)

Suggested citation. Birth defects surveillance training: facilitator's guide, second edition. Participant workbook. Geneva: World Health Organization; 2024. Licence: CC BY-NC-SA 3.0 IGO.

Cataloguing-in-Publication (CIP) data. CIP data are available at https://iris.who.int/.

Design and layout by Green Ink Publishing Services Ltd.

Contents

This participant workbook accompanies the *Birth defects surveillance training: facilitator's guide, second edition*, available at https://iris.who.int/handle/10665/379508

Module 2

Introduction to planning activities and tools

Activity 2.1

Use the table below to complete a logic model for development of a congenital anomalies surveillance programme in your country.

Resources	Activities	Outputs	Short- and long-term outcomes	Impact
Need the following resources in order to accomplish activities:	Need to accomplish the following activities in order to address the problem:	Once activities are accomplished, expect to have the following product(s) or services:	If activities are accomplished they will lead to the following changes in 1–3 years:	If activities are accomplished, they will lead to the following changes in 4–6 years:

Activity 2.2

- Give examples of partners in your country who will be important to include in the planning and implementation of a surveillance programme.
- Consider some ways partners might help a surveillance programme to succeed.

Activity 2.3

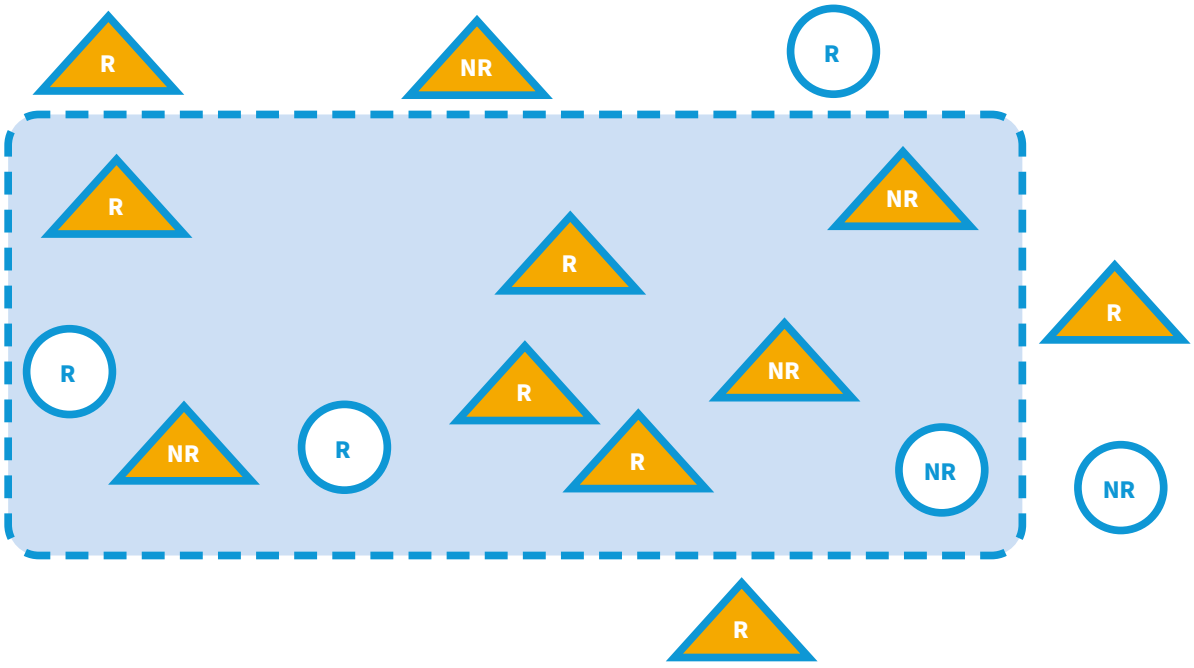
Use the table below to complete a stakeholder's worksheet for development of a congenital anomalies surveillance programme in your country.

Likely users of outputs	Communication message	Dissemination strategy	Evaluation
Ministries of health			
Hospitals and, if relevant, hospital associations and clinics			
Champions			
Community health workers/community health volunteers			
Congenital anomalies associations, foundations and other nongovernmental organizations			
International organizations			
Medical schools/ research agencies			

Module 3

Introduction to surveillance approaches

Activity 3.1



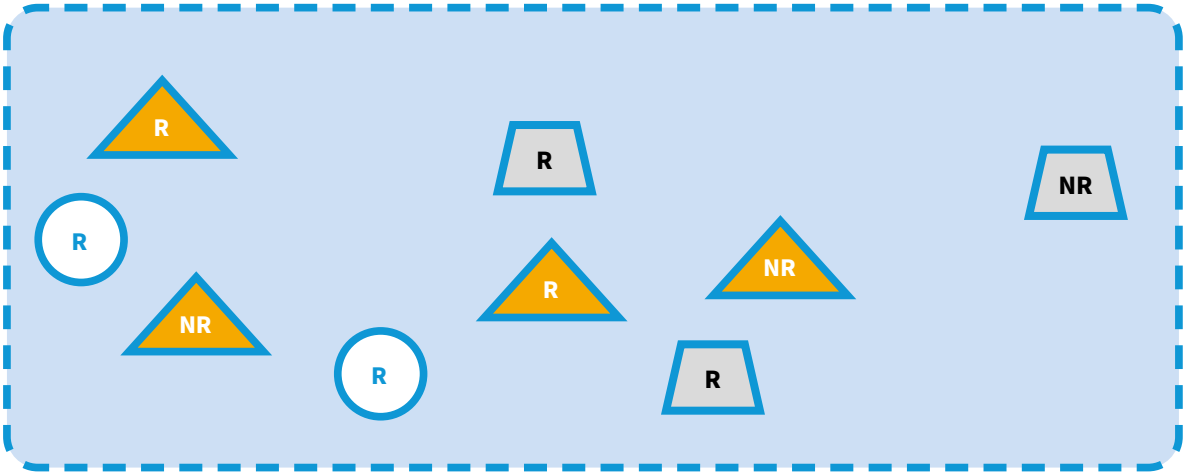
Geographic boundary defining residence (catchment area)

Hospital, clinic or treatment facility

Home births

NR: non-resident, i.e. fetus or neonate with congenital anomaly whose mother is a non-resident;
R: resident, i.e. fetus or neonate with congenital anomaly whose mother is a resident.

- Questions
  - Does the figure represent a population-based or hospital-based surveillance programme?
  - What is the numerator (cases that should be registered) in this surveillance programme?
  - Is maternal residence important for this type of surveillance?
  - Are home births with congenital anomalies counted in this type of surveillance?



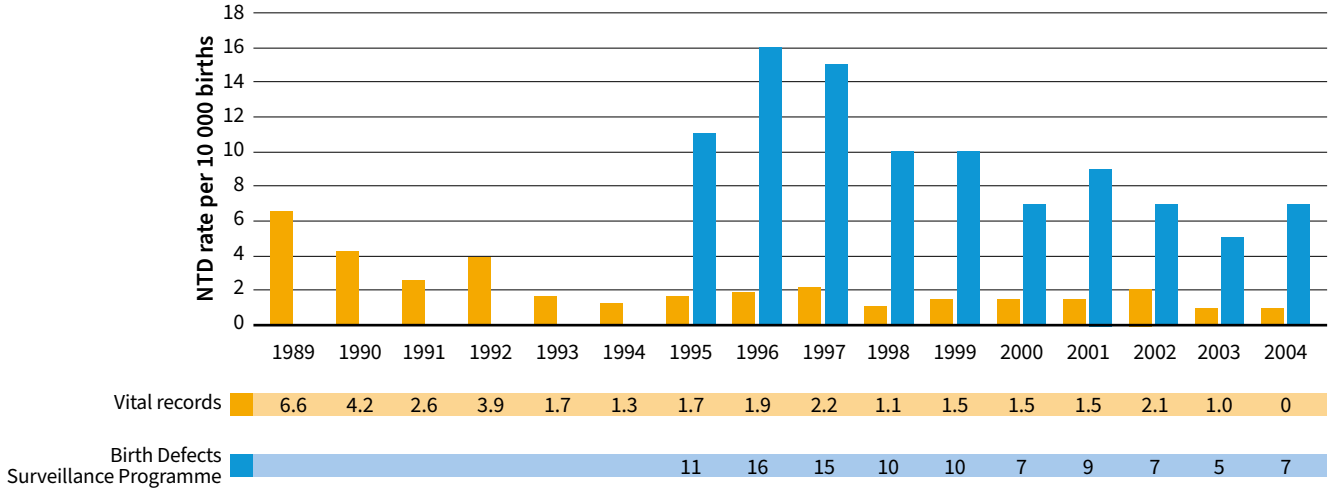
Geographic boundary for participating hospitals | Participating hospitals | Home births | Non-participating hospitals

NR: non-resident, i.e. fetus/neonate with a birth defect whose mother is a non-resident;
R: resident, i.e. fetus/neonate with a birth defect whose mother is a resident.

- Questions
  - Does the figure represent a population-based or hospital-based surveillance programme?
  - What is the numerator (cases that should be registered) in this surveillance programme?
  - Is maternal residence important for this type of surveillance?
  - Are home births with congenital anomalies counted in this type of surveillance?

Activity 3.2

Study the figure below.



What does this figure demonstrate?

Activity 3.3

Create inclusion and exclusion criteria for population-based or hospital-based surveillance programmes. Keep in mind capacity and available data sources. Remember that inclusion and exclusion criteria will be different, depending on whether the programme is hospital or population based.

Activity 3.4

Answer the following questions:

- What defines a case?
- If only live births are going to be counted in your surveillance programme, do you include an infant who was alive during labour but at delivery shows no sign of life? Explain your response.
- If a mother delivers a neonate with a congenital anomaly on her way to the hospital and the surveillance programme is hospital based, would the neonate be included in the programme? Explain your response.
- If a mother delivers at home and the neonate is brought to the hospital within hours of delivery because a congenital anomaly has been noticed, would the neonate be included in a hospital-based surveillance programme? Explain your response.
- If a mother delivers a neonate with congenital anomalies in a hospital in your country, but she is from another country or not a resident of your catchment area, would her neonate be included in a population-based programme? What about a hospital-based programme? Explain your response.
- If a mother, born in another country, who has been living in your country for three months, delivers a neonate with a congenital anomaly in a hospital in your country, would her neonate be included in a population-based programme? What about a hospital-based programme? Explain your response. What if she has lived in your country for at least one year? Would this make a difference? Explain your response.
- If your surveillance programme is collecting information during the first three days of life, and an infant with a congenital anomaly is identified in the paediatric ward at 1 month of age, would the infant be included? What if the child is identified at 2 years of age? Explain your response.
- How would the exclusion of stillbirths affect your prevalence estimate? For which congenital anomalies would it be important to include stillbirths?
- In a hospital-based surveillance programme, if a neonate with a congenital anomaly is born alive by caesarean section in the obstetric operating theatre/room and not in the delivery room, and your only data source of information in the hospital is the delivery room log, would the neonate be counted in the programme? Explain your response.
- If a neonate with anencephaly weighs 600 g, but your inclusion criteria define weight as 1000 g or more, would the neonate be included in the surveillance programme? Explain your response.
- If a fetus is identified prenatally with a congenital anomaly, and examination at birth shows that the congenital anomaly is not present, would the fetus be included in the surveillance programme? Explain your response.
- Explain how the inclusion and exclusion criteria can change the birth prevalence estimate.

Activity 3.5

Study the images below.

Verbatim description format

Selected congenital anomaly	Description/comments/details
1. Cleft lip	Baby born with unilateral, left cleft lip; palate is intact. Baby also has microcephaly and clenched hands.

Checkbox format

Neural tube defects:
☐ Anencephaly ☐ Encephalocele ☐ Spina bifida
Orofacial clefts:
☒ Cleft lip ☐ Cleft palate ☐ Cleft lip and palate
☒ Other

What do these examples demonstrate?

Activity 3.6

- Review the table below and consider the suggested core ascertainment variables.

Category	Variable name	Why this variable should be collected
Report	Case record identification	
	City, province, state or territory	
Father	Name(s)	
Mother	Name(s)	
	Mother's date of birth, or age if date of birth is not available	
	Total number of pregnancies	
Infant	Date of birth	
	Sex	
	Outcome at birth	

Activity 3.7

- Review the form below, and consider which variables you would add or delete, and why.

Birth Defects Surveillance Programme

Case record ID: Date of report: (dd/mm/yyyy)	Name of health facility: City: Province/State/Territory:

FETUS / NEONATE

Name, if available:

Date of birth: (dd/mm/yyyy) Date of diagnosis of congenital anomaly: (dd/mm/yyyy)

Sex:
◯ male ◯ female ◯ ambiguous ◯ missing/unknown

Outcome at birth:
◯ live birth ◯ stillbirth
◯ elective termination of pregnancy with fetal anomaly

Gestational age: (completed weeks)

Best estimation: ultrasound: LMP: other:

Weight: (grams) Length: (cm)

Head circumference: (cm)

Multiple birth: ◯ Yes ◯ No If yes, specify:

Photographs taken: ◯ Yes ◯ No

Did neonate die? ◯ Yes ◯ No

If yes, specify date of death: (dd/mm/yyyy)

Cause of death:

Autopsy: ◯ Yes ◯ No If yes, specify details on back of this sheet.

PARENTS

Father's given name(s):

Father's family name(s):

Father's date of birth: (dd/mm/yyyy) Father's age: (completed years)

Race/ethnicity:

Mother's given name(s):

Mother's family name(s) (including maiden name):

Mother's date of birth: (dd/mm/yyyy) Mother's age: (completed years)

Race/ethnicity:

Primary address during 1st trimester of pregnancy:

Town/city: Province:

Current address (If different from above):

Town/city: Province:

Telephone number:

Total number of previous: live births: stillbirths:

spontaneous abortions: terminations of pregnancy:

Are parents of fetus/neonate related? ◯ Yes ◯ No
If yes, specify: ◯ first cousins ◯ second cousins ◯ aunt – nephew ◯ uncle – niece ◯ other (specify):

Congenital anomaly present	Full description of congenital anomaly (use back of form if needed)	ICD-10 code	C or P*
1.			◯ C ◯ P
2.			◯ C ◯ P
3.			◯ C ◯ P
4.			◯ C ◯ P
5.			◯ C ◯ P
6.			◯ C ◯ P
7.			◯ C ◯ P
8.			◯ C ◯ P
9.			◯ C ◯ P
10.			◯ C ◯ P

Diagnostic tests performed, pending results, notes and comments:

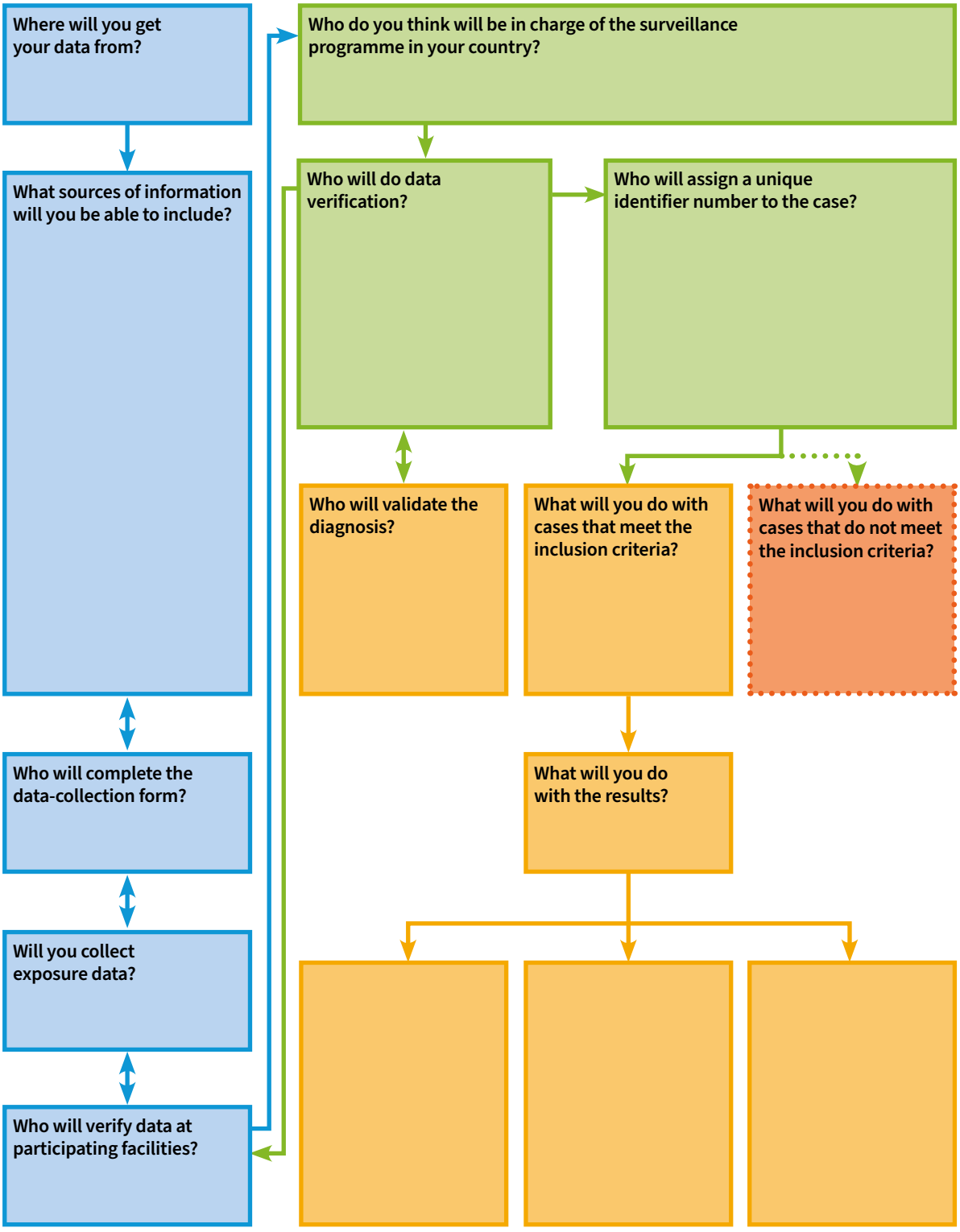
* C = Confirmed diagnosis
P = Possible diagnosis

Name of professional completing the form: Contact information:
◯ physician ◯ nurse ◯ other (specify):

Version: January 2014

Activity 3.8

Complete the flow chart.



Activity 3.9

Read the case example below.

Case example: Cases of neural tube defects by type of ascertainment, United States of America (USA), 2010–2014

The United States National Birth Defects Prevention Network collects state-specific congenital anomalies surveillance data for annual publication of prevalence estimates and collaborative research projects. In 2019, national estimates were presented for 29 major birth defects using data from 2010–2014. The data presented in the table below are from population-based programmes that have different types of case ascertainment: active, hybrid and passive. Active ascertainment occurs when there is active review of multiple data sources to identify cases. Active ascertainment usually requires that the programme hires trained personnel to conduct abstraction from data sources. Passive ascertainment occurs when hospital staff report cases directly to the programme without verification of cases by the programme staff. An example of hybrid ascertainment is when hospital staff report cases and programme staff verify them.

Cases of neural tube defects by type of ascertainment, USA, 2010–2014

	Number of cases			
Neural tube defects	Active ascertainment[a]	Hybrid ascertainment[b]	Passive ascertainment[c]	National
Anencephaly	1306	379	466	2151
Spina bifida	2082	1293	1241	4616
Encephalocele	559	326	285	1170
Total neural tube defects	3947	1998	1992	7937

[a] N=16 programmes; number of live births = 5 502 807

[b] N=10 programmes; number of live births = 4 792 252

[c] N=13 programmes; number of live births = 3 528 713

Source: Mai CT, Isenburg JL, Canfield MA, Meyer RE, Correa A, Alverson CJ et al. National population-based estimates for major birth defects, 2010–2014. Birth Defects Res. 2019;1–16 (https://doi.org/10.1002/bdr2.1589).

Questions

- Estimate the national prevalence for each neural tube defect and for the total neural tube defects per 10 000 live births.
- Estimate the birth prevalence for each neural tube defect per 10 000 live births by type of ascertainment.
- Estimate the birth prevalence for total neural tube defects per 10 000 live births by ascertainment.
- Enter your prevalence estimates in the table below.

Cases of neural tube defects by type of ascertainment, USA, 2010–2014

	Active ascertainment[a]		Hybrid ascertainment[b]		Passive ascertainment[c]		National	
Neural tube defects	**Cases**	**Prevalence**	**Cases**	**Prevalence**	**Cases**	**Prevalence**	**Cases**	**Prevalence**
Anencephaly	1306		379		466		2151	
Spina bifida	2082		1293		1241		4616	
Encephalocele	559		326		285		1170	
Total neural tube defects	3947		1998		1992		7937	

[a] N=16; number of live births = 5 502 807

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Questions

- Describe the differences in prevalence by ascertainment method and provide some reasons for why differences might exist.
- What are some possible reasons why the three ascertainment methods have different prevalence estimates for spina bifida?

Activity 3.10

Read the case study below.

Case study: Pre- and post-fortification birth prevalence of neural tube defects in the USA, 1999–2011

In 1996, folic acid fortification of cereal grain products labelled as enriched became voluntary in the USA. In 1998, a mandate was passed requiring that these products be fortified with folic acid, to ensure an adequate supply of folate for women of childbearing age.

The United States National Birth Defects Prevention Network collects information on neural tube defects by three major race/ethnic groups and has data from the time period prior to mandatory folic acid fortification and following the folic acid fortification mandate. The estimated annual prevalence of neural tube defects for 19 population-based birth defects surveillance programmes in the USA during these time periods is presented in the table below.

Prevalence of neural tube defects in the USA per 10 000 live births by race/ethnicity (1995–2011)

Race/ ethnicity	Year																
	1995	1996	1997	1998	1999	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011
Caucasian	7.1	7.8	6.6	5.5	5.4	5.3	5.1	4.5	4.6	5.1	4.6	4.9	5.3	4.4	5.2	4.7	4.8
Black	4.9	5.8	3.6	4.8	4.8	4.5	4.8	5.2	4.2	3.7	3.9	3.4	3.8	4.8	4.1	3.5	3.7
Hispanic	9.2	10.8	9.6	7.4	7.8	6.4	6.6	6.9	6.8	6.5	6.2	5.7	6.0	6.4	6.9	7.1	6.5

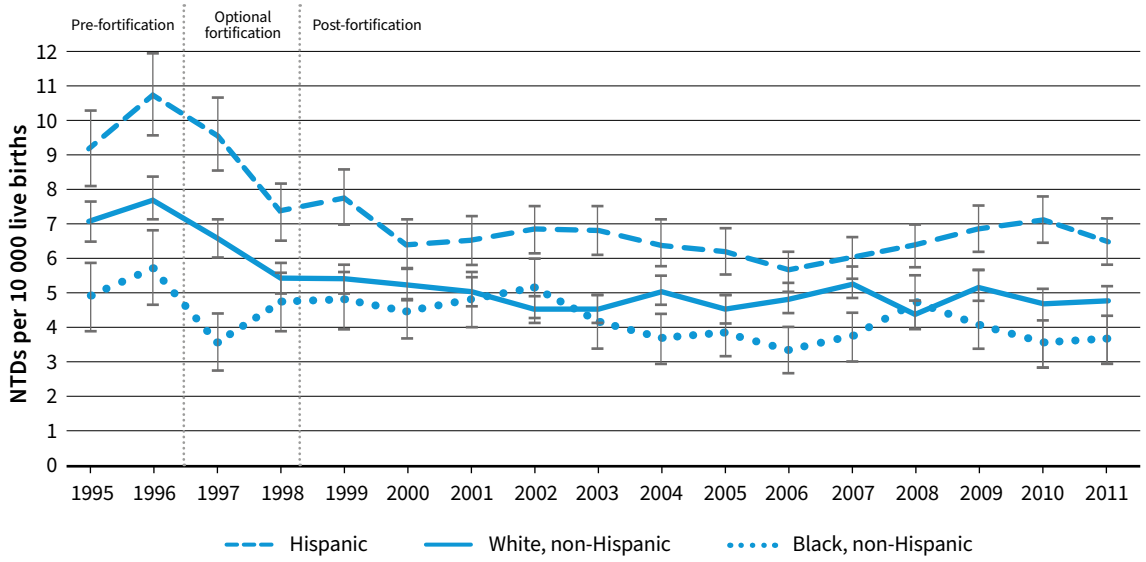
Source: Williams J, Mai CT, Mulinare J, Isenburg J, Flood TJ, Ethen M et al. Updated estimates of neural tube defects prevented by mandatory folic acid fortification — United States, 1995–2011. MMWR Morb Mortal Wkly Rep. 2015;64:1–5 (https://www.cdc.gov/mmwr/preview/mmwrhtml/mm6401a2.htm).

Questions

- Has folic acid fortification of staple foods impacted the prevalence of neural tube defects?
- If so, how has it impacted the prevalence of neural tube defects?
- If you have a computer and access to Excel or other spreadsheet program, make a graph with the data provided.

- Public health agencies have a long tradition of monitoring trends in rates of disease and death, and in medical, social and behavioural risk factors that may contribute to these adverse events. Trends in observed rates provide information for needs assessment, programme planning, programme evaluation, and policy development activities. Examining data over time also allows predictions to be made about future frequencies and rates of occurrence.
- Typically in public health, trend data are presented as population-based rates. These data are accessed from large database systems and show how rates change over relatively long periods of time (e.g. 10 years or more). Trend data can be visually presented through tables and graphs. The figure below shows secular trend data for the prevalence of neural tube defects in the USA by race/ethnicity.

Prevalence of neural tube defects (per 10 000 births) by maternal race/ethnicity, USA, 1995–2011



Source: Williams J, Mai CT, Mulinare J, Isenburg J, Flood TJ, Ethen M et al. Updated estimates of neural tube defects prevented by mandatory folic acid fortification — United States, 1995–2011. MMWR Morb Mortal Wkly Rep. 2015; 64:1–5 (https://www.cdc.gov/mmwr/preview/mmwrhtml/mm6401a2.htm).

Questions

- Describe the prevalence of neural tube defects and the secular (long-term) trend. Is there a change in the prevalence of neural tube defects? What is the direction of the change?
- When was this change first evident?
- What are some possible reasons for some of the changes observed in the prevalence of neural tube defects?
- What are some factors that could impact the prevalence of a health condition?

Activity 3.11

Using the sample surveillance data provided for Activity 3.9, discuss how you would communicate and disseminate the surveillance data information to your assigned group. The groups are given below.

Target audience

- Group 1: Nongovernmental organization
- Group 2: Clinic/public health practitioners
- Group 3: General public
- Group 4: Policy-makers

Activity 3.12

You are a group of paediatricians working in a large maternity facility in your country. You are seeing many babies (see table below) with congenital anomalies being born in the facility and the group thinks it would be good to provide information to your target audience (assigned), to interest them in supporting a surveillance programme.

- In the letter, you should include a description of how the data will be organized, which data will be collected and how they will be presented to make the case to your target audience.
- Using the sample surveillance data in the table below, draft an advocacy letter requesting support for a local congenital anomalies surveillance programme to your assigned target audience.

Target audience

- Groups 1 and 2: Ministry of health (government agency)
- Groups 3 and 4: Clinic/public health practitioners (from other maternity facilities within the country)

Birth prevalence of congenital anomalies by race/ethnicity per 10 000 live births

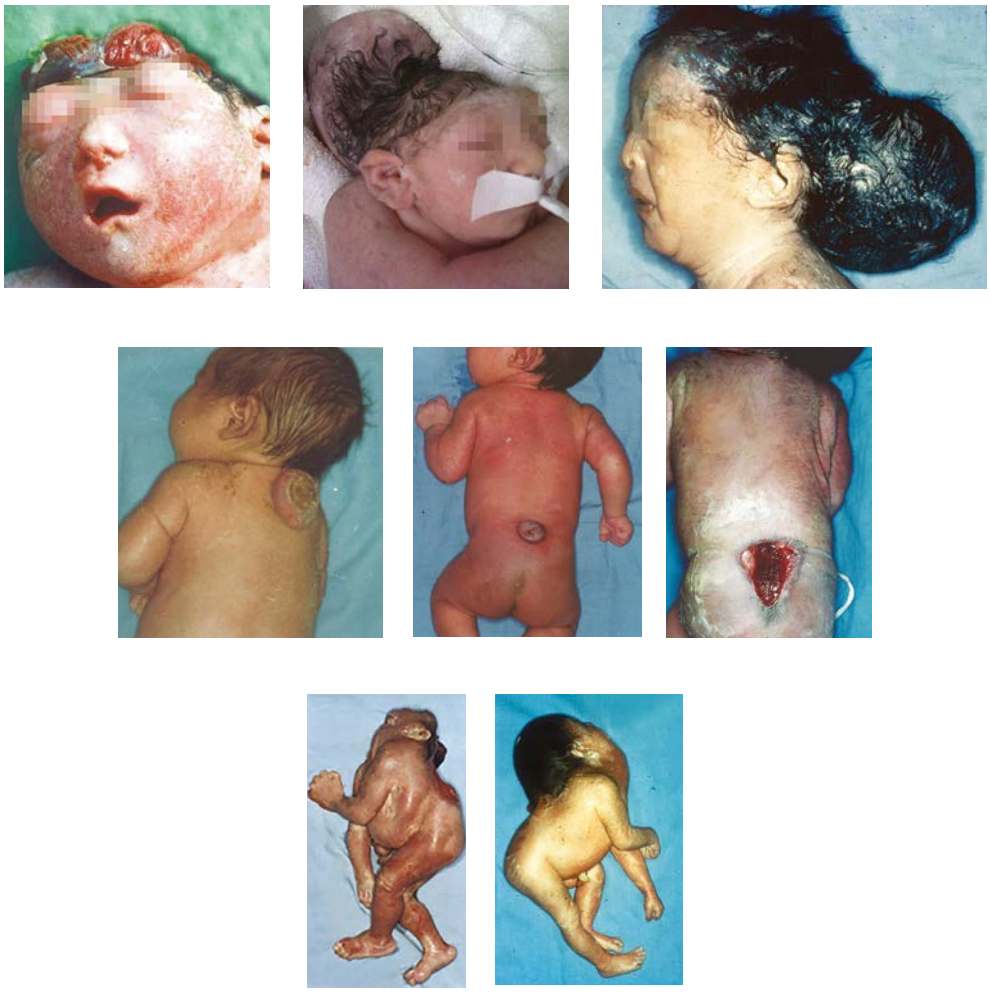
	Ethnic group 1	Ethnic group 2	Ethnic group 3
Cleft lip	243 (10.59)	136 (6.19)	91 (11.28)
Spina bifida	76 (3.31)	53 (2.41)	35 (4.34)
Anencephaly	40 (1.74)	30 (1.37)	21 (2.60)
Encephalocele	19 (0.83)	31 (1.41)	9 (1.12)

Module 4

Introduction to surveillance of congenital anomalies

Activity 4.1

Compare the photos of the different types of neural tube defects below (anencephaly, encephalocele, spina bifida craniorachischisis and iniencephaly). Using the information in the *Birth defects surveillance: quick reference handbook of selected congenital anomalies and infections* (QRH),[1] describe the key findings using the documentation checklists, and then describe the differences between these conditions.



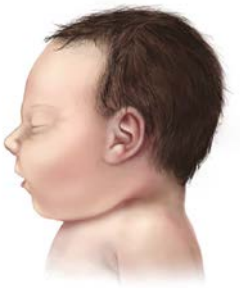
Photograph source: © Latin American Collaborative Study of Congenital Malformations; CDC–Beijing Medical University collaborative project; Dr Jaime Frías.

1 World Health Organization, Centers for Disease Control and Prevention and International Clearinghouse for Birth Defects Monitoring Systems. Birth defects surveillance: quick reference handbook of selected congenital anomalies and infections. Geneva: World Health Organization; 2020 (https://apps.who.int/iris/handle/10665/338485). Licence: CC BY-NC-SA 3.0 IGO.

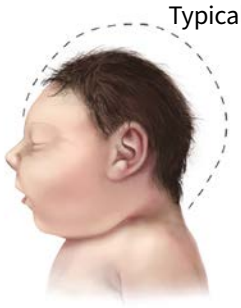
Activity 4.2

Compare photos of a normal head size, microcephaly and severe microcephaly and describe how to properly measure the head circumference. Using the information in the QRH, including the documentation checklist, describe the key findings.

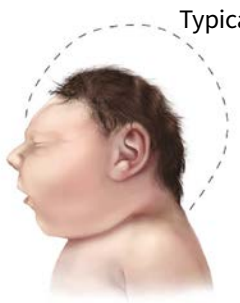
Baby with typical head size



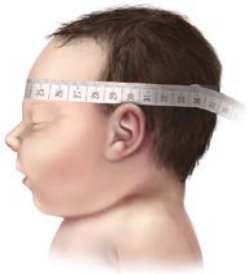
Baby with microcephaly



Baby with severe microcephaly

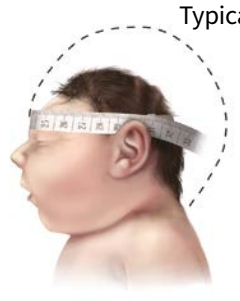


Baby with typical head size



Baby with microcephaly

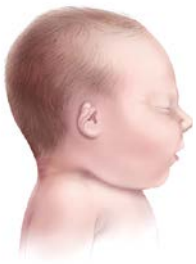
Baby with severe microcephaly



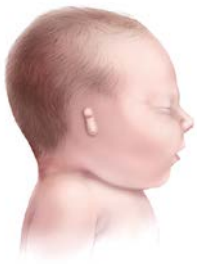
Normal infant ear

Microtia I

Microtia II



Microtia III

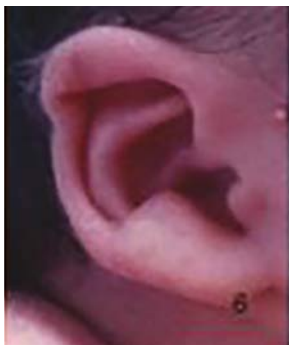


Microtia IV or Anotia

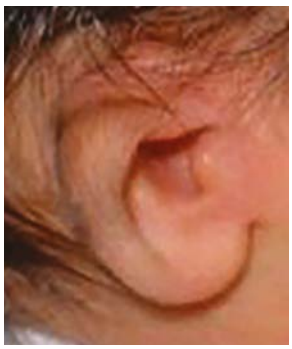
Activity 4.3

Compare photos of the four types of microtia (microtia I, II, III and IV-anotia). Using the information in the QRH, including the documentation checklist, describe the key finding of each type of microtia (I–IV).

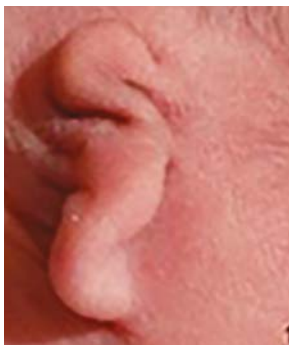
Microtia I



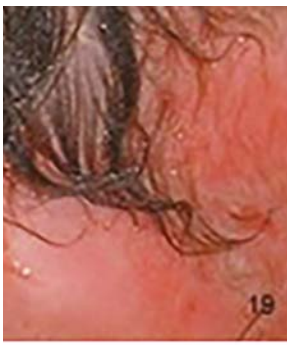
Microtia II



Microtia III



Microtia IV



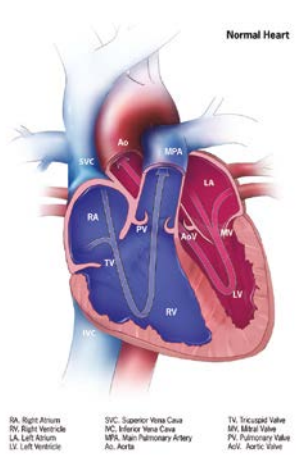
Photograph source: © Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC).

Activity 4.4

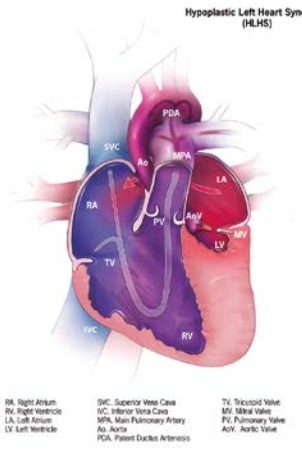
Compare the normal cardiac structures with the two cardiac abnormalities (hypoplastic left heart syndrome and interrupted aortic arch). Using the information in the QRH, including the documentation checklist, describe the key finding of each type of congenital heart defect in the figure below.

Normal and congenital heart defects

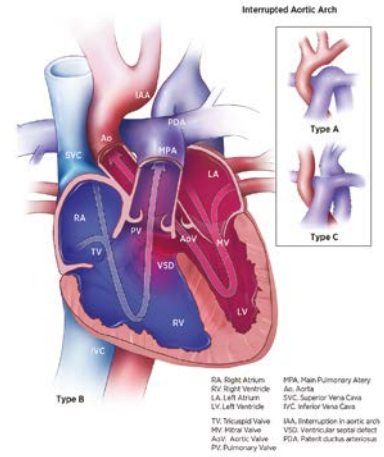
Normal heart



Hypoplastic left heart syndrome



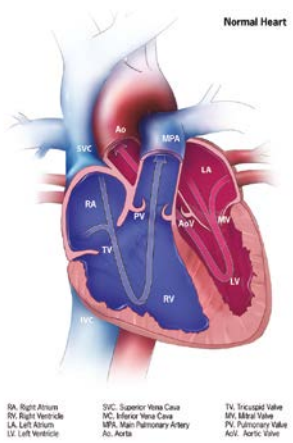
Interrupted aortic arch



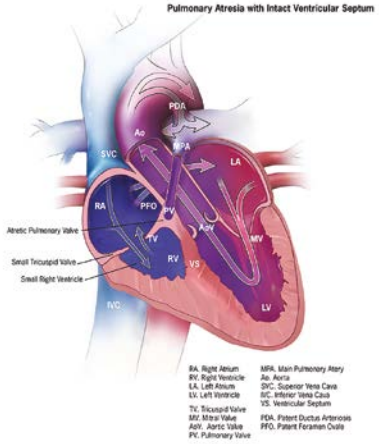
Compare the normal cardiac structures with the two cardiac abnormalities (pulmonary and tricuspid valve atresia). Using the information in the QRH, including the documentation checklist, describe the key finding of each type of congenital heart defect in the figure below.

Normal and abnormal cardiac structures

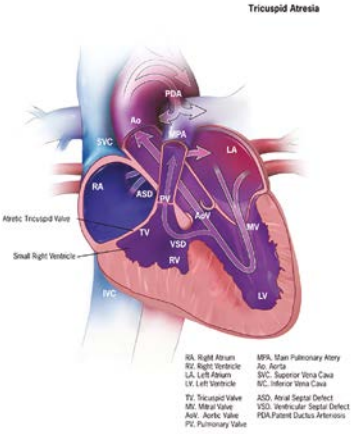
Normal heart



Pulmonary atresia with intact ventricular septum



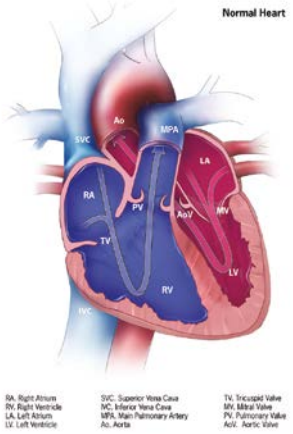
Tricuspid atresia



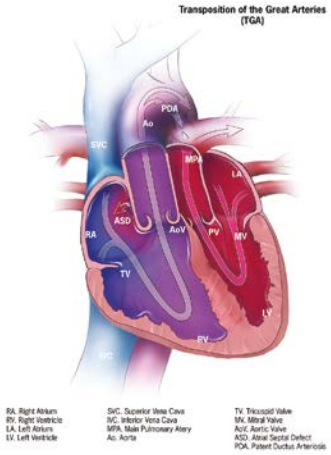
Compare the normal cardiac structures with the three cardiac abnormalities (tetralogy of Fallot, transposition of the great arteries, common truncus). Using the information in the QRH, including the documentation checklist, describe the key finding of each type of congenital heart defect in the figure below.

Normal and abnormal cardiac structures (transposition of the great arteries, tetralogy of Fallot, common truncus)

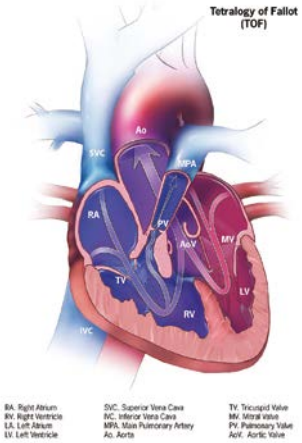
Normal heart



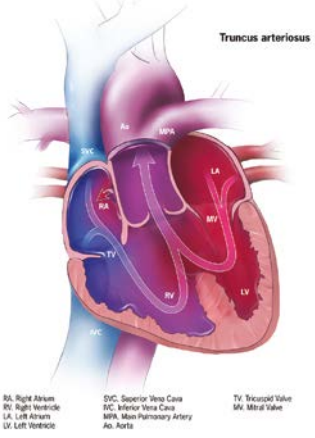
Transposition of the great arteries



Tetralogy of Fallot



Common truncus (truncus arteriosus)

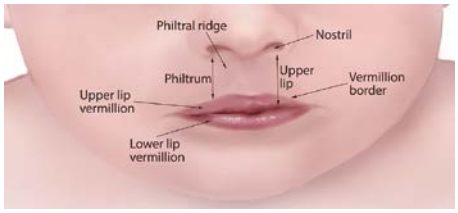


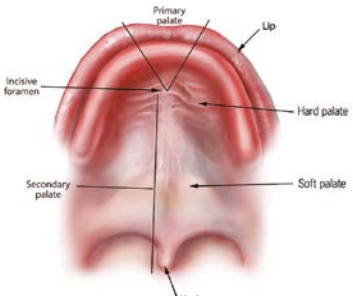
Activity 4.5

Orofacial clefts

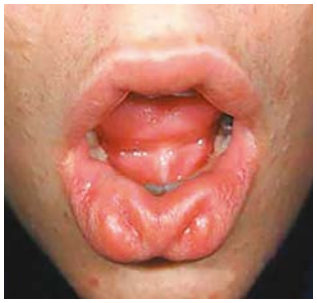
Compare the normal lip and palate anatomy to the different types of orofacial clefts (cleft palate, cleft lip, and cleft palate with cleft lip). Using the information in the QRH, including the documentation checklist, describe the key findings of each type of orofacial cleft below.

Anatomy of the lip and palate

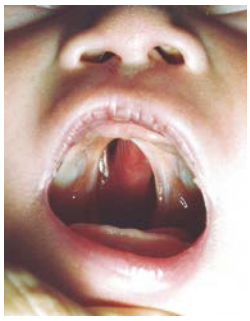


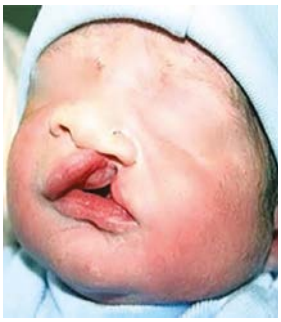


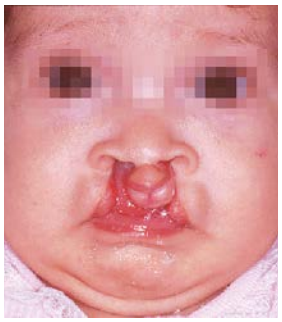
Lip pit

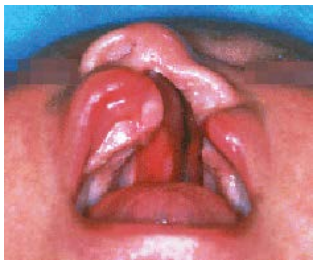


Orofacial clefts









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

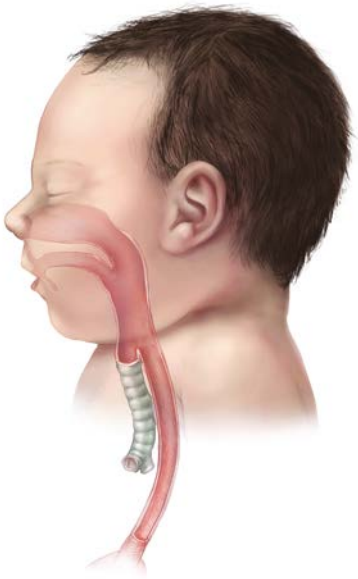
Activity 4.6

Digestive system anomalies

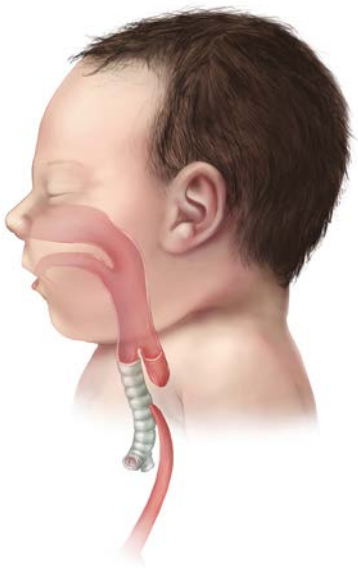
Compare photos of normal structures to the different types of oesophageal atresia/tracheo-oesophageal fistula (see the figure below). Using the information in the QRH, including the documentation checklist, describe the key finding of each type of oesophageal atresia/trachea-oesophageal fistula.

Normal oesophagus, trachea and oesophageal atresia

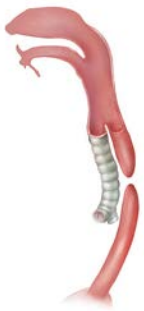
Normal oesophagus and trachea



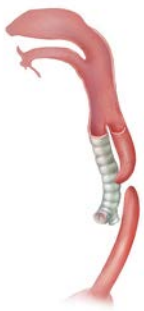
Oesophageal atresia, type C



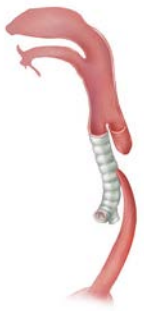
Type A



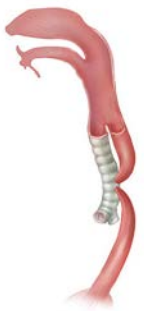
Type B



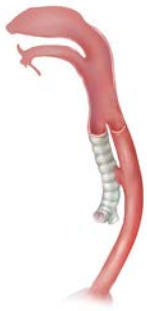
Type C



Type D



Type E

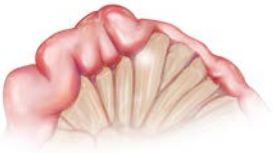


Compare photos of normal structures to the different types of large intestinal atresia (see the figure below). Using the information in the QRH, including the documentation checklist, describe the key finding of each type of large intestinal atresia/stenosis.

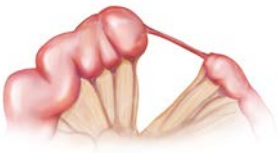
Large intestinal atresia

Large intestinal atresia

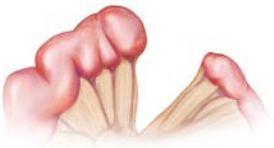
Type 1



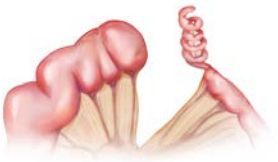
Type 2



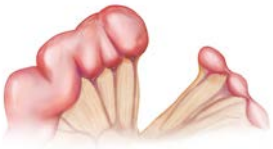
Type 3a



Type 3b

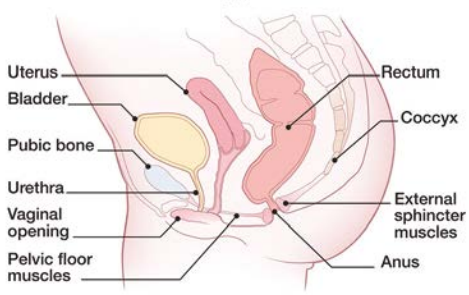


Type 4

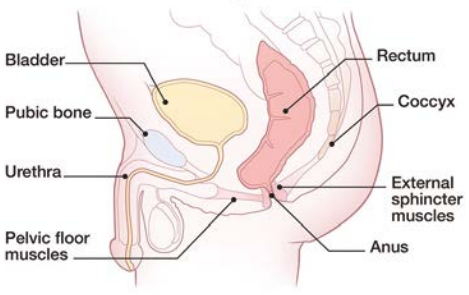


Compare the normal female and male anatomy to the abnormal structures for both anorectal atresia and stenosis. Using the information in the QRH, including the documentation checklist, describe the key finding in females and males.

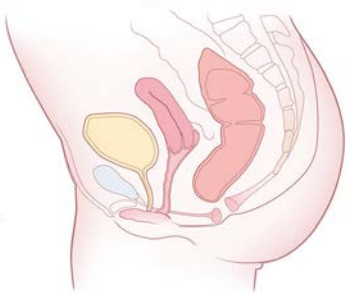
A. Normal anatomy female



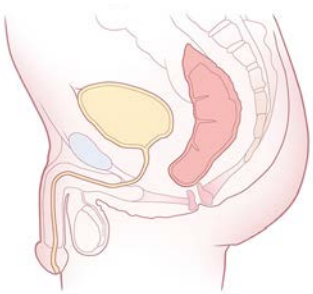
D. Normal anatomy male



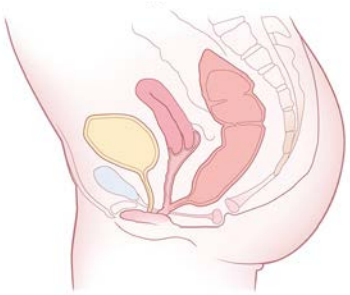
B. Anal opening missing or closed off



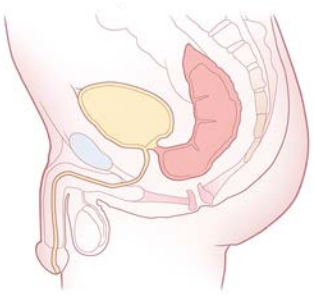
E. Anal opening missing or closed off



C. Rectum connects to vagina



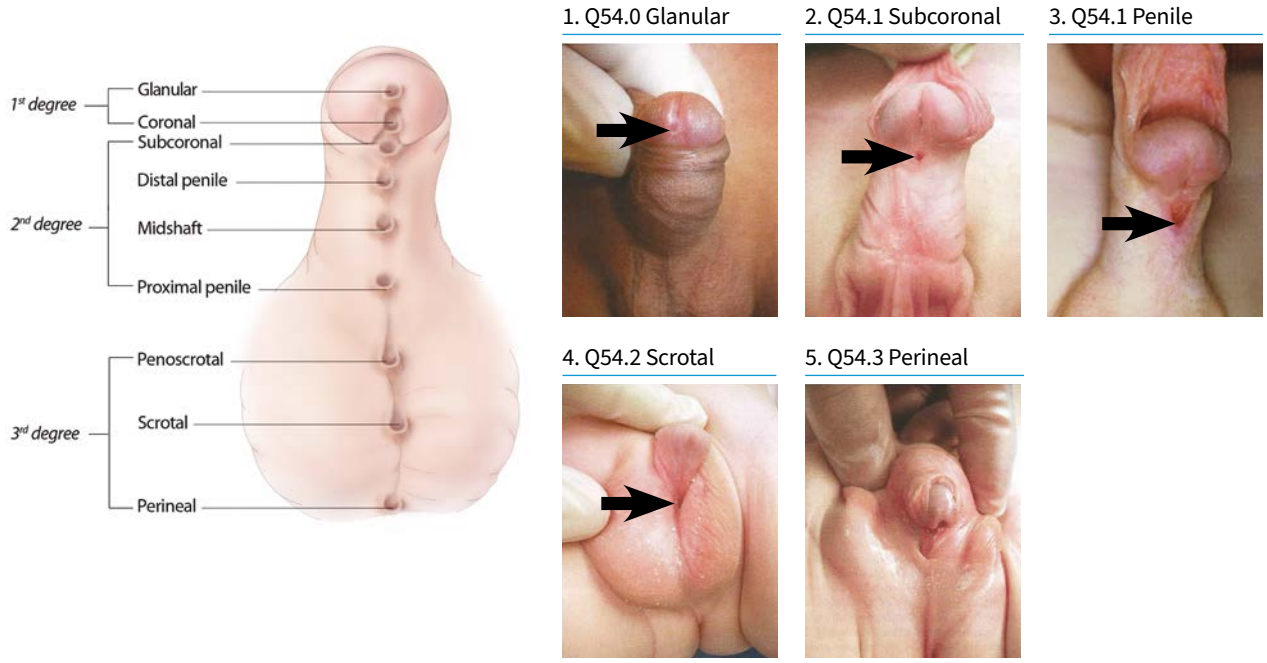
F. High rectum connects into bladder



Activity 4.7

Compare the normal male anatomy and the different types of hypospadias (see the figure below). Types of hypospadias are based on the (ventral) placement of the external urethral meatus. Using the information in the QRH, including the documentation checklist, describe the key finding in males.

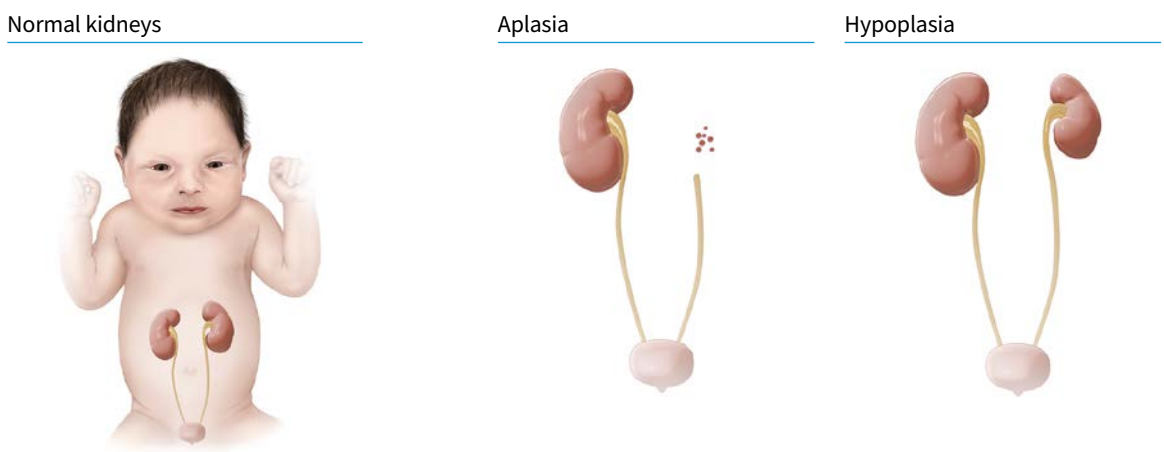
Hypospadias



Photograph source: © Kliegman RM, St. Geme III JW, editors. Nelson textbook of pediatrics, two-volume set. Philadelphia: Elsevier; 2024.

Compare photos of the normal renal anatomy with the different anomalies that occur with renal agenesis (unilateral or bilateral), renal aplasia and renal hypoplasia (see the figure below). Using the information in the QRH, including the documentation checklist, describe the key findings.

Normal kidneys and agenesis: aplasia and hypoplasia



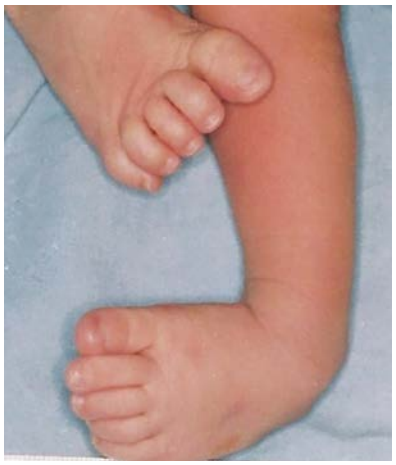
Activity 4.8

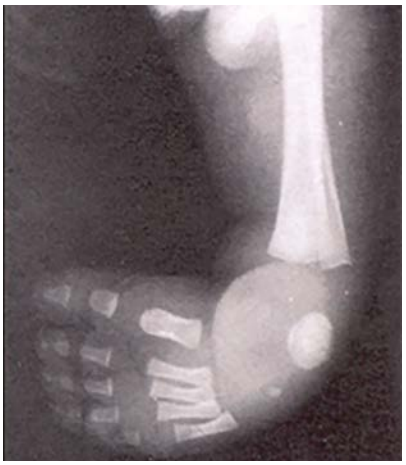
Musculoskeletal anomalies

Compare photos of the different anomalies of the ankle and foot (see the figure below). Using the information in the QRH, including the documentation checklist, describe the key findings.

Talipes equinovarus

Talipes equinovarus





Supinatus

Varus

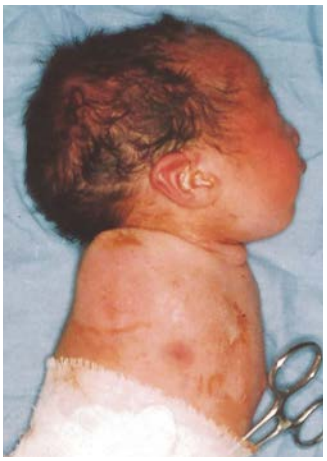
Equinus

Photograph and radiograph source: © Dr Idalina Montes and Dr Rajael Longo.

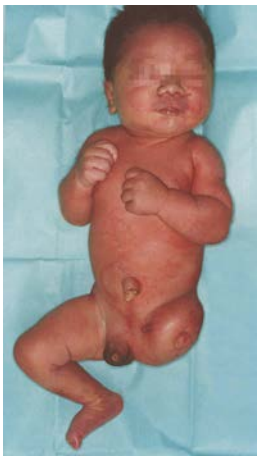
Compare photos of the different anomalies of the upper and lower extremities that may occur (arms and legs). Using the information in the QRH, including the documentation checklist, describe the key findings.

Amelia

Amelia of the upper limb



Amelia of the lower limb



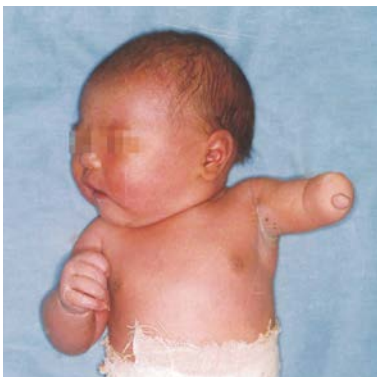
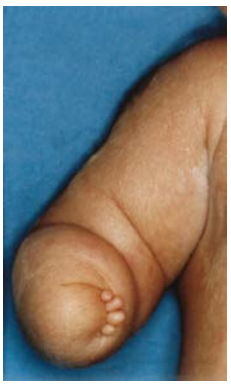
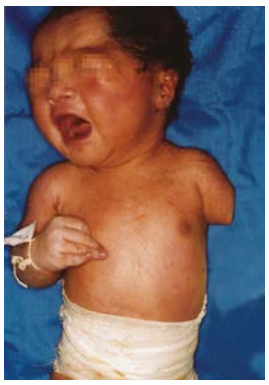
Photograph source: © CDC–Beijing Medical University collaborative project.

Compare photos of the different anomalies involving the upper and lower extremities (arms and legs) resulting in either a transverse terminal or transverse intercalary limb deficiency (see the figure below). Using the information in the QRH, including the documentation checklist, describe the key findings.

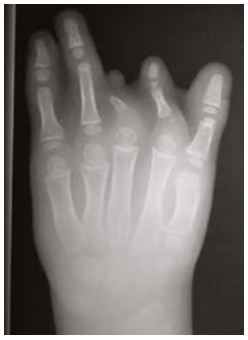
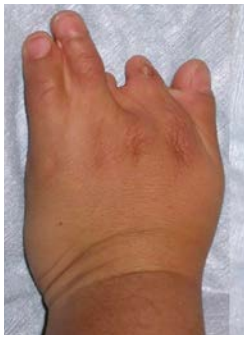
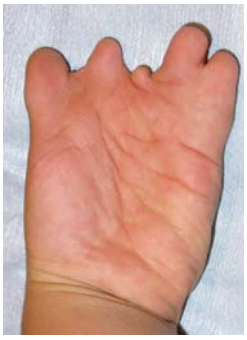
Transverse terminal and intercalary limb deficiency

Transverse terminal limb deficiency

Congenital absence of both forearm and hand (Q71.2)

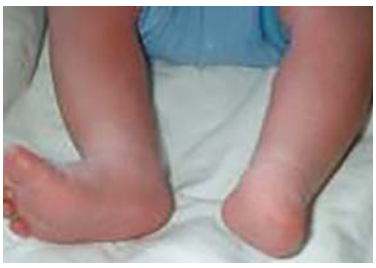
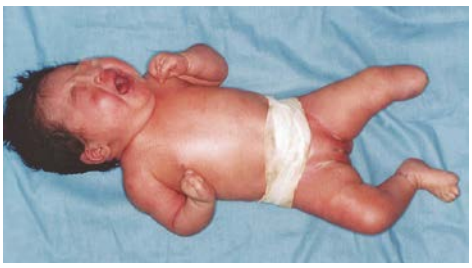




Congenital absence of finger(s) (remainder of hand intact) (Q71.30)

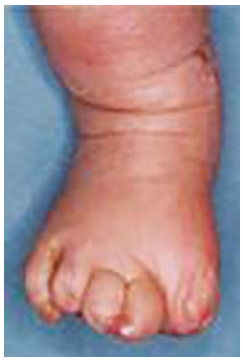
Congenital absence of foot and toes (Q72.3)

Congenital absence of both lower leg and foot (Q72.2)

Congenital absence or hypoplasia of toe(s) with remainder of foot intact (Q72.30)

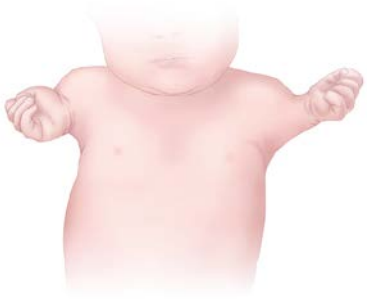
Constriction ring (Q84.81)

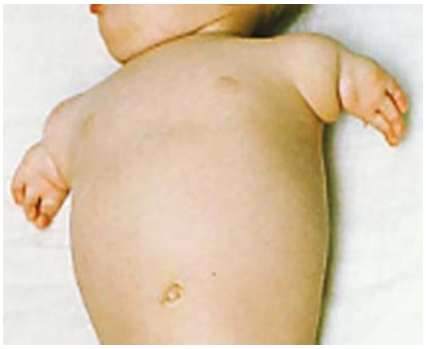



Photograph source: © QRH, 2020 (5).

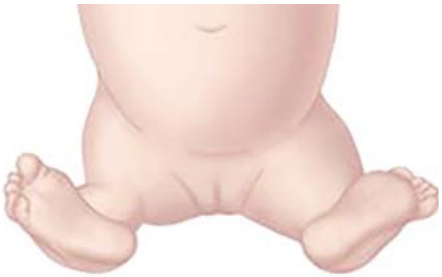
Transverse intercalary limb deficiency

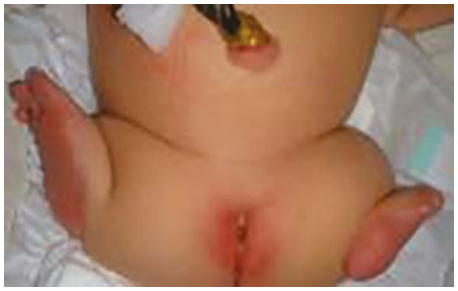
Congenital absence of upper arm and forearm with hand present



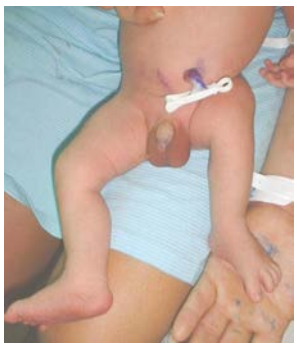


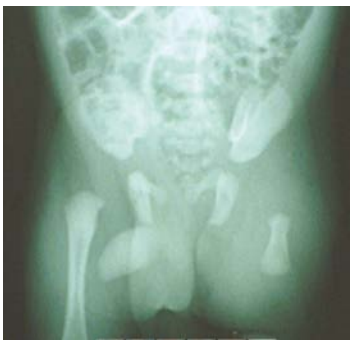
Congenital absence of thigh and lower leg with foot present





Longitudinal reduction defect of femur





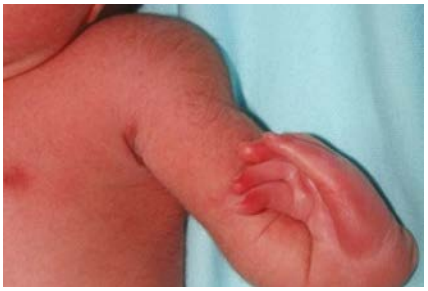
Photograph source: © QRH, 2020 (5).

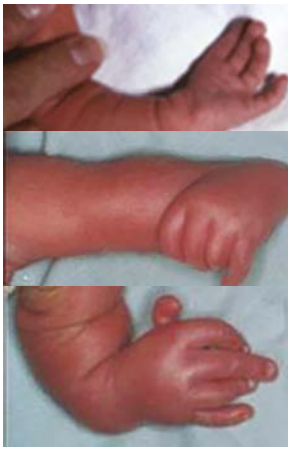
Compare photos of the different anomalies involving the upper and lower extremities (arms and legs) resulting in either a preaxial, axial, or postaxial defect (see the figure below). Using the information in the QRH, including the documentation checklist, ask participants to describe the key findings.

Longitudinal preaxial, longitudinal axial and longitudinal postaxial limb deficiency

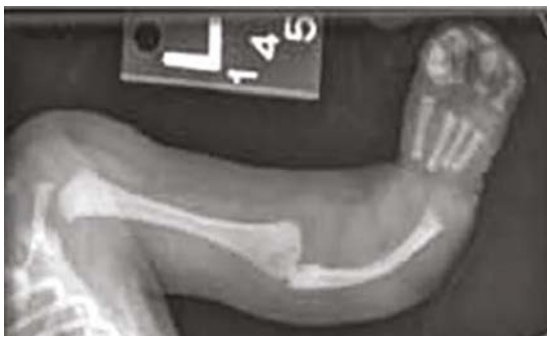
Longitudinal preaxial

Absence/hypoplasia of thumb (Q71.31)

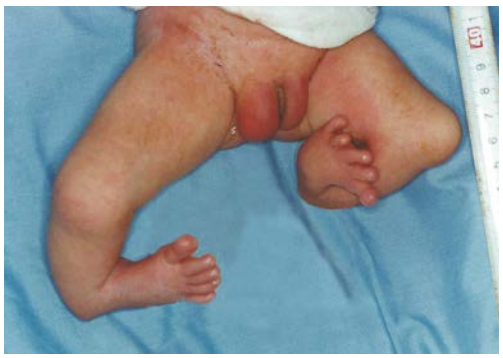




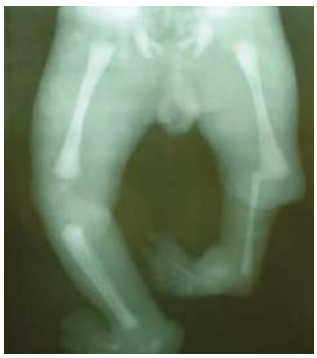
Longitudinal reduction defect of radius (Q71.4)



Hypoplasia of first toe with other digits present (Q72.31)



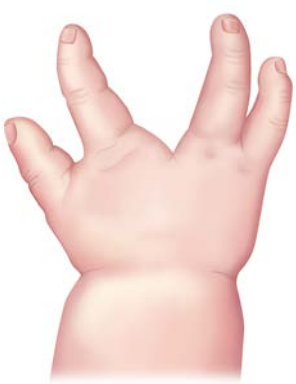
Longitudinal reduction defect of tibia (Q72.5)

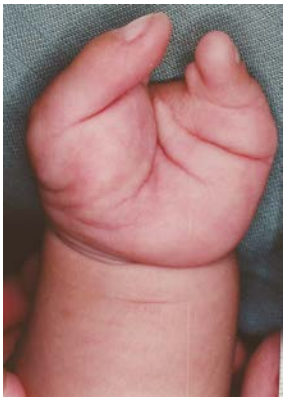


Photograph and radiograph source: © ECLAMC; CDC–Beijing Medical University collaborative project.

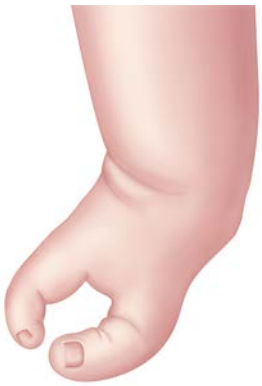
Longitudinal axial

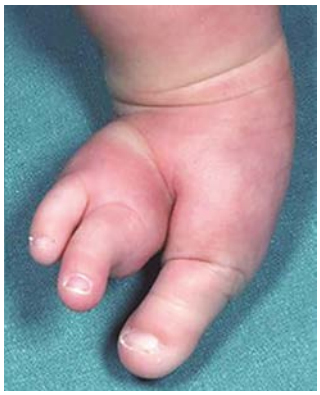
Congenital cleft hand (Q71.6)





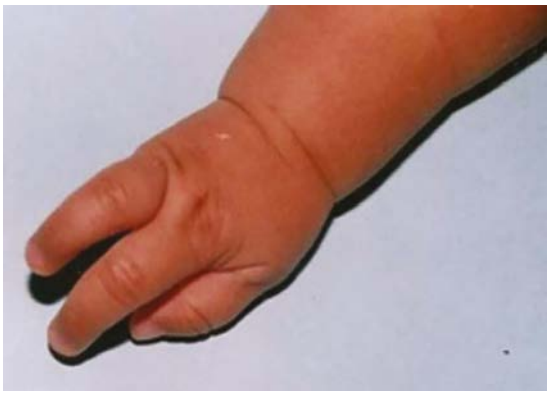
Split foot (Q72.7)

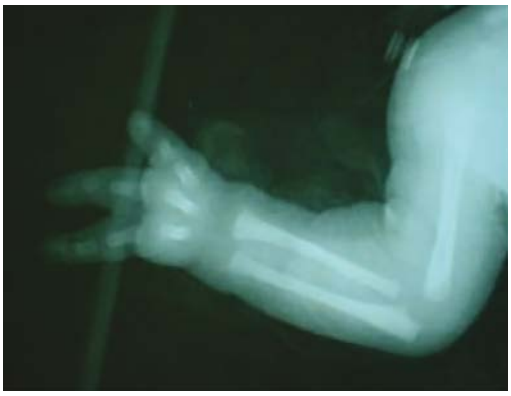




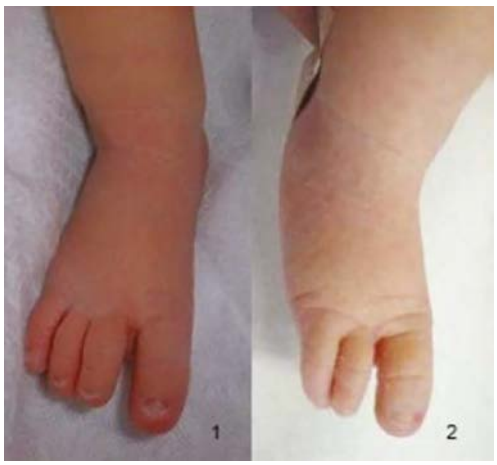
Longitudinal postaxial

Congenital absence of fourth and fifth fingers (Q71.30)





Congenital absence or hypoplasia of toe(s) with remainder of foot intact (Q72.30)



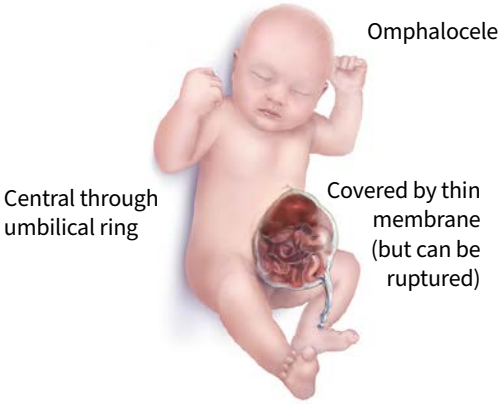
Photograph and radiograph source: © ECLAMC; CDC–Beijing Medical University collaborative project.

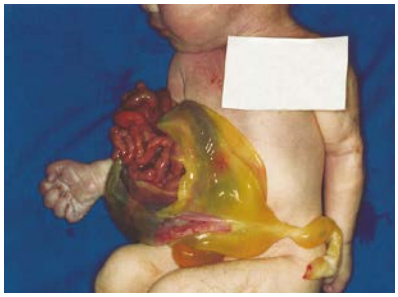
Activity 4.9

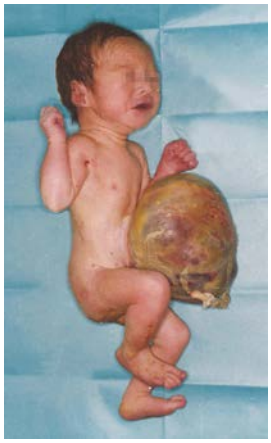
Abdominal wall defects

Compare photos of the two different types of abdominal defects (gastroschisis and omphalocele). Using the information in the QRH, including the documentation checklist, describe the key finding of each type of abdominal defect.

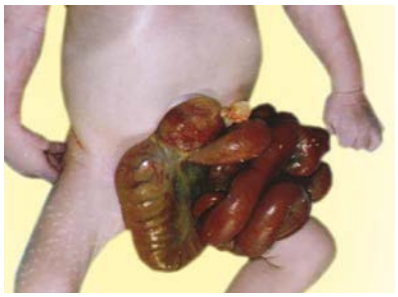
Omphalocele







Gastroschisis



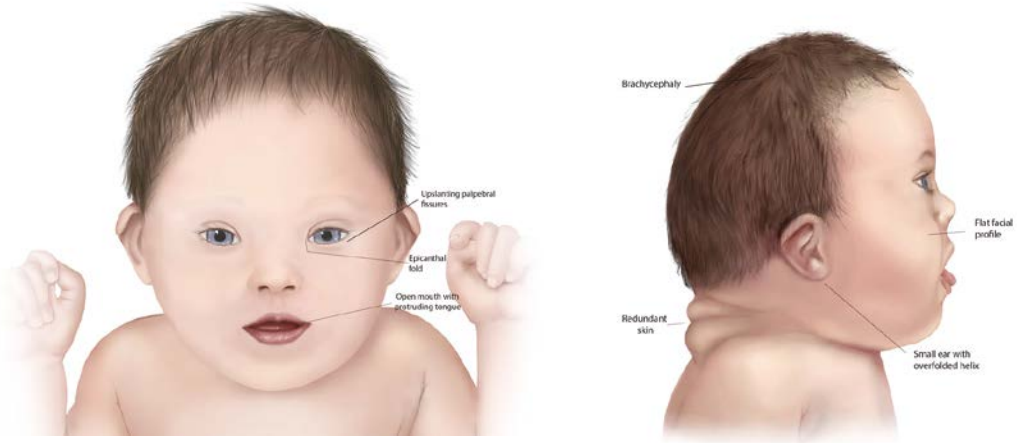
Photograph source: © CDC–Beijing Medical University collaborative project.

Activity 4.10

Chromosomal abnormalities

View the photo of the typical face of an infant with trisomy 21. Using the information in the QRH, including the documentation checklist, describe the key findings (major and minor) in an infant with this syndrome.

Down syndrome (trisomy 21)



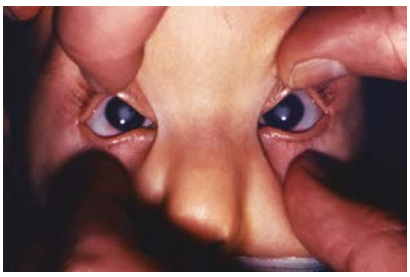
Activity 4.11

Congenital infectious syndromes

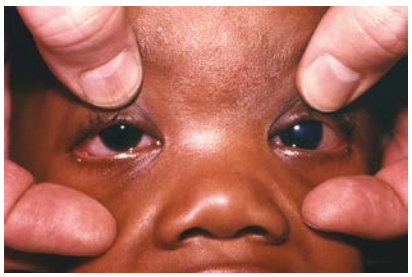
View the photo of the typical findings in an infant exposed to the rubella virus early in pregnancy (see the figure below). The critical window of exposure from maternal infection is < 18 weeks of pregnancy, with the most vulnerable period during pregnancy between 8 and 10 weeks. Using the information in the QRH, including the documentation checklist, describe the key findings in an infant with prenatal rubella infection.

Clinical findings in the infant with congenital rubella syndrome

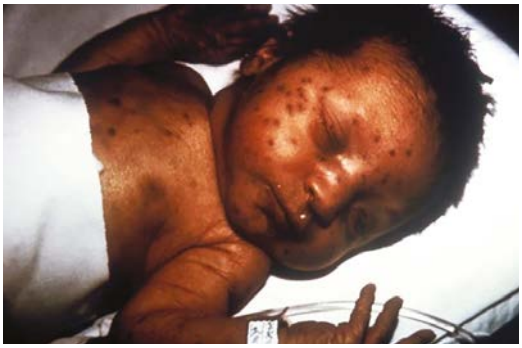
Cataracts



Congenital glaucoma (and cataract)



"Blueberry muffin" skin lesions



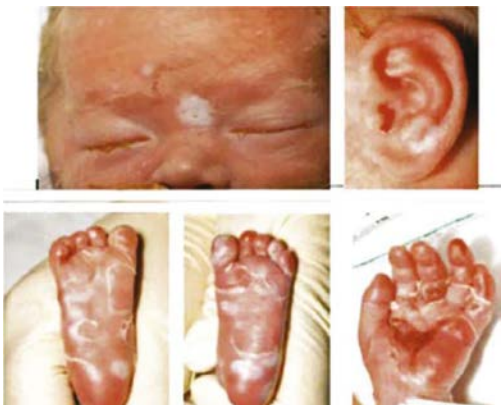
Radiolucent bone disease

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

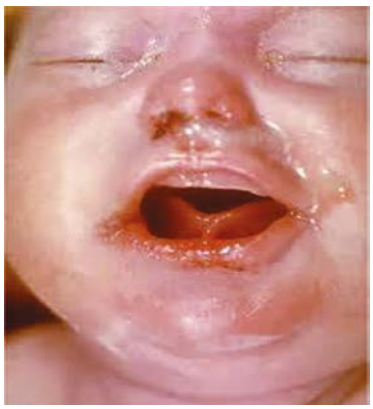
View the photo of the typical findings in an infant exposed to syphilis in pregnancy (see the figure below). Using the information in the QRH, including the documentation checklist, describe the key findings in an infant with a prenatal exposure to a maternal syphilis infection.

Clinical findings in the infant with congenital syphilis syndrome

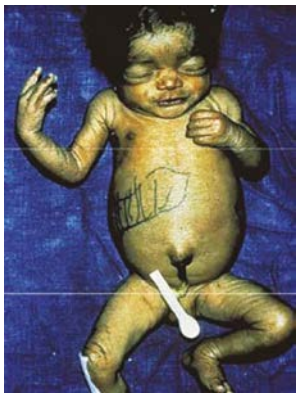
Desquamating maculopapular skin lesions



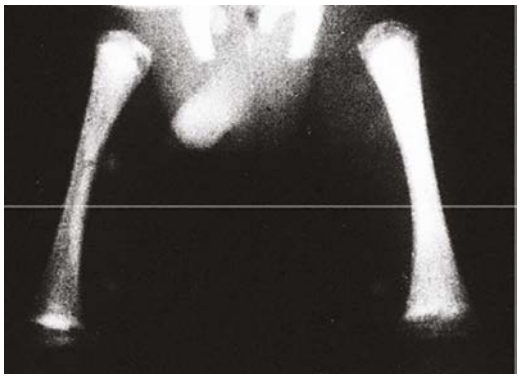
Rhinitis with mucopurulent nasal discharge



Hepatosplenomegaly and jaundice



Syphilitic metaphysitis

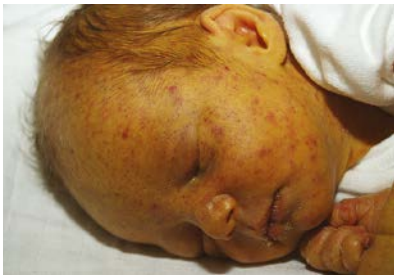


Photograph and radiograph source: © Dr Ronald Ballard.

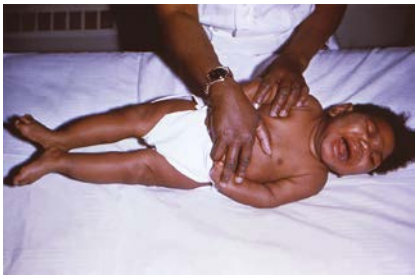
View the photo of the typical findings in an infant infected with the cytomegalovirus in pregnancy (see the figure below). The highest risk for congenital cytomegalovirus infection is from a primary infection occurring in a pregnant woman during the first and second trimester. Using the information in the QRH, including the documentation checklist, describe the key findings in an infant with a prenatal exposure to a maternal cytomegalovirus infection.

Clinical findings in the infant with congenital cytomegalovirus syndrome

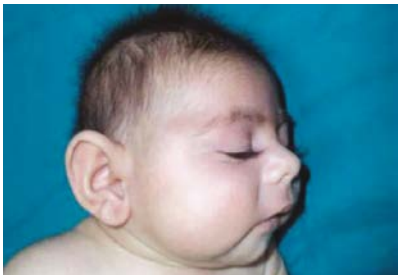
Petechial rash ("blueberry muffin" rash) and jaundice



Microcephaly and lower limb spasticity



Microcephaly

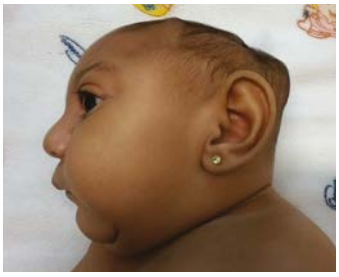


Photograph source: © Jacob Johan; CDC Public Health Image Library; Isikay S, Yilmaz K. Congenital cytomegalovirus infection and finger anomaly. BMJ Case Rep. 2013:bcr2013009486 (https://doi.org/10.1136/bcr-2013-009486).

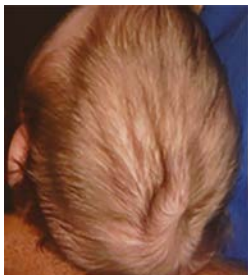
View the photo of the typical findings in an infant infected by the Zika virus in pregnancy (see the figure below). The highest risk for adverse outcomes in an infant result from a primary Zika virus infection occurring in a pregnant woman during the first and early-second trimester. Third trimester infection is associated with less severe defects of the brain and eyes. Using the information in the QRH, including the documentation checklist, describe the key findings in an infant with a prenatal exposure to a maternal Zika virus infection.

Clinical findings in the infant with Zika syndrome

Collapse of cranium and extreme reduction in the height of the cranial vault



Redundant scalp with folds (rugae)



Upper and lower limb deformities

Photograph source: © Moore CA, Staples JE, Dobyns WB, Pessoa A, Ventura CV, Borges da Fonseca E et al. Characterizing the pattern of anomalies in congenital Zika Syndrome for pediatric clinicians. JAMA Pediatr. 2017;171(3):288–95. https://doi.org/10.1001/jamapediatrics.2016.3982.

Module 5

Introduction to coding

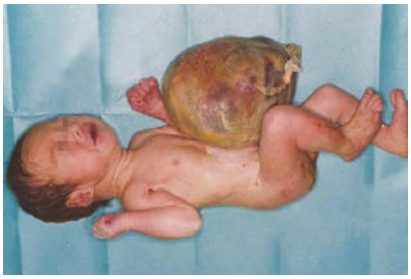
Activity 5.1

There are 27 cases in this activity.

- Break out into your small group.
- Each group will receive a set of pictures labelled with numbers.
- Write the number of the photo and describe it on the answer sheet.

DO NOT write down the ICD-10 or ICD-10-RCPCH code

- Exchange answer sheets (but not photos) with another group
- Based on the description, write down the ICD-10 or ICD-10-RCPCH code.
- Provide photos and, if necessary, re-code.
- Discuss all responses in a larger group.



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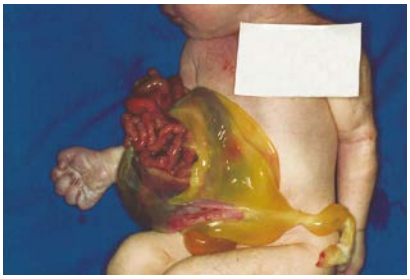

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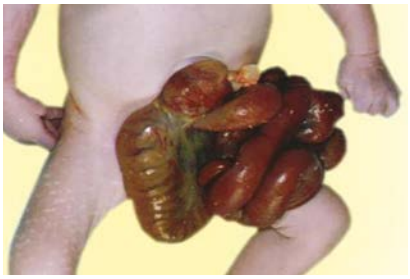

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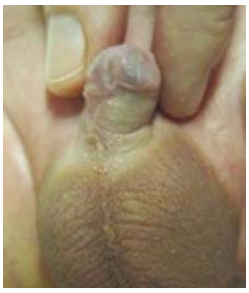

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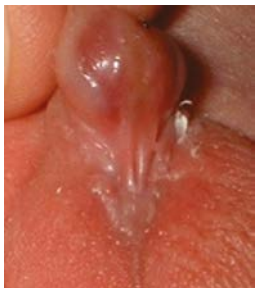

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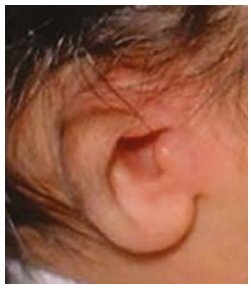

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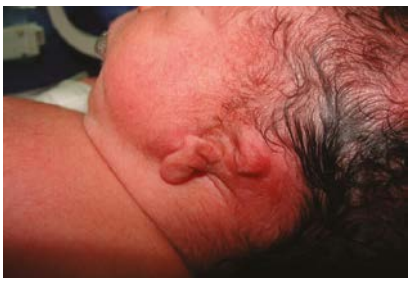

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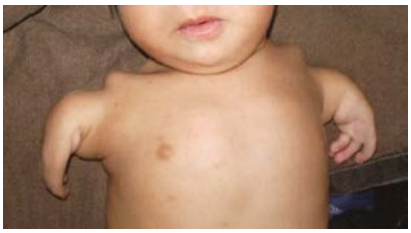

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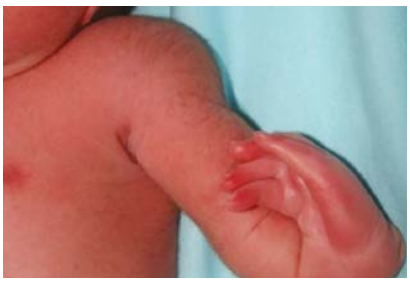

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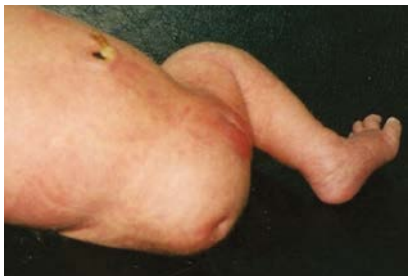

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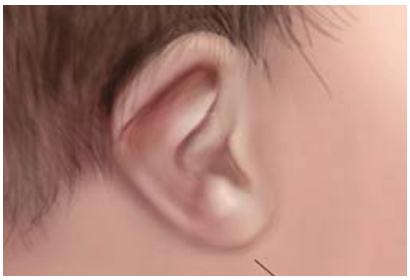

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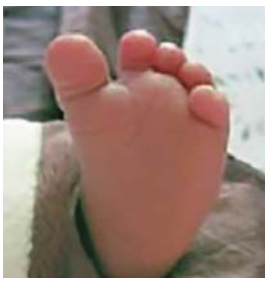

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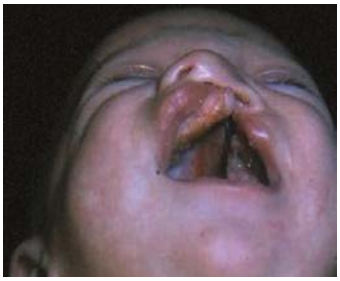

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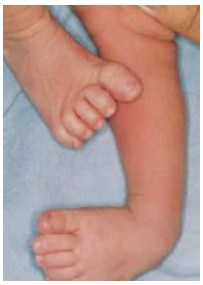

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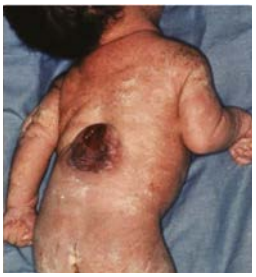

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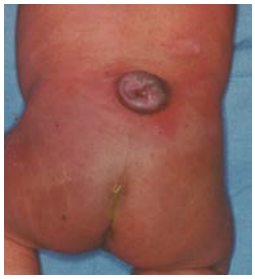

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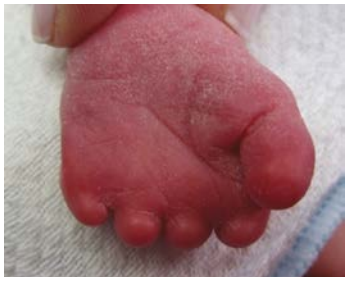

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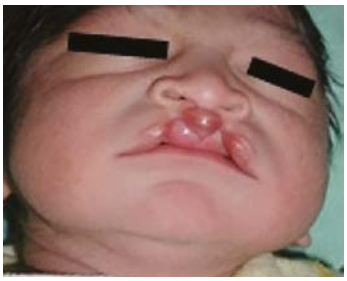

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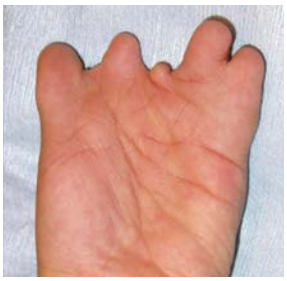

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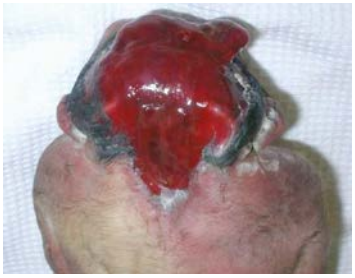

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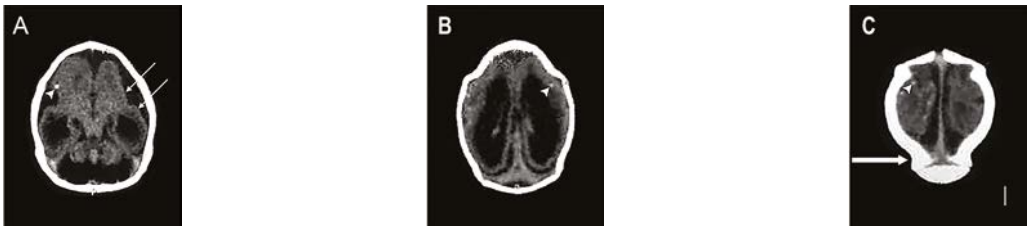

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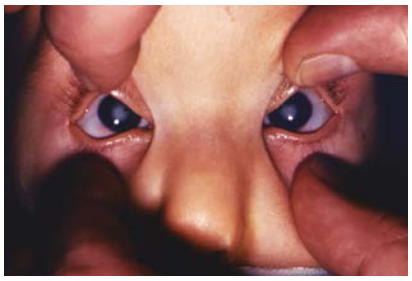

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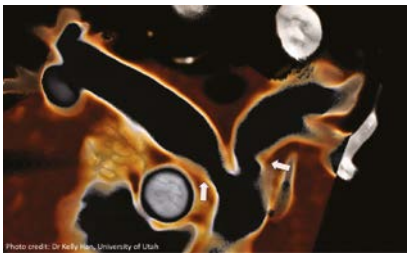

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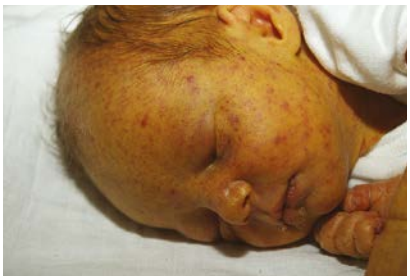

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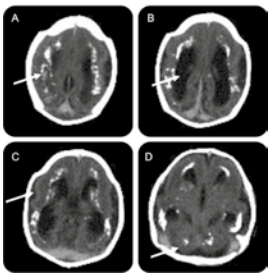

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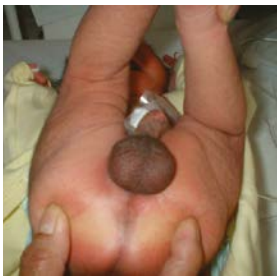

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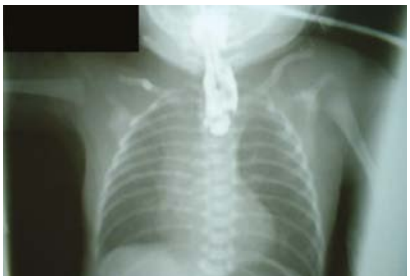

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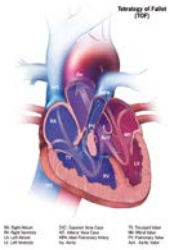

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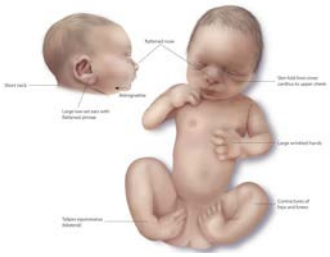

27.

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Activity 5.2

Assign an ICD-10-RCPCH code or codes, based on the available clinical description of the different fetuses or infants with congenital anomalies.

- **Case 1**
  Spina bifida with LS myelomeningocele (open); massive hydrocephalus, talipes equinovarus
- **Case 2**
  Encephalocele; unilateral cleft lip
- **Case 3**
  Cleft lip and palate; defect of the abdomen, with central herniation of the gut and liver through an enlarged umbilical ring; covered by a translucent membrane
- **Case 4**
  Facial cleft; amniotic band evidence on face
- **Case 5**
  Omphalocele, macrosomia, macroglossia, ear creases and pits

The next 25 cases can be done in your own time.

- **Case 6**
  Horseshoe kidney, ventricular septal defect, dysplastic ears, clenched hand with overlapping of fingers, prominent heels, left radial aplasia
- **Case 7**
  Hypospadias, penoscrotal; hydrocephalus
- **Case 8**
  Holoprosencephaly, aplasia cutis, bilateral cleft lip and cleft palate, patent ductus arteriosus, bilateral postaxial polydactyly, hydronephrosis

- **Case 9**
  Cleft palate, microcephaly, interrupted aortic arch type B
- **Case 10**
  Severe microcephaly, hepatosplenomegaly, petechiae, chorioretinitis, periventricular brain calcifications
- **Case 11**
  Contiguous anencephaly and open spina bifida
- **Case 12**
  Unilateral cleft lip with cleft palate; lower-lip pits, bilateral and paramedian
- **Case 13**
  Stenosis at the pulmonary valve, ventricular septal defect, aorta overriding ventricular septal defect, right ventricular hypertrophy
- **Case 14**
  Severe microcephaly with partially collapsed skull and redundant scalp with extra skin folds; hypertonia; thin cerebral cortices with subcortical calcifications; macular scarring and focal pigmentary retinal mottling; congenital contractures of major joints
- **Case 15**
  Microcephaly, congenital cataract, stenosis of pulmonary artery
- **Case 16**
  Bilateral absence of the radii, with thumbs present; thrombocytopenia
- **Case 17**
  Defect of the occipital bone, cervical and thoracic spine anomalies; head tilted with very short neck; cleft palate
- **Case 18**
  Anterior abdominal defect, lateral to an umbilical cord attached on the left; intestine outside abdomen; atresia of the bowel
- **Case 19**
  Stillbirth with autopsy showing bilateral renal agenesis, lung hypoplasia, club feet and flat facies
- **Case 20**
  Holoprosencephaly, microcephaly, cyclopia, absence of premaxilla, severely flattened nose
- **Case 21**
  Anencephaly, facial dysmorphic features
- **Case 22**
  Meningocele, omphalocele, exstrophy of the cloaca, anal atresia
- **Case 23**
  Absent left hand; soft tissue nubbins
- **Case 24**
  Bilateral absence of arm and forearm; hand present
- **Case 25**
  Small jaw, glossoptosis, upper airway obstruction, U-shaped cleft palate

- **Case 26**
  Microtia of the right side, two right-sided preauricular skin tags, facial asymmetry, fusion of cervical vertebrae
- **Case 27**
  Bilateral preaxial upper limb deficiency, oesophageal atresia with tracheo-esophageal fistula, atrial septal defect, anal atresia
- **Case 28**
  Short limbs (possible achondroplasia)
- **Case 29**
  Upslanting palpebral fissures, flat nasal bridge and midface, decreased muscle tone, brachycephaly, incurving of the fifth finger, and single transverse crease in the palm of the hand; common atrioventricular canal

Module 6

Clinical scenarios

Clinical scenario: baby Adaeze

- Q1. Describe in detail what you see, then use the visual aids and documentation checklist in the QRH to check and revise your description.
- Q2. What important components are missing or not sufficiently clear from the photograph?
- Q3. 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?
- Q4. What is the spina bifida sequence? What is, in general, a sequence?
- Q5. Adaeze has a form of spina bifida called myelomeningocele. What is the difference between meningocele and myelomeningocele? Why is it important?
- Q6. Complete the report of the anatomic findings in Adaeze's clinical presentation with appropriate coding.
- Q7. What does this presentation suggest to you? Why might it be important?
- Q8. Now look at the different clinical presentations in spina bifida. What are main points on the summary slides that you find relevant to Adaeze's case?
- Q9. What is your final description, diagnosis, and coding of Adaeze at this time? Describe the anomalies (using the checklists as guides), try and provide an overall clinical classification, and code.
- Q10. Discuss some lessons learned from this case – from the newborn exam to approaches to birth defect ascertainment and case classification.
- Q11. You have heard of folic acid. What is the relation between the vitamin folic acid and risk for spina bifida (and neural tube defects in general)?
- Q12. Besides folic acid, what are other factors that influence the risk for spina bifida? Discuss also to what extent these risk factors might be present in your specific population or community.
- Q13. This case study covers a lot of ground. How would you summarize and perhaps expand on the key points and lessons learned? Which of these do you think are particularly relevant to your local setting?

Clinical scenario: baby Esther

- Q1. What, in particular, should you be suspicious of in your exam?
- Q2. What do you do next?
- Q3. Using the figures and the documentation checklist in the QRH (pages 37 and following) describe the elements of the palate, and then the cleft palate.
- Q4. What could this be? Could it be related to the cleft palate? Check the QRH and review the description, clinical tips and checklist. What do you see now that was missed before? What do you do next?
- Q5. Can cleft palate be missed at initial examination? How can one diagnose cleft palate early so it is not missed, and prevent complications such as respiratory obstruction?
- Q6. Complete the reporting of this component of Esther's clinical presentation – namely, cleft palate and Pierre Robin sequence – with appropriate coding. How would you code these anomalies, using ICD-10 with RCPCH modifications? (Hint: codes are well described on pp. 86–87 of the MPM2)[2].
- Q7. With these findings – murmur, cyanosis, hypoxia – what do you think Esther might have? Can you list a few specific diagnoses (refer also to the QRH)?
- Q8. You alert the team. A diagnostic evaluation is done, and this diagram is shown to you, together with a diagram of an average heart. What are the key structural anomalies in Esther's heart? What is her specific cardiac diagnosis?
- Q9. How would you code tetralogy of Fallot, using ICD-10 with RCPCH modifications? (Hint: codes are mentioned in the QRH and well described on pp. 70–71 of the MPM2)[2].
- Q10. What elements do you find potentially applicable to Esther's presentation? What else might you do to complete the evaluation?
- Q11. What is your final description and diagnosis of Esther at this time? Describe the anomalies (using the checklists as guides), code, and try and provide an overall clinical classification.
- Q12. Discuss some lessons learned from this scenario – from the newborn exam to approaches to birth defect ascertainment and case classification.
- Q13. What are main points on the summary slides that you find relevant to Esther's case?
- Q14. What are the main (non-genetic), potentially preventable exposures that can increase the risk for cleft palate? Discuss also to what extent these risk factors might be present in your specific population or community.

2 MPM2 is the abbreviation for: World Health Organization, United States Centers for Disease Control and Prevention and International Clearinghouse for Birth Defects Monitoring Systems. Birth defects surveillance: a manual for programme managers, second edition. Geneva: World Health Organization; 2015 (https://apps.who.int/iris/handle/10665/337425).

Clinical scenario: baby Matias

- Q1. Describe in detail what you see.
- Q2. How would you classify this type of limb deficiency?
- Q3. What might be going on – what is the likely congenital anomaly?
- Q4. What are the associations shared by both oesophageal atresia and preaxial limb deficiencies? What other internal anomalies would you now actively be looking for, based on clinical history and these clinical notes?
- Q5. Describe these anomalies (using the checklists and visual aids in the QRH).
- Q6. Now you take stock of the clinical phenotype. You re-examine the baby and do not find other anomalies. What are his major anomalies and what is your provisional diagnostic classification?
- Q7. Describe the relevance of this association in birth defect surveillance (hint: the internal anomalies to look for when an external anomaly or another component is present).
- Q8. Try and assign codes (e.g. ICD-10 RCPCH) to the case.
- Q9. To what extent is maternal diabetes relevant to Matias's presentation? What are some congenital anomalies associated with maternal diabetes? How is this relevant to prevention?
- Q10. Discuss some lessons learned from this scenario – from the newborn exam to approaches to birth defect ascertainment and case classification.

Module 7

Data quality exercise

Activity 7.1

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

Congenital anomaly cases in Loa

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?

After you ask for more information, you receive the table below.

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

Congenital anomaly	ICD-10 code	Number of cases
Neural tube defects	Q00, Q01, Q05	189
Spina bifida	Q05	124
Anencephaly	Q00	55
Critical congenital heart defects	[a]	325
Tetralogy of Fallot	Q21.3	59
Transposition of the great arteries	Q20.3	54
Hypoplastic left heart syndrome	Q23.4	50
Orofacial clefts	[b]	348
Cleft lip alone	[b]	81
Cleft lip with cleft palate	[b]	164
Cleft palate alone	[b]	103
Abdominal wall defects	Q79.3, Q79.2	171
Gastroschisis	Q79.3	17
Omphalocele	Q79.2	154

[a] 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).

[b] cleft lip alone (Q36, Q36.0, Q36.9, Q36.90, Q36.99), cleft lip with cleft palate (Q37, Q37.0-Q37.5, Q37.8, Q37.9, Q37.99), cleft palate alone (Q35, Q35.1, Q35.3, Q35.5, Q35.59).

- Q: What changes would you suggest? Using the given data, can you add more information?

Prevalence of congenital anomalies in Loa, 2016–2020, among 245 070 births

Congenital anomaly	ICD-10 code	Number of cases	Prevalence per 10 000 births
Neural tube defects	Q00, Q01, Q05	189	7.7
Spina bifida	Q05	124	5.1
Anencephaly	Q00	55	2.2
Critical congenital heart defects	[a]	325	13.3
Tetralogy of Fallot	Q21.3	59	2.4
Transposition of the great arteries	Q20.3	54	2.2
Hypoplastic left heart syndrome	Q23.4	50	2.0
Orofacial clefts	[b]	348	14.2
Cleft lip alone	[b]	81	3.3
Cleft lip with cleft palate	[b]	164	6.7
Cleft palate alone	[b]	103	4.2
Abdominal wall defects	Q79.3, Q79.2	171	7.0
Gastroschisis	Q79.3	17	0.7
Omphalocele	Q79.2	154	6.8

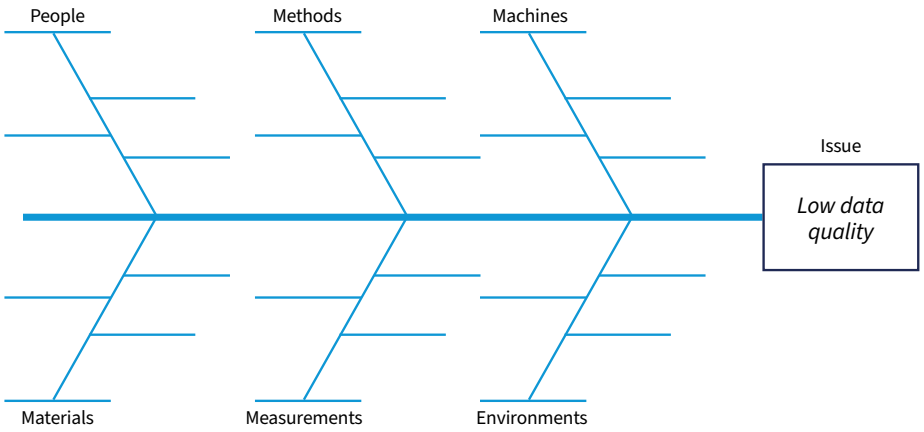
[a] Persistent truncus arteriosus (Q20.0), Double outlet right ventricle (Q20.1), D-transposition of great arteries (Q20.3), Single ventricle (Q20.4), Tetralogy of Fallot (Q21.3), Pulmonary valve atresia (Q22.0), Tricuspid atresia (Q22.4), Aortic valve stenosis (Q23.0), Hypoplastic left heart syndrome (Q23.4), Coarctation of the aorta (Q25.1), Interrupted aortic arch (Q25.2), Total anomalous pulmonary venous return (Q26.2).

[b] Cleft lip alone (Q36, Q36.0, Q36.9, Q36.90, Q36.99), Cleft lip with cleft palate (Q37, Q37.0-Q37.5, Q37.8, Q37.9, Q37.99), Cleft palate alone (Q35, Q35.1, Q35.3, Q35.5, Q35.59).

- Q: Now that you have prevalence figures, you can compare the data of your programme with other programmes. A good practice to compare 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 neighbouring country, and in fact, it is quite lower than the previous five-year period. What might be the reasons for this low prevalence?
- Q: 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 spina bifida with hydrocephalus, talipes equinovarus

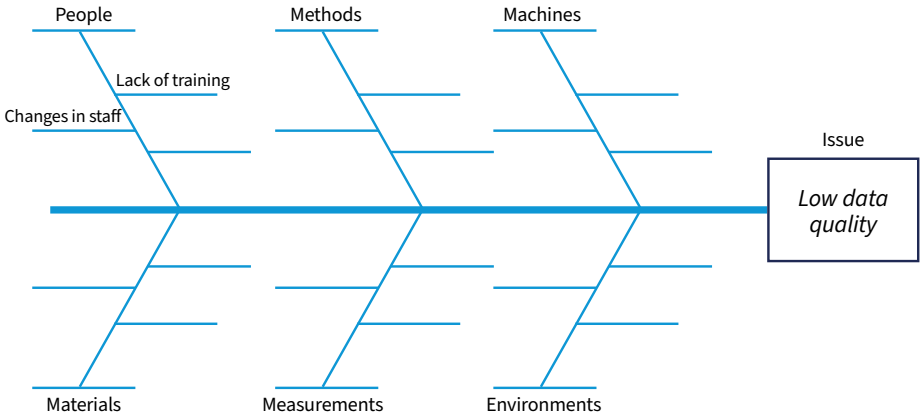
What information is missing from these case descriptions?

- 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?
- What can you do to diagnose what might be causing the 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, such as low data quality. Use the fishbone diagram template to add in the problem (i.e. low data quality) in the issue box. Next, add in the six categories of potential causes in the branches: people, methods, machines, materials, measurements and environment.



Note: "people" refers to those involved with the process (e.g. frontline workers, programme coordinators); "methods" refers to procedures described in standard operating procedures; "machines" refers to diagnostic equipment, computers and tools; "materials" refers to paper forms and measuring tape for head circumference; "measurements" refers to data analysis, calculation of prevalence and data quality indicators; and "environment" refers to the conditions and culture of the reporting site and central coordination.

- Q: Add potential causes of low data quality as sub-rays within categories. Please complete.



- Q: Once you diagnose the potential causes of the issue, you can start with an intervention. How would you improve data quality?
- Q: Comment on champions. Who are the champions of a surveillance system? Why are they important?
- Q: We found out that data quality was low after we looked at the reported data from 2016–2020. (And the data have already been produced). What would you do to prevent this from happening again? Discuss the difference between quality control and quality improvement.

For more information, please contact:

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