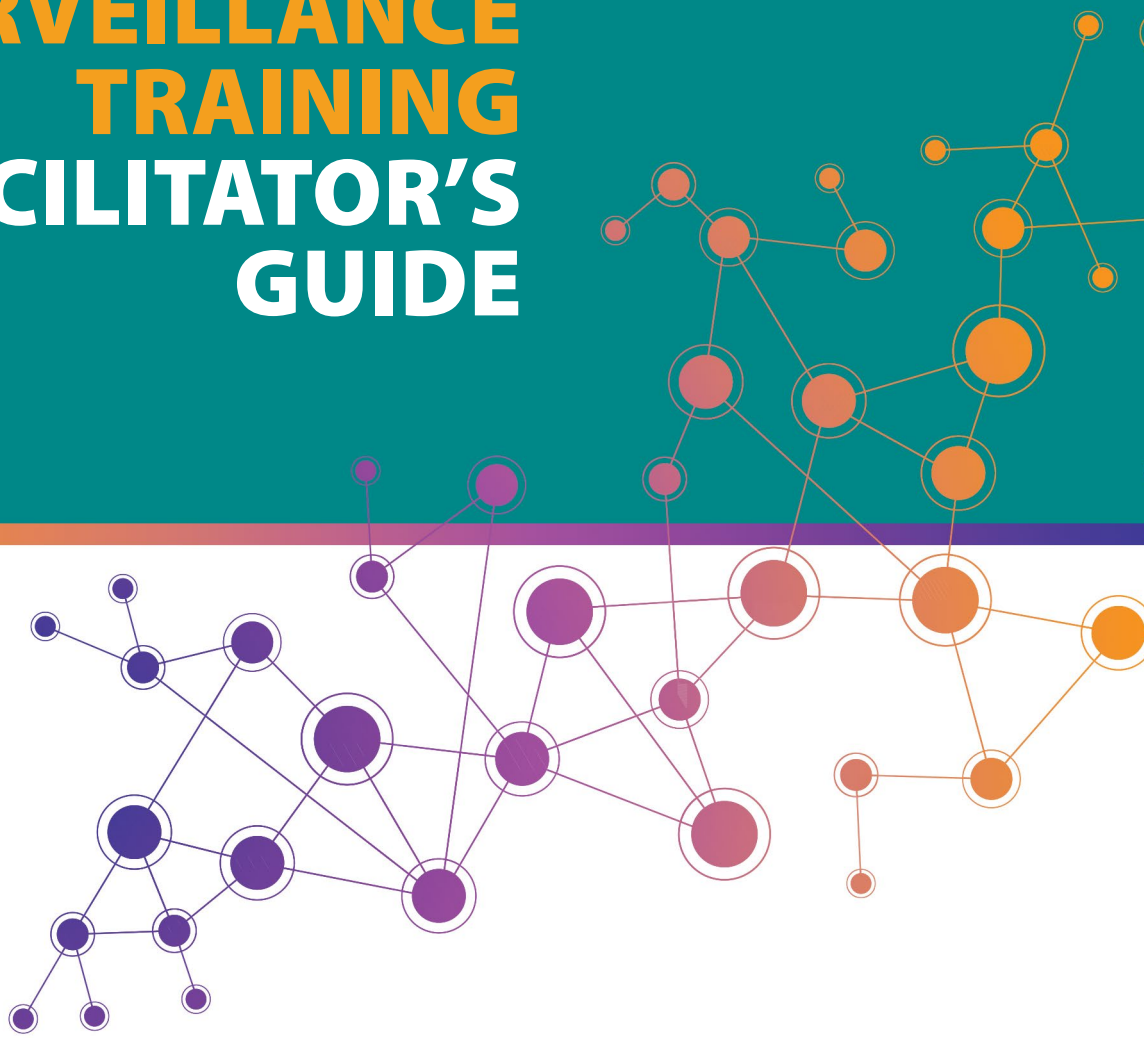
# BIRTH DEFECTS SURVEILLANCE TRAINING FACILITATOR'S GUIDE



World Health Organization

CDC
Centers for Disease Control and Prevention

International Clearinghouse for Birth Defects Surveillance and Research

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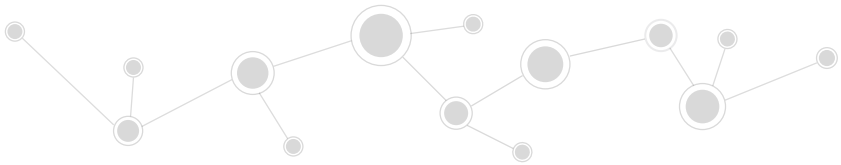

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## Acknowledgements

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We would like to thank the WHO Regional Office for South-East Asia, for providing valuable feedback during the development process. We acknowledge the Division of Birth Defects and Developmental Disabilities Surveillance Workgroup for their contributions toward the development of content for this resource.

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## Contents

## Abbreviations

| | |
|---|---|
| **CDC** | United States Centers for Disease Control and Prevention |
| **COUNT** | Countries and Organizations United for Neural Tube Defects Prevention |
| **ETOPFA** | elective terminations of pregnancy with fetal anomalies |
| **ICBDSR** | International Clearinghouse for Birth Defects Surveillance and Research |
| **ICD-10** | *International statistical classification of diseases and related health problems*, 10th revision |
| **RCPCH** | Royal College of Paediatrics and Child Health |
| **USA** | United States of America |
| **WHO** | World Health Organization |

## Introduction

Congenital anomalies, also known as birth defects, are structural or functional (e.g. metabolic disorders) anomalies that occur during intrauterine life and can be identified prenatally, at birth or later in life *(1)*. Synonymous terms that are often used are "birth defects", "congenital abnormalities" and "congenital malformations", but the latter has a more specific meaning. For the purposes of this guide, the term "congenital anomalies" will be used throughout.

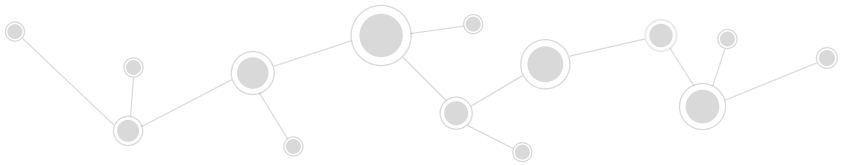
According to the World Health Organization (WHO) in 2010, an estimated 270 358 deaths globally were attributable to congenital anomalies during the first 28 days of life, with neural tube defects being one of the most serious and most common of these anomalies. In an effort to decrease the number of congenital anomalies worldwide, the Sixty-third World Health Assembly adopted a *Birth defects* resolution. Among other objectives, this resolution encourages countries to build in-country capacity related to the prevention of congenital anomalies and to raise awareness about their effects *(2)*. Through the development of a population-based surveillance programme that accurately captures congenital anomalies, countries can gain a better understanding of the burden of and risks for these conditions, refer identified infants to services in a timely manner, and use prevalence estimates to evaluate any current prevention or clinical management programmes. Countries can also use the information gathered to inform stakeholders and policy-makers about the importance of investing in programmes aimed at reducing the occurrence of congenital anomalies, and help them plan for appropriate services.

The International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) is a voluntary nongovernmental organization in official relations with WHO. ICBDSR brings together congenital anomalies surveillance and research programmes from around the world, with the aim of investigating and preventing congenital anomalies and lessening their impact.

ICBDSR was first established in 1974 in Helsinki, Finland, where representatives of malformation registries in 10 countries were present. There are 43 member programmes in the current year, which are actively engaged in systematic collection and analysis of data. Their main objective is to monitor and prevent congenital anomalies. ICBDSR established a centre, currently in Rome, which is funded in part by the United States Centers for Disease Control and Prevention (CDC) and collaborates with WHO and CDC to develop training courses for surveillance of congenital anomalies.

CDC's National Center on Birth Defects and Developmental Disabilities has developed Birth Defects COUNT (Countries and Organizations United for Neural Tube Defects Prevention), a global initiative to significantly reduce mortality and lifelong morbidity resulting from neural tube defects. This initiative contributes to the achievement of United Nations Millennium Development Goal 4 (Reduce child mortality) and supports the 63rd World Health Assembly resolution on birth defects *(2)*.

Through Birth Defects COUNT, CDC provides scientific and programmatic technical expertise to expand neural tube defects prevention efforts and strengthen surveillance of congenital anomalies worldwide.



## Course overview

### About this surveillance course

The goal of this course is to provide participants with the foundational skills needed to begin the development, implementation and ongoing improvement of a congenital anomalies surveillance programme, in particular for countries with limited resources. It focuses on the methodology needed to develop either population-based or hospital-based surveillance programmes.

A set of congenital anomalies will be used as examples throughout this course. The specific examples used are typically severe enough that they would probably be captured within the first few days after birth, have a significant public health impact and, for some of them, have the potential for primary prevention.

### Target audience

This course is designed for individuals who are directly involved in the development, implementation and daily activities related to congenital anomalies surveillance programmes in their countries and who will directly teach others.

### Learning objectives

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

1. describe how to use data for the development of prevention campaigns and policy/programme evaluation;
2. describe how to best present data to policy-makers;
3. identify selected major external congenital anomalies for monitoring;
4. describe the tools needed to ascertain and code congenital anomalies;
5. describe the processes for managing and analysing data;
6. demonstrate how to calculate the birth prevalence of congenital anomalies.

### Course competencies

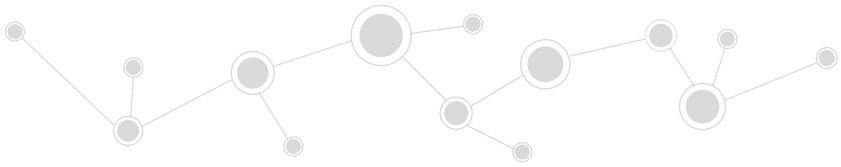
This course is based on a set of competencies that every participant is expected to learn during the course and subsequent practice and follow-up at their place of work. To become competent at something, a certain amount of knowledge is needed, as well as proficiency in certain skills. Table 1 lists the competencies for the course (column 1) and the knowledge (column 2) and skills (column 3) required for each competency.

### Structure of the course

Conducted in its entirety, the course length is approximately 4–5 days. However, the actual length will depend on the needs and experience of the country and the participants; therefore, the facilitator will need to decide which chapters to cover and adapt the time frame of the course accordingly. The sessions use a variety of teaching methods, including lectures, demonstrations and work in smaller groups, as well as practicums and exercises.

**Table 1. Course competencies and their knowledge and skills requirements**

| Competency | Knowledge | Skills |
|---|---|---|
| 1. Identify congenital anomalies | • Define congenital anomalies<br>• Describe terms that are used as synonymous with congenital anomalies<br>• List common risk factors associated with congenital anomalies | • Recognize and explain major external congenital anomalies |
| 2. Describe public health surveillance | • Define public health<br>• Define surveillance<br>• Define public health surveillance<br>• List the purposes of public health surveillance | • Explain public health surveillance and its purposes to key partners |
| 3. Use logic models for surveillance of congenital anomalies | • Define logic models<br>• List the components of logic models<br>• Describe how logic models can support programme planning | • Create a logic model for congenital anomalies surveillance<br>• Revise a logic model as a programme evolves |
| 4. Help engage partners for a congenital anomalies surveillance programme | • List partners and their potential role(s)<br>• Describe different ways to communicate with different groups of partners | • Apply competencies<br>• Identify key partners as appropriate for a country or setting<br>• Advocate with key partners using the logic model |
| 5. Help set up a reporting system for congenital anomalies surveillance | • List advantages and disadvantages of mandatory and voluntary reporting<br>• Describe confidentiality and privacy issues to be considered when setting up a reporting system for congenital anomalies surveillance | • Develop a reporting system |
| 6. Define the surveillance programme that is most appropriate for a given setting | • Define the population under surveillance<br>• Identify areas of coverage<br>• Define population-based and hospital-based surveillance programmes<br>• List advantages and disadvantages of each type of surveillance programme<br>• List factors to consider when deciding the most appropriate surveillance programme | • Select the surveillance programme that is most appropriate, considering the population under surveillance and areas of coverage<br>• Select the population under surveillance<br>• Select the geographic area under surveillance |



| Competency | Knowledge | Skills |
|---|---|---|
| 7. Determine how to ascertain cases | • Describe case-ascertainment approaches<br>• List advantages and difficulties related to each approach<br>• List potential data sources<br>• Describe the different characteristics to be considered for case inclusion<br>• Describe potential case inclusion/ exclusion criteria, according to the type of surveillance<br>• List core variables to be included in a surveillance programme | • Help identify the best ascertainment approach in a given setting<br>• Define cases to be included, as appropriate for a given setting<br>• Define data sources that will be used<br>• Define core variables to be considered for a given surveillance programme |
| 8. Help set up a coding system for a congenital anomalies surveillance programme | • Describe methods for describing congenital anomalies in the data-collection process<br>• Define the international standard diagnostic classification system<br>• Describe ways to improve the coding system for certain congenital anomalies not thoroughly addressed in the *International statistical classification of diseases and related health problems*, 10th revision (ICD-10) *(3)* | • Help identify a coding system using the ICD-10/Royal College of Paediatrics and Child Health (RCPCH) coding system<br>• Assign appropriate codes to diagnoses of congenital anomalies |
| 9. Help to disseminate congenital anomalies surveillance data | • Describe the use of data<br>• List potential end users of the data<br>• Describe the dissemination method that is most appropriate for each stakeholder/audience | • Develop dissemination messages according to different audiences<br>• Help develop dissemination tools |
| 10. Use public health surveillance for a congenital anomalies programme | • List the objectives of a congenital anomalies surveillance programme<br>• Describe the main types of congenital anomalies surveillance programmes<br>• List potential congenital anomalies that will be collected by the surveillance programme | • Explain how to develop a congenital anomalies surveillance programme<br>• Identify the congenital anomalies that will be collected by the surveillance programme |
| 11. Help identify the best congenital anomalies surveillance programme for a given setting | • Describe key components that will influence which surveillance programme is most appropriate for a given setting | • Apply competencies to a decision about which surveillance programme is most appropriate for a given setting<br>• Describe the congenital anomalies surveillance programme that is most appropriate for a given setting |

## Course materials

### *The Facilitator's guide*

The *Facilitator's guide* contains what you, the facilitator, need in order to lead participants through the course. It contains detailed instructions on how to conduct each session. This is your most essential tool as a facilitator. It is recommended that you use it at all times and add notes to it as you work. These notes will help you in future courses.

### *PowerPoint slides*

Many sessions use slides. These are provided on a CD for projection onto a screen, or can be downloaded from a site. Please contact CDC for further details (birthdefectscount@cdc.gov).

### Birth defects surveillance: a manual for programme managers

This manual *(4)* can be used for reference after the course. If resources are available, provide a copy of the *Facilitator's guide* to each participant, so it is not essential for participants to take detailed notes.

### Birth defects surveillance: atlas of selected congenital anomalies

This atlas *(5)* can be used for reference after the course; it can also be used for some of the coding activities during the training.

### *Participant workbook*

The workbook, which can be found at the end of this *Facilitator's guide,* is to be provided to participants on the first day of the training, and includes specific activities that they will do by themselves or in groups during the training. One copy of the participant workbook should be made for each participant.

### Training aids

As a facilitator, you will need a flipchart, and blackboard and chalk, or white board and suitable markers, for most sessions, and a means of fixing flipchart pages to the wall or notice board – such as masking tape.

You will also need a computer/laptop, an LCD (liquid-crystal display) projector and a screen.

## Organizing a training programme

This section is for the use of a senior facilitator or a course coordinator.

### Preparation

In order to hold a successful course, you may need to arrange:

- classroom space for the course and classroom space for preparation;
- a listing of hotels and restaurants for the facilitators and participants.

### Classroom facilities

A large classroom to accommodate the whole class, including facilitators and visitors or observers, is required. The classroom should have space for each group and their facilitators to sit at a table during the sessions. Additional table space to lay out the materials used during the course is suggested. The classroom should be in a place where the participants are not disturbed by too much background noise.

### Accommodation

For a residential course, it is necessary to arrange for suitable accommodation near the place where the classroom is. Unsatisfactory accommodation can hinder participants' learning. Ideally, a training venue will be central to most participants.

### Clerical and logistical support

It would be ideal for photocopies and other training materials to be made available prior to the course; however, if this is not possible, make sure that clerical and support staff will be available at the site to assist with this. They should be able and willing to help with anything that requires their attention.

### Funds required

Ideally, sufficient funds should be available to cover the following:

- participants' travel and per diem, if required;
- facilitators' travel and per diem and special compensation, if required;
- payment for clerical support staff, if required;
- stationery, equipment;
- refreshments;
- accommodation and meals (if not covered by per diem);
- midday meal and refreshments, such as coffee and tea;
- costs of photocopying.

It is very important that decisions about how costs will be covered are made well in advance, as what is covered by the organizer versus the participant can vary.

## Opening and closing ceremonies

You may wish to have an opening and closing ceremony for the participants. There may be an invited speaker to open the course and to close it and present certificates to the participants and any new facilitators. It is important to involve representatives from the government and key institutions, so that they are aware of the training, and to acknowledge or obtain their support for congenital anomalies surveillance.

Decide in good time who to invite. Send an invitation with a short description of the course and the participants. Make it clear whether or not you want those whom you invite to make a speech. If you do wish them to speak, stress the exact time that will be available. Send them relevant information that would be appropriate for them to mention.

If possible, before the course, try to make personal contact with those who accept the invitation and ensure that they fully understand the context in which they make their speech.

Prepare the course timetable to include the time needed for opening and closing ceremonies. It is important that your course schedule does not get disturbed by lengthy speeches, particularly on the first day.

## Selecting and preparing the facilitators

### *Profile of a facilitator*

A facilitator for this course ideally will be a public health professional who has participated in a congenital anomalies surveillance workshop sponsored by ICBDSR, CDC and WHO. He or she should have substantial experience with congenital anomalies surveillance. Further, it is recommended to have more than one facilitator, since organizing and implementing this workshop is labour- and time-intensive.

### *Inviting faculty*

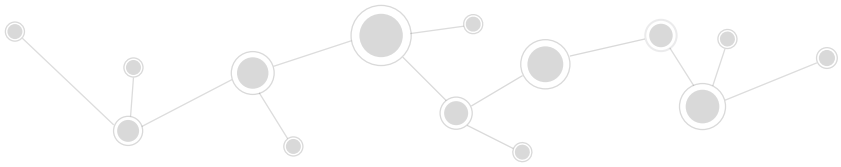
Invite faculty early and confirm their availability, so that you know how many participants you can invite.

Give the exact dates, and make it clear that you expect them to attend the entire course, including the preparation. Explain that the preparation is necessary for the faculty to become familiar with the content and methods of the course. Give any additional administrative details, such as arrangements about finance and accommodation.

If faculty members live near to where the course will be held, it may be useful to involve them early in the preparations for the course.

Preparation of faculty takes place before the participants' training and is the responsibility of the course director or a senior more experienced faculty individual. The preparation of faculty will depend on the experience they have already. All faculty need time to review the agenda, visit site facilities, check materials and equipment for their sessions, and spend time learning how to assess participants.

If the faculty have different levels of experience, you will need to arrange the preparation time to ensure their different preparation needs are met.



### *Distribute materials*

The facilitator should give each invited faculty a copy of the *Facilitator's guide*, *Birth defects surveillance: a manual for programme managers (4)*, *Birth defects surveillance: atlas of selected congenital anomalies (5)*, and the agenda for the course.

### *Explain the objectives of the preparation*

The objectives are:

- to learn how to use the course materials, especially the *Facilitator's guide;*
- to decide the content of the course;
- to become familiar with the information in the materials, and to discuss any points that are not clear;
- to practise the facilitations skills;
- to prepare to teach the different kinds of sessions;
- to discuss the management of the course.

### *Practising the sessions*

- Assign practice sessions to faculty. It is useful to ensure that new faculty practise giving a lecture and facilitating a group work session; after the practice, discuss the teaching practice.
- Check that the projector, electrical extension cords if needed, flipchart, and all other equipment is in place, or that the facilitators know where to obtain it.
- Define roles and responsibilities and discuss among the facilitators who is responsible for providing materials, stationery and equipment.
- Appoint a senior representative from the involved agencies, whom the facilitators can contact if they need something.
- Inform all the facilitators about the daily facilitators' meetings of about one half to one hour, which are very important for the success of the course. Agree on an acceptable time (usually at the end of the day) and place.
- Time may be needed in the evenings after the session to prepare and practise the next day's sessions.

### *Facilitator and faculty meetings*

Facilitators should coordinate faculty meetings at the end of each day for about 30–60 min. Keep the meetings brief. Discuss the activities of the day, including progress made by participants, to identify any difficulties impeding progress and any skill, exercise or section of the sessions that participants found especially difficult to understand or carry out. Identify solutions to any problems related to any particular group's progress, or to difficult skills or sections of the sessions. Discuss teaching techniques that the faculty have found to be successful.

Review important points to emphasize in the sessions the next day. Remind the facilitators to consult the *Facilitator's guide* and gather together any supplies needed for the next day.

## Selecting participants

The number of participants who can be invited for a course depends on the budget and the capacity of the classroom and availability of residential accommodation for participants and facilitators. It is recommended that you do not invite more than 25 participants to a course.

### *Example of a course announcement*

**Course on congenital anomalies surveillance**

Date:

Venue:

Course organizers: [name, affiliations, email]

Objectives of the course: After completing this course, participants will be able to…

Who should attend:

Outline of course: The course requires full-time attendance for a minimum of 4 complete days.

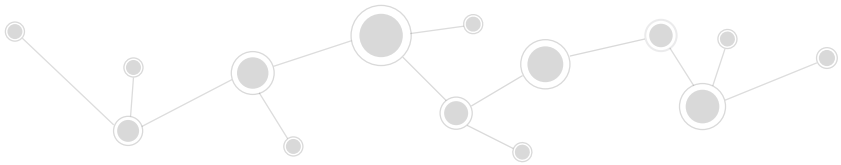
Accommodation:

Registering for the course: Send the names and contact details of candidates who wish to apply to [name and address] before [date]. When participants have been selected, further information will be sent to them and to their health facility. In some cases, the organizing agency (i.e. the Ministry of Health) may decide who should be trained, based on needs.

## Arrangements at the course site, before the course begins

Confirm arrangements for:

- lodging for all facilitators and participants;
- classrooms;
- daily transportation of participants from lodgings to classroom;
- meals and refreshments;
- opening and closing ceremonies with relevant authorities; confirm participation of invited guests and speakers;
- a course completion certificate (if one will be given) and when a group photograph will be taken in time to be developed before the closing ceremony (optional);
- typing/word processing and copying of materials during the course (for example, timetables, lists of addresses and emails of participants and trainers).



Ideally, arrange to welcome facilitators and participants at the lodging facility, airport or railway/bus station, if necessary.

Prepare the course agenda (see Annex 1: Congenital anomalies surveillance: tentative programme) and timetable for facilitators' preparation.

Prepare the evaluation form and make enough copies.

### Actions during the course

- After registration, create groups of three to four participants. Post up the list of names where everyone can see it.
- Provide all participants and faculty with a course directory, which includes the names and addresses and emails of all participants and faculty, including the facilitators.
- Arrange for a course photograph, if desired, to be taken.
- Prepare a course completion certificate for each participant.
- Make arrangements to reconfirm or change airline, train or bus reservations for faculty and participants, if necessary.
- If needed, allocate a time for payment of per diem and for travel/lodging arrangements that does not take time from the course.

## Resources and materials

| Resources | Materials |
|---|---|
| For the facilitator and faculty:<br>• *Facilitator's guide*<br>• PowerPoint slides<br>• Group discussion guide<br>• Activities guide<br>• Chapter evaluation forms<br>• Course evaluation forms<br>• Pens and pencils<br>• Markers<br>• Tape to secure flipchart pages to wall if needed<br>• Attendance log<br>• *Birth defects surveillance: a manual for programme managers (4)*<br>• *Birth defects surveillance: atlas of selected congenital anomalies (5)*<br>• Video of Jay Walker, chairman of TEDMED<br>• A copy of the ICD-10-RCPCH codes for the congenital anomalies chapter | For the facilitator and faculty:<br>• Flipcharts and easels<br>• Laptop<br>• LCD projector<br>• Calculator<br>• Name tags<br>• Power cords<br>• Power outlet adapters if needed |
| For the participants:<br>• Participant workbook (make sure to have enough copies), and prepare necessary materials for all activities – extra paper, photographs with numbers for Activity 5.2 etc.<br>• *Birth defects surveillance: a manual for programme managers (4)*<br>• *Birth defects surveillance: atlas of selected congenital anomalies (5)*<br>• Pens and pencils | For the participants:<br>• Laptop (if possible)<br>• Power cords<br>• Calculators |
| *Note:* Prior to the workshop, the facilitator will send a set of questions that the participants should bring with them to the workshop. These questions can be found in Annex 2. | |

## Facilitator's notes

Suggested actions and script for the facilitator are located in the "Script/key points" sections, to facilitate each session. Also included are references to the page numbers in the participant workbook, as well as instructions on when to use the activities. Use these materials as you prepare for your session, to guide you during the workshop. Be sure to refer participants to the appropriate page number in their workbooks throughout the session.

This course is intended to be used as a teaching guide and introduction to the WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*. The facilitator will see in this guide that there are specific references to sections of the manual.

Script for the facilitator to SAY is written in plain text like this.

**Instructions for the facilitator to DO are written in bold text like this.**

*Possible responses are written in italicized text like this.*

## Icon glossary

Specific slide or PowerPoint presentation

Flipchart use

Activity

Group discussion

Questions for facilitator to ask participants

Video presentation

## Role of the facilitator

This section is adapted from *A workshop curriculum on policy advocacy strategy development: facilitator's guide*. Seattle: Programme for Appropriate Technology in Health (PATH); 2013:9–10 (http://www.path.org/publications/files/APP_advocacy_workshop_fac_guide.pdf).

As the facilitator, you are critical to participant learning and to the overall success of the workshop. Below are some basic facilitation tips to keep in mind.

- Be enthusiastic! The energy level of participants will reflect your own energy level.
- Arrive at least one hour before the starting time to organize the room and materials, and to make other final preparations.
- Make sure to introduce yourself and greet participants as they arrive.
- Learn the names of each participant and use their names frequently. This helps participants feel valued and included.
- Do not turn your back to the participants, but be sure to walk around during the course of the workshop. Moving around engages the participants, keeps their attention and helps everyone hear you.
- Stay flexible with the agenda. It is important that participants become familiar with the material so they can produce high-quality work. If you spend more time on one subject area, plan to make up time elsewhere so all of the material will be covered.
- Acknowledge all participants' responses, to encourage them to try again. Comment briefly on their answer, or say "Thank you", or "Yes". If participants give an incorrect answer, do not say "No – that is wrong!" or some may hesitate to make other suggestions. Accept all answers, and say something non-committal, such as "That is an interesting idea" or "I haven't heard that one before". Ask them to say more to clarify the idea, or say, "What does anyone else think?" or ask for other suggestions. Make participants feel that it is good to make a suggestion, even if it is not the "correct" answer. Then clarify the information so that participants have the correct information.
- When someone answers correctly, repeat the response, expand it if necessary, and make sure that everyone else has understood.
- Do not let several participants talk at once. If this occurs, stop the talkers, and give them an order to speak in. For example, say "Let's hear from someone who has not yet had a chance to speak". People will usually not interrupt if they know that they will have a turn to talk.
- Do not let the same one or two people answer all the questions. If a talkative participant tries to answer several questions, ask him or her to wait for a minute, or move away and focus attention on others. Try to encourage quieter participants to talk. Ask by name someone who has not yet spoken to try to answer a question, or walk towards someone to bring attention toward him or her, and make him or her feel that he or she is being asked to talk.
- The "Expected time" provided at the start of each module is variable, based on the amount of discussion and participation by the group members.

*Note:* If there is a facilitation team, they should meet briefly at the end of each day to review the daily participant feedback and decide whether there is a need to adjust the next day's content and agenda accordingly. These meetings are generally very important to the success of a workshop.

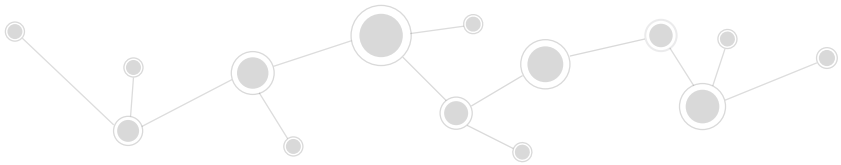
During small group work, the facilitator(s) will need to be sure to work with each team as mentors. As a mentor, your primary role is to make sure participants follow instructions, understand the learning concepts and apply those concepts correctly in their work. You will be able to identify areas where participants may be confused, and answer questions as they arise.

During small group work, follow these general guidelines:

- Allow groups to work independently. Be available for guidance, but encourage groups to work through activities on their own first.
- Pay attention to the conversation. If groups begin to lose focus or clearly don't understand a concept correctly, do not hesitate to redirect or clarify ideas.
- Encourage the group's critical thinking. When asked a question, respond with another question, to allow them to think through a response on their own.
- Watch the clock. Groups may get into lengthy discussions, easily lose track of time, and thus fail to complete an activity. Offer time reminders at mid-point and 5–10 min before the activity ends.
- Encourage the group to assign roles. Groups can function in an efficient and effective manner if a timekeeper, recorder and spokesperson are assigned at the beginning of an activity.
- *Note:* Many of the questions posed during group discussions and activity times ask the participants about situations in their countries. There will be a variety of responses, as each country is unique and some may be further ahead in congenital anomalies surveillance than others. It is important to let participants know that these kinds of questions do not have a right or wrong response, and to encourage participants to openly share experiences from their countries.

Before beginning the training, one of the facilitators should:

- provide a brief overview of the entire course and the agenda for day 1;
- provide a brief overview of *Birth defects surveillance: a manual for programme managers (4)*; participants should have been asked to read the manual prior to the course;
- provide orientation on what is expected from the participants: indicate that every morning, one participant, or a group of participants, will be asked to provide a summary of what they learnt during the previous day's workshop. The summary presentation should be no longer than 10 slides and it should show a clear understanding of definitions, principles and key issues presented the previous day.



## Using the *Facilitator's guide*

### Before you lead a session

Be completely familiar with this *Facilitator's guide* before beginning a session. Make sure you are aware of what your responsibilities are, and read the "Objectives", to find out what the participants should be able to do at the end of the session.

Read the text for the session, so that you are clear what you will have to do. The text includes detailed point-by-point instructions about how to conduct the session.

Consider splitting the session between two or more trainers, particularly if the session is long. Trainers can also work together, with one trainer writing on the flipchart or assisting with a demonstration while the other trainer is conducting the session.

### When you lead a session

Keep the *Facilitator's guide* handy and use it as a reference during the training. You do not need to try to memorize what you have to do. It is extremely difficult to do so. Use the guide as your session notes, and follow it carefully. Also keep handy *Birth defects surveillance: a manual for programme managers (4)* and *Birth defects surveillance: atlas of selected congenital anomalies (5)*. It would be useful to have means that help you identify the sections of the manual and the atlas that relate to the session you are leading.

If the participants appear tired or their attention is wandering, the facilitator may consider pausing for a short break. Encourage everyone to stretch and take some deep breaths. Perhaps a short activity, song or game may revive them.

### Checklist of training skills

On page 3 of this *Facilitator's guide* is a summary "list of competencies". The senior facilitator may decide to demonstrate these competencies at the time of preparing the trainers before a course, or it may be useful to study them individually. Refer to the list from time to time, to remind youself how to make your session effective.

**During the week, the facilitators should try to spend as much time as possible with their groups, to learn what the participants feel competent at and where they need more help and practice.**

## Pre-course instruction

If participants have to follow the pre-course instruction, a section where the pre-course is briefly described, including the expected time for completing, it will be made available.

## Course instruction

### Script/key points

- Welcome the participants and introduce yourself.
- Ask the participants to introduce themselves and have them indicate what their job duties are with regard to public health surveillance in their countries.
- Ask the participants to indicate the reason(s) for attending the course, their experience and their expectations.
- Discuss break times, restroom (bathroom, toilet) locations, and any other relevant information, as needed.
- Present an overview of the workshop.
- Explain the workbook and how it will be used to complete exercises and take notes.
- Consider having the participants speak briefly about the state of congenital anomalies surveillance in their countries.

For group activities or discussions, consider ways to divide participants that will promote participation from all. It is recommended that groups should not have more than five people, to encourage full participation.

# INTRODUCTION TO CONGENITAL ANOMALIES SURVEILLANCE

MODULE 1

Expected time: 4 hours

## Objectives

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

- describe the purpose and importance of public health surveillance for congenital anomalies.

*Note*: This module is linked to Chapter 1 of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*.

## Public health and surveillance of congenital anomalies

Expected time: 1 hour

### Script/key points

- Let's start by discussing public health. What comes to mind when you think of public health? **Let participants respond, and write responses on the flipchart.**
- Can anyone define public health? **Ask participants to provide responses aloud, and write responses on the flipchart.**
    - *Response:*
        - *According to CDC, "public health is the science of protecting and improving the health of families and communities through education, policies, services and research. Public health is concerned with protecting the health of the population, such as individuals living in a neighborhood, city, country, region, or world" (6).*
- Now let's talk about surveillance. What comes to mind when you think of surveillance? Can anyone define surveillance? **Ask participants to provide responses aloud, and write responses on the flipchart.**
    - *Possible responses:*
        - *Surveillance means to monitor. It is focused and systematic observation.*
        - *Police monitoring*
        - *Weather monitoring*
- Now we will put the two together. Can anyone give me a definition of public health surveillance? **Let participants respond, and write responses on the flipchart. After participants have provided their responses, give the definition below as given by CDC.**

- *Public health surveillance is the*
  - *ongoing*
  - *systematic collection*
  - *analysis and interpretation of health data,*
  - *for public health purposes.*

- Now we are going to watch a video by Jay Walker, chairman of TEDMED, which shows how the first official compilation of public health data revealed unexpected statistical trends. **Display video on screen** (https://www.youtube.com/watch?v=5IRsqDnPzSE).


- What are some purposes for which surveillance data could be used? **Ask participants to provide responses aloud, and write responses on the flipchart.** Some of these purposes include:
  - To assess the prevalence of certain conditions in a country
  - To identify areas with high or low rates, to try and identify epidemics or clusters of illness and disease
  - To disseminate public health information for assessment and public health response
  - To plan, implement and evaluate health strategies:
    - For example, surveillance data are critical to help determine whether a programme is having an impact, to evaluate if new strategies are necessary, and to identify problem areas and target populations that require more intensive intervention and follow-up
  - To integrate data with the decision-making process for the prevention or care of adverse health conditions
  - To document burden, to develop public health priorities
  - To develop political and social will
  - To document the burden of congenital anomalies to plan for services
- We are now going to define congenital anomalies. Congenital anomalies are abnormalities of structure or function that are identified before birth, at birth, or later in life and are of prenatal origin.


- **Ask participants:** Does anyone know of other terms used to refer to congenital anomalies? **Ask participants to provide responses aloud, and write responses on the flipchart.**
  - *Possible responses*:
    - *Congenital anomalies*
    - *Congenital abnormalities*
    - *Congenital malformations*: the use of this term to refer to congenital anomalies in general is not appropriate because malformations are only one type of congenital anomaly, which in addition include, as mentioned in the ICD-10, Chapter XVII, deformations, disruptions and dysplasias

- **Ask participants:** Which term do you prefer to use and why? **Ask participants to provide responses aloud, and write responses on the flipchart.**
- **Tell participants:** For the remainder of the course, we will be using the preferred term since it is the term most commonly used in this region. Given the stigmatization of people with disabilities, it is important to discuss that congenital anomalies is preferred terminology at WHO, but for clarity the terms are used interchangeably with congenital anomalies, in this material.

*Note to facilitator*: if there is another term more commonly used, consider using that term throughout the course.

- Now let us think about the objectives of a congenital anomalies surveillance programme. What do you think are some of the desired outcomes of surveillance? **Ask participants to provide responses aloud, and write responses on the flipchart.**
- The following are objectives of a congenital anomalies surveillance programme:
  - To collect systematic, valid, reliable and timely data on congenital anomalies
  - To define the magnitude and distribution of congenital anomalies by time, person and place
  - To identify high-risk populations or identify clusters (aggregation of cases)
  - To monitor trends in the prevalence of different types of congenital anomalies in a defined population
  - To provide scientific data and information for priority setting, planning, implementing and evaluating congenital anomalies programmes
  - To refer affected infants to appropriate services in a timely manner
  - To disseminate findings and interpretations to appropriate partner organizations and government agencies in a timely fashion
  - To provide a basis for epidemiologic research and prevention programmes
  - To inform public health and health-care policies and programmes
  - To plan for needed services among the affected population
- Ultimately, the purpose of a congenital anomalies surveillance programme can be to evaluate programmes; to define, characterize and help support prevention efforts for these congenital anomalies and their associated complications; and to help identify resources for individuals living with a congenital anomaly.

## Begin Group discussion 1.1

- **Ask participants:** How do you think surveillance programmes can be used to assess congenital anomalies? Allow participants time to write responses. **Encourage group discussion about responses before reviewing how surveillance has actually been used in assessing congenital anomalies.**
  - *Possible responses:*
    - *To measure the burden of congenital anomalies and identify high-risk populations*

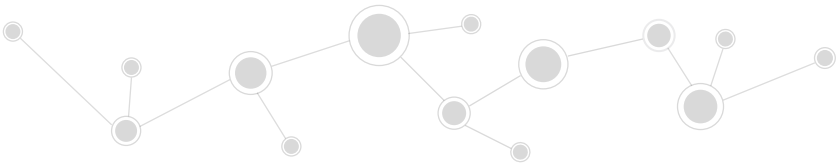
- *To identify disparities in prevalence and outcomes by factors, which could include race/ethnicity, maternal age, socioeconomic level or geographic region*
- *To assess the impact of prenatal screening and diagnosis, and other changes in diagnostic technologies, on the birth prevalence of congenital anomalies*
- *To describe short-term and long-term outcomes for children with congenital anomalies, and provide information relevant to long-term management of individuals who are affected by serious congenital anomalies*
- *To inform public health and health-care policies and programmes and plan for needed services in the population*

## Congenital anomalies

Expected time: 1.5 hours

### Script/key points

- According to WHO, an estimated 270 358 deaths globally were attributable to congenital anomalies during the first 28 days of life in 2012, with neural tube defects being one of the most serious and most common of these congenital anomalies.
- The 63rd World Health Assembly in 2010 adopted a resolution on congenital anomalies *(2)*, to encourage countries to build in-country capacity related to the development of congenital anomalies surveillance systems and the prevention of congenital anomalies, and to raise awareness about their impact. The resolution calls on Member States to prevent congenital anomalies wherever possible, implement screening programmes, and provide ongoing support and care to children with congenital anomalies and their families. WHO is to support Member States in implementing these services and to strengthen research and data collection in this area.
- Structural congenital anomalies can be classified as one of two types: major anomalies or minor anomalies. Major and minor anomalies may sometimes be present in one individual.
    - Major anomalies are structural changes that have significant medical, surgical, social or cosmetic consequences for the affected individual, and typically require medical intervention. Examples include spina bifida, anencephaly, heart defects and orofacial clefts. Major anomalies account for most of the mortality, morbidity, and disability related to congenital anomalies.
        - **Tell participants:** Because the focus of *Birth defects surveillance: a manual for programme managers (4)* and this workshop is on major structural anomalies, the examples we will use throughout the course are major anomalies.
    - Minor anomalies are structural changes that pose little or no significant health problem and tend to have limited social or cosmetic consequences for the affected individual. Minor anomalies are more common than major anomalies and can be a useful tool for clinicians to identify syndromes. Examples of minor anomalies include single palmar crease and clinodactyly (mild curvature of a finger).



- **Ask participants:** What external congenital anomalies are most often seen in your country? **Ask participants to provide responses aloud, and write responses on the flipchart**.
- We are now going to discuss risk factors. In the case of congenital anomalies, risk factors include genetic factors; maternal conditions (e.g. diabetes and obesity); maternal age; and behaviours and environmental exposures that may put a woman at risk for having a pregnancy affected by a congenital anomaly.
- **Ask participants:** Is anyone familiar with risk factors associated with congenital anomalies? **Ask participants to provide responses aloud, and write responses on the flipchart.**
  - *Possible responses:*
    - *Nutritional deficiencies/insufficiencies (e.g. folate)*
    - *Maternal age*
    - *Maternal illnesses (e.g. diabetes, hypothyroidism)*
    - *Infectious diseases (e.g. rubella, syphilis)*
    - *Alcohol abuse*
    - *Obesity*
    - *Tobacco use*
    - *Certain medications*
    - *Environmental pollution (e.g. pesticides)*
    - *Low socioeconomic status*
    - *Consanguinity*
    - *Genetic factors*
- **Ask participants:** Can any of the risk factors you mentioned be modified to help prevent congenital anomalies? **Ask participants to provide responses aloud, and write responses on the flipchart.**
  - *Possible responses:*
    - *Have vaccinations up to date prior to pregnancy*
    - *Maintain a healthy weight*
    - *Consume adequate micronutrients, like folic acid, through fortified food products or vitamin supplements prior to pregnancy*
    - *Control diabetes prior to pregnancy*
    - *Use iodine to prevent hypothyroidism*
    - *Abstain from alcohol abuse or smoking prior to, and during, pregnancy*
    - *Speak with a health-care provider about any medication use*

## Surveillance

Expected time: 1 hour

### Script/key points

- **Ask participants**: Can you name some surveillance programmes that currently exist? **Ask participants to provide responses aloud, and write responses on the flipchart.**
    - *Possible responses:*
        - *Occupational health surveillance programme*
        - *Environmental public health surveillance programme*
        - *Behavioural risk factor surveillance system*
        - *Laboratory-based surveillance*
        - *Chronic disease surveillance*
        - *Congenital anomalies surveillance programme*
        - *Infectious diseases surveillance*
        - *Vaccine-preventable disease surveillance*

- We are now going to discuss types of surveillance programmes that are most relevant for the documentation and classification of congenital anomalies.
- There are two main types of surveillance programmes often used for congenital anomalies surveillance. These are population-based and hospital-based programmes.
    - Population-based surveillance programmes capture pregnancy outcomes of interest for the condition under surveillance, such as congenital anomalies, in a population living in a defined geographical area.
    - Hospital-based surveillance programmes capture pregnancy outcomes of interest for the condition under surveillance, such as congenital anomalies that occur in selected hospitals in a well-defined location.
- Countries with limited resources may choose to start with a hospital-based surveillance programme and expand it over time into a population-based programme. Population-based and hospital-based surveillance programmes are discussed in more detail in WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4).*

### Begin Group discussion 1.2

- **Divide participants into groups of 3–4 and have them discuss within their groups which type of surveillance programme would be best suited for the following scenarios. After each smaller group has had time to discuss, review responses with the whole group.**

**Scenarios**

1. You are a public health professional in a country with few resources, where 48% of births occur in the home. You are asked to set up a surveillance programme to assess the prevalence of neural tube defects among the population. Which type of surveillance programme would you use? Why?
    - *Possible response:*
        - *Because there are few resources and 52% of births occur in hospital settings, it may be best to begin with a select number of large maternity hospitals. Once the hospital-based surveillance is well established, one can then begin to identify neonates with congenital anomalies born at home who are taken to hospitals or local clinics for services, to assess the possibility of implementing a population-based surveillance programme. If the country has community health workers and midwives who can cover home births and report to the surveillance programme, a population-based programme can be considered.*
2. You are public health professional in a country where 95% of the total births occur in hospitals (only resident mothers are considered in the programme). You are asked to assess the prevalence of neural tube defects among the population. Which type of surveillance programme would you use? Why?
    - *Possible response:*
        - *Because most of the births in hospitals are to resident mothers, the programme would be considered a population-based surveillance programme. The programme could implement a way to capture the 5% of births from resident mothers that occur at home or at other facilities, which would help in the referral of children to services.*

## Evaluation questions 1

Expected time: 30 minutes

Correct answers are presented in **bold**.

1. Define congenital anomalies.

**Answer:** Abnormalities of body structure or function that originate during intrauterine life and may be evident prenatally, at birth, or later in life.

2. Define public health surveillance.

**Answer:** Ongoing, systematic collection, analysis and interpretation of health data for public health purposes and the timely dissemination of public health information for assessment and public health response to reduce morbidity and mortality.

3. What is a primary goal of a congenital anomalies surveillance programme?

**Answer:** To provide valid and timely data that can guide the development of prevention initiatives for congenital anomalies.

4. Which of the following is NOT an objective of a surveillance programme for congenital anomalies?

    a. To monitor trends in the prevalence of different types of congenital anomalies in a defined population

    b. To collect systematic, valid and timely data on congenital anomalies

    c. To detect clusters of congenital anomalies (outbreaks)

    d. **To medically treat individuals with congenital anomalies**

    e. To refer affected infants to appropriate services in a timely manner

    f. To disseminate findings and interpretations to appropriate partner organizations and government agencies in a timely fashion

    g. To provide a basis for epidemiologic research and prevention programmes

    h. To allow evaluation of the prevention programme

5. What are two main types of congenital anomalies surveillance programmes?

**Answer:** Population-based, hospital-based

6. True or **false**: Two minor anomalies are considered a major anomaly.

7. **True** or false: In some cases, internal anomalies have external manifestations that allow the observer to suspect its diagnosis.

8. What is an example of a risk factor associated with congenital anomalies?

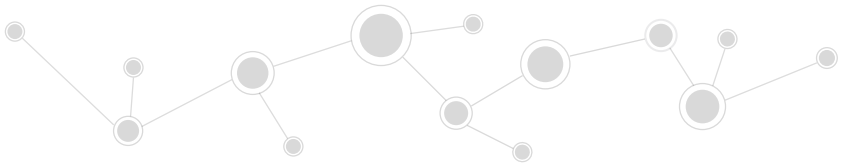
    a. Good nutrition

    b. Healthy weight

    c. **Folate insufficiency**

# INTRODUCTION TO PLANNING ACTIVITIES AND TOOLS

MODULE 2



Expected time: 6.5 hours

## Objectives

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

- identify the basic elements and importance of legislation to support congenital anomalies surveillance;
- identify two types of reporting related to congenital anomalies surveillance legislation;
- describe voluntary and mandatory reporting, and their advantages and disadvantages;
- identify the importance of partnerships to support congenital anomalies surveillance;
- identify potential types of partners;
- describe the roles partners can play in the surveillance of congenital anomalies;
- identify ways to engage partners in surveillance work;
- identify the purpose and components of logic models;
- understand the benefits of creating a logic model;
- develop a logic model;
- describe how logic models help guide programme planning and evaluation.

*Note*: This module is linked to Chapter 2 of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*.

## Logic models

Expected time: 2.5 hours

### Script/key points

- We are now going to discuss logic models and how they can be helpful tools when planning, implementing and evaluating congenital anomalies surveillance programmes.
- Logic models are graphic representations of how a surveillance programme will work.
    - Logic models can not only identify resources that are available and those that are needed, but also identify potential activities and outcomes. Logic models can outline the order in which these activities would occur, and how the outcomes would be achieved.

- Most logic models will include the following components: resources, activities, outputs, outcomes and impact.
  - Resources: What resources currently exist? What resources are needed?
  - Activities: What activities are required for the surveillance programme to function (purpose and reasons needed)? The activities will differ depending on the intended audience. For example, one activity can be to identify partners, while another could be training hospital staff on how to collect data.
  - Outputs: What are the expected outputs that will result from the activities (e.g. training programmes, tangible products)?
  - Outcomes: What are the short-term, intermediate and long-term outcomes for the programme (e.g. benefits for the population, improvement of health)?
  - Impact: What is the impact that you want the programme to ultimately have?

- Also consider how the surveillance programme might improve the quality of life of affected individuals. **Ask participants:** What are some ways in which a surveillance programme might improve the quality of life for the population at risk?
  - *Possible responses:*
    - *Identifying gaps in services*
    - *Increasing the number of services available*
- For examples of information to include in a logic model, please refer to Appendix D in WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*.

## Begin Activity 2.1

- **Ask participants to refer to their workbooks, page 2.**
- Have participants complete a logic model for development of a congenital anomalies surveillance programme in their country, using the worksheet provided in their workbooks. After they have had time to complete the logic model, display the following completed logic model and discuss components with the group.

| Resources | Activities | Outputs | Short-term 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: |
| • Staff with the necessary skills<br>• Funding<br>• Infrastructure<br>• Partnerships<br>• Leadership support<br>• Legislative support<br>• Tools for data collection and analysis<br>• Identification of existing resources and anticipation of needed resources<br>• Identification of champions | • Develop surveillance system<br>• Identify goals<br>• Develop and distribute baseline survey for situational analysis report<br>• Identify appropriate stakeholders<br>• Select sites<br>• Develop and implement surveillance protocol with uniform guidelines<br>• Establish pilot for congenital anomalies surveillance<br>• Assess data quality and utility<br>• Engage partners<br>• Create task force<br>• Advocate<br>• Establish evaluation and monitoring for each step of the programme | • Implement surveillance system<br>• Produce reports and recommendations<br>• Create an upgradeable model surveillance programme<br>• Identify risk factors and prevent those congenital anomalies that have modifiable ones | • Uniform national implementation of a surveillance programme<br>• Enhancement of knowledge<br>• Provision of data to develop policies<br>• Improvement of necessary infrastructure to manage congenital anomalies | • Improved quality of life for affected individuals and their families<br>• Reduction in mortality in children aged under 5 years<br>• Reduction in preventable congenital anomalies |

## Legislation

Expected time: 1 hour

### Script/key points

We are now going to discuss the types of reporting that are used for congenital anomalies surveillance. There are two types of reporting: mandatory and voluntary.

- Mandatory reporting means that staff from participating facilities, such as hospitals, are required to keep a log and report all cases of live births and stillbirths with congenital anomalies to the surveillance programme, within a specified timeframe and in a specified format. Mandatory could mean mandated by either normative or legislative regulations.

- **Ask participants:** What might be some benefits of mandatory reporting? **Allow participants to answer**.
  - *Possible responses:*
    - *Provides structured/reliable data*
    - *Provides countries with evidence of the need to invest in sustainable congenital anomalies prevention programmes*
    - *Provides timely information to help develop policies to distribute resources for babies born with congenital anomalies*
    - *Facilitates the analysis of prevalence and trends for monitoring congenital anomalies*
    - *Informs legislation that permits reporting of private health information for public health*
    - *Provides access to data*

- **Ask participants:** What might be some unintended consequences of mandatory reporting? **Allow participants to answer**.
  - *Possible responses:*
    - *Increased workload*
    - *Reduced quality of services because time spent may be taken away from other services*
    - *Mandatory reporting does not guarantee that staff will comply*
    - *Mandatory reporting does not guarantee data quality*
- Although there are some disadvantages to mandatory reporting, it is important to identify these disadvantages, in order to find the proper solution for them.
- Voluntary reporting means that staff from facilities participating in the surveillance programme, such as hospitals, are encouraged by the officials responsible for the programme to keep a log and report all live births and stillbirths with congenital anomalies to the surveillance programmes, but they do not have to comply.

- **Ask participants:** What might be some benefits of voluntary reporting? **Allow participants to answer.**
  - *Possible response:*
    - *Voluntary reporting might be a good starting point that can lead to mandatory reporting*

- **Ask participants:** What may be some disadvantages to voluntary reporting? **Allow participants to answer.**
  - *Possible responses:*
    - *Unstructured/unreliable data*
    - *Potential for skewed prevalence trends due to underreporting or overreporting of data*
    - *Uneven distribution of resources, based on actual need*
    - *Reporter bias*
- Neither mandatory nor voluntary reporting will be successful if the reporting process does not include feedback to the reporting staff. Each report should be acknowledged, and missed or incomplete information should be corrected.
- **Ask participants whether a mandatory or a voluntary reporting system would or would not work in their countries and why. Ask participants to provide responses aloud, and write responses on the flipchart.**

## Privacy and confidentiality issues

Expected time: 30 minutes

### Script/key points

- We are now going to discuss privacy and confidentiality as they relate to patient data protection and to data acquisition and management in public health surveillance.
- Privacy and confidentiality laws vary by country. It is important to know the laws in the country in which you work.
  - There could be standards on how to collect, use, disseminate and protect information.
- All surveillance personnel involved with data collection, management and dissemination may need to sign confidentiality agreements.
- The three components of patient data protection are privacy, confidentiality and security.
- Privacy is an individual's right to control the collection, use and disclosure of their identifiable health information. This relates to both the parents and the fetus or neonate.

- **Ask participants**: What is one way to protect the privacy of health information? **Allow participants to answer, and discuss.**
  - *Possible response:*
    - *Each fetus or neonate could be assigned a unique identifier, such as a numeric code, to protect their privacy*
- Confidentiality is an individual's right to have their identifiable health information kept secure. These data must be accessible only to health-care providers and those directly involved in the surveillance programme.
- **Ask participants:** What are some ways to ensure confidentiality? **Allow participants to answer and discuss.**
  - *Possible responses:*
    - *Remove all identifiers*
    - *Lock and secure files*
    - *Password protect files*
    - *Require signed confidentiality agreements for surveillance workers*
- Security refers to the safeguards and practices designed to protect data systems against unwarranted disclosure, modification or destruction. All individuals have the right to have their health data secured.
- We are now going to briefly discuss privacy, confidentiality and security issues for photographs that might be used as part of a surveillance programme for congenital anomalies.
  - Photographs can serve as diagnostic tools, like other tools available for diagnosis (e.g. echocardiogram).
    - Photographs of the congenital anomaly are taken to increase the quality of information transmitted to a surveillance programme.
  - Photographs should always be transferred in encrypted files and stored securely.
  - When photographs of congenital anomalies are taken, countries may require that parents sign an authorization form. Requirements for authorization forms may be different in different countries.
  - For recommendations on how to take photographs to be used for the surveillance programme, please refer to Appendix J in WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4).*

## Begin Group discussion 2.1

- **Tell the group:** You are preparing a protocol for a congenital anomalies surveillance programme in your country.
- **Ask participants:** What kind of questions would you need to ask regarding privacy and confidentiality before beginning the surveillance programme?
  - *Possible responses:*
    - *What type of data will be collected? Why?*
    - *How will the data be collected? On paper, electronically or both?*

- *Who will have access to the data?*
- *How will the data be used?*
- *Where will the information be stored and secured?*
- *How long does the law require it to be archived?*

- **Ask participants:** What type of information regarding laws do you need before beginning the surveillance programme?
  - *Possible responses:*
    - *Laws available in a country for protection of medical data*
    - *Country and local laws for confidentiality*
    - *Laws on public health surveillance*

## Partnerships and funding

Expected time: 1.5 hours

### Script/key points

- We are now going to discuss the important role that partners play in surveillance. When implementing and maintaining a surveillance programme, having a wide variety of partners committed to the programme's success is essential.
- Partners can help a surveillance programme succeed and be sustained, by developing goals, policies and access to funding, and providing access to data.
  - They may include health-care professionals and non-profit organizations.
  - **Ask participants to give examples of partners in their countries who will be important to include in the planning and implementation of a surveillance programme. Ask participants to provide responses aloud, and write responses on the flipchart.**
- For a list of ideas of potential partners and their roles, please refer to Appendix E in WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*. It is important to involve partners from the start of the programme, to fully benefit from their expertise.
- Now let's consider some ways partners might help a surveillance programme to succeed. **Allow participants to provide responses aloud, and write responses on the flipchart.**
  - *Possible responses:*
    - *Developing goals and objectives for the surveillance programme*
    - *Ensuring implementation and long-term sustainability*
    - *Developing policy measures and moving measures through appropriate channels*
    - *Identifying champions that will maintain the interest in the programme*

## Begin Activity 2.2

- **Ask participants to refer to their workbooks, page 3.**
- **Have participants complete a stakeholder's worksheet for development of a congenital anomalies surveillance programme in their country. Tell participants:** We are now going to complete a stakeholders' worksheet. Think of potential roles for each partner and how they will contribute to your surveillance programme.

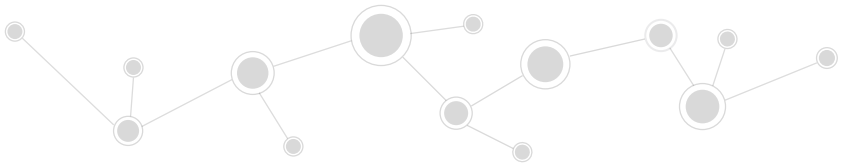
| Sample worksheet: Partners/stakeholders | |
|---|---|
| **Partners/stakeholders** | **Potential roles** |
| Ministry of health | Set policies and regulations for health-care services and delivery |
| Hospitals | Serve as data sources |
| Community health workers | Serve as potential data sources; help increase awareness of congenital anomalies, risk factors, other. |
| Primary health centres | Data sources; source for prevention and outreach activities |
| Universities | Opportunity to help increase knowledge |
| International organizations | Provide advocacy; technical assistance and expertise |

- **After they have had time to complete the worksheet, display the following completed worksheet and discuss components with the group.**

*Note*: Before participants begin the activity, the facilitator can choose to complete one row with the participants to show them an example of how it is completed. These are *examples*, and participants can include other stakeholders appropriate to their needs.

## Script/key points

- We are now going to discuss the importance of communicating with different types of partners to keep them engaged in the surveillance process. The following table provides ideas for different communication messages and strategies for disseminating these messages to a variety of partner audiences.



| Likely users of outputs | Communication message | Dissemination strategy | Evaluation |
|---|---|---|---|
| Ministries of health | • Prevalence of congenital anomalies is high<br>• Need for continued support for surveillance programme<br>• Need for identification of specialty services<br>• Need for prevention activities and intervention policies | • Report<br>• Press conferences | • Evaluation of report for clarity of messages<br>• Number of places that provide services<br>• Effect of prevention policies |
| Hospitals and, if relevant, hospital associations and clinics | • Importance of valid and reliable reporting, and feedback on their performance<br>• Number of reports; distribution of reported congenital anomalies by hospital<br>• Consistency of reports/data quality | • Report<br>• Datasets<br>• Newsletters for staff<br>• Information given at training sessions, workshops and seminars | • Evaluation of report<br>• Pre- and post-tests at trainings or workshops |
| Champions | • Importance of their support<br>• How they can promote and support implementation of a surveillance programme | • Report<br>• Letter | • Number of champions participating in the programme |
| Community health workers/community health volunteers | • Importance of notification of congenital anomalies seen in the community and with home births, to provide referrals to clinics or ambulatory care centres | • Information given at training sessions, workshops and seminars | • Pre- and post-tests at training sessions or workshops |
| Congenital anomalies associations, foundations and other nongovernmental organizations | • Need for improved quality of care for individuals living with disabilities | • Report<br>• Media<br>• Educational materials<br>• Newsletters | • Use of report<br>• Requests for educational materials |
| International organizations | • Information about surveillance programme implementation, global impact, how they can support efforts | • Report<br>• Media, written materials<br>• Newsletters | • Use of reports<br>• Requests for educational material |
| Medical schools/ research agencies | • Importance of congenital anomalies mortality and morbidity<br>• Importance of congenital anomalies surveillance<br>• Prevention of congenital anomalies | • Curricula<br>• Coursework<br>• Seminars | • Number of medical schools/ agencies integrating the message |

## Communicating with parents

Expected time: 30 minutes

### Script/key points

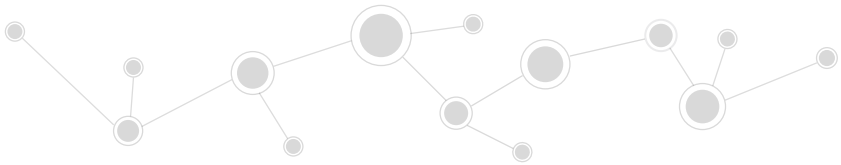
- Public health surveillance is not meant to provide clinical care recommendations or guidance, and it is inappropriate for surveillance staff to communicate diagnoses, prognoses or clinical guidance to parents or other family members. Only the family's primary health-care provider can communicate on clinical issues. However, it is important for surveillance staff to understand that having a child with a congenital anomaly is a very sensitive issue and it is essential to protect confidentiality.
- Only health-care providers who have been appropriately trained should communicate the diagnosis and prognosis of a congenital anomaly to parents.
  - For more information about this topic, please refer to Appendix F of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*.
- Parents can be involved in the surveillance programme.
- **Ask participants to give examples of ways in which parents can be involved**.
  - *Possible responses:*
    - *Parents can be effective advocates of the surveillance programme and help create and maintain interest among government officials and others, on the importance of having a congenital anomalies surveillance programme.*
    - *Parents can be advocates to help implement a prevention strategy when one exists (e.g. folic acid).*
    - *Parents can also help advocate for new services or improvement of current services for children born with congenital anomalies.*

## Evaluation questions 2

Expected time: 30 minutes

Correct answers are presented in **bold**.

1. **True** or false: A logic model is a graphic representation of how the surveillance programme will work.
2. Which of the following is NOT a major category in the development of logic models?
   a. Resources
   b. Activities
   c. **Recruitment**
   d. Outputs
3. Partnerships are important because they can help to … (*List at least one response discussed during the training*).



**Answer:** *(Any of the following is correct)* Develop goals and objectives for the surveillance programme; develop policy measures and shepherding measures through appropriate channels; identify funding support; implement and/or manage the programme.

4. **True** or false: Mandatory reporting of congenital anomalies and regular feedback on reported information can improve data quality.

5. Privacy and confidentiality are components of the laws and regulations related to _____, _____, _____ and _____ of personal information. (*List the four responses discussed during the training*)

**Answer:** collection, use, dissemination, protection.

6. How can surveillance data be used effectively when assessing congenital anomalies? (*List two of the responses discussed during the training*)

**Answer:** *(Any two of the following are correct)* Identifying clusters, populations at risk, potential risk factors, disease burden, trends of congenital anomalies; programme evaluation, planning, implementing, and evaluating evidence-based interventions; motivating action in the community; informing policy-makers and government officials; informing clinical/public health practitioners, nongovernmental organizations, and the public; identifying and referring children with special needs to services.

7. Which of the following is NOT a systematic approach for collecting data on congenital anomalies?

   a. Paper-based collection

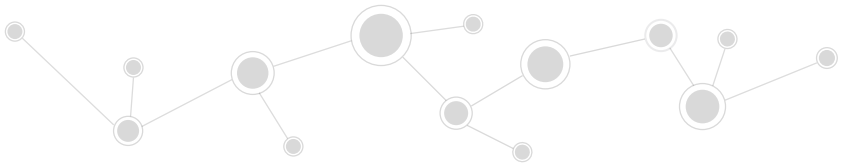
   b. Electronic collection

   c. **Word of mouth**

8. **True** or false: Parents can also help advocate for new services or improvement of current services for children born with congenital anomalies.

# INTRODUCTION TO SURVEILLANCE APPROACHES

MODULE 3



Expected time: 15.5 hours

## Objectives

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

- describe population coverage as used in surveillance of congenital anomalies;
- describe population-based surveillance;
- describe hospital-based surveillance;
- compare and contrast population-based surveillance versus hospital-based surveillance;
- describe the three types of case ascertainment;
- describe the advantages and disadvantages of active and passive case ascertainment;
- describe case-finding in congenital anomalies surveillance;
- identify the differences between a single data source and multiple data sources;
- describe considerations for inclusion criteria used in the surveillance of congenital anomalies;
- identify the congenital anomalies to include in the programme;
- describe considerations for inclusion of particular congenital anomalies;
- describe how data can be used for decision-making;
- identify considerations for communicating and disseminating surveillance data;
- identify different communication methods;
- identify variables that will reflect the objectives of the programme;
- describe core variables;
- describe additional variables;
- identify sources to collect variables;
- understand the elements of quality and value in congenital anomalies surveillance;
- recognize the importance of shared procedures and protocols;
- identify selected processes and data elements that can be tracked for ongoing quality improvement;
- understand the meaning of birth prevalence;
- compute birth prevalence;
- comment on data that are similar to those presented in the exercises.

*Note*: This module is linked to Chapter 3 of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*.

## Epidemiology

Expected time: 15 minutes

### Script/key points

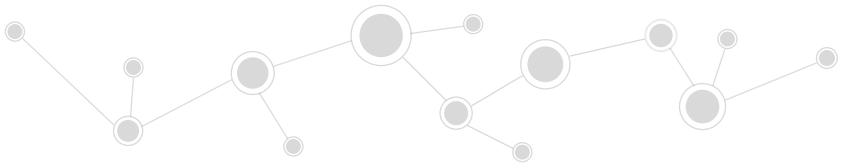
- Now let's define epidemiology in the context of a congenital anomalies surveillance programme.
- **Ask participants:** Can anyone define epidemiology? **Ask participants to provide responses aloud, and request a volunteer to write responses on the flipchart.**
  - *Possible responses:*
    - *The study of the frequency and distribution of health events and their determinants in human populations, and the application of such research to the prevention and control of health problems.*
    - *The study of how diseases vary according to person, time and place. When the distribution of prevalence is not uniform according to person, time and place, you are able to identify high-risk groups for prevention or research purposes.*
- Epidemiology is the study of the distribution and determinants of health-related states or events in specified populations and the application of this study to control health problems.
- Surveillance is one of the methods used by epidemiological investigations.

## Population coverage

Expected time: 2 hours

### Script/key points

- We will now discuss why birth prevalence and not incidence is the recommended form of reporting the frequency of congenital anomalies.
- First, let's distinguish between incidence, prevalence and birth prevalence of congenital anomalies.
  - Incidence refers to all new cases of congenital anomalies within a time period and within a given population. Incidence is not usually used to describe the occurrence of congenital anomalies. It is not possible to capture a true incidence, since not all conceptuses (products of conception at any time between fertilization and birth) can be ascertained – for example, because of early spontaneous abortions.
  - Prevalence is a measure of the total number of existing cases of a disease or condition for a given point in or period of time, and in a given population, regardless of whether or not they are new cases.
  - The following expression is used to calculate the birth prevalence of congenital anomalies, with the assumption that both live births and late fetal deaths are being captured:



Birth prevalence = $a/b \times 10\,000$

$a$ = Number of live births and late fetal deaths (stillbirths) with a specific congenital anomaly (e.g. spina bifida) counted among the source population in a given year

$b$ = Number of live births and late fetal deaths (stillbirths) during the same year

- In order to calculate total prevalence in a population-based congenital anomalies surveillance system, the numerator includes cases among live births, known fetal deaths and pregnancy terminations (if possible). The denominator comprises the total number of subjects evaluated, including those cases in the numerator. However, if the total number of fetal death subjects or elective terminations of pregnancy for fetal anomaly (ETOPFA) is unknown, the total number of live births (and stillbirths if available) can be used. This is because the numerator of ETOPFA (and/or stillbirths) is relatively small, compared with the number of live births, and its exclusion from the denominator has little effect on the prevalence estimate. Spontaneous abortions (also called miscarriages) are not included in either the numerator or the denominator because it is practically impossible to assess the total number of spontaneous abortions.

- **Ask participants:** How do you define birth prevalence in congenital anomalies surveillance?
  - Birth prevalence includes a specific timeframe; the estimate is for babies being born during a defined period of time.

- **Ask participants:** In your country, what information is collected through your vital statistics (vital registration) programmes? Are data for congenital anomalies collected in your country?
- There are two main types of surveillance programmes that may be used for congenital anomalies surveillance: population-based and hospital-based programmes.
- Population-based programmes collect data from the entire population (all mothers giving birth either within or outside of a maternity facility, in a defined geographic area). The denominator would be the total number of births (live births and stillbirths).
  - Inclusion is typically defined by the maternal residence.
  - For example, a woman giving birth in the catchment area but residing elsewhere would not be considered a resident. Therefore, the fetus or neonate would not be included for surveillance or in the total number of births.
  - Alternatively, a woman who gives birth outside the catchment area, but who maintains a permanent residence in the catchment area, would be considered a resident mother. Therefore, the fetus or neonate would be included for surveillance and in the total number of births.
    - It is important to note that the surveillance system would need a mechanism to identify and exclude records of non-eligible mothers/births.

- Also, the surveillance system would need a mechanism to capture births to residents identified with congenital anomalies outside of the catchment area, such as in treatment centres, paediatric wards, rehabilitation centres or any other health facility. Therefore, more resources are needed to ascertain all fetuses or neonates with congenital anomalies. Programmes will need to have a mechanism in place to ensure that fetuses/neonates identified with a congenital anomaly and reported from more than one source are not counted twice (e.g. having a report from the delivery hospital and a report from a treatment centre).

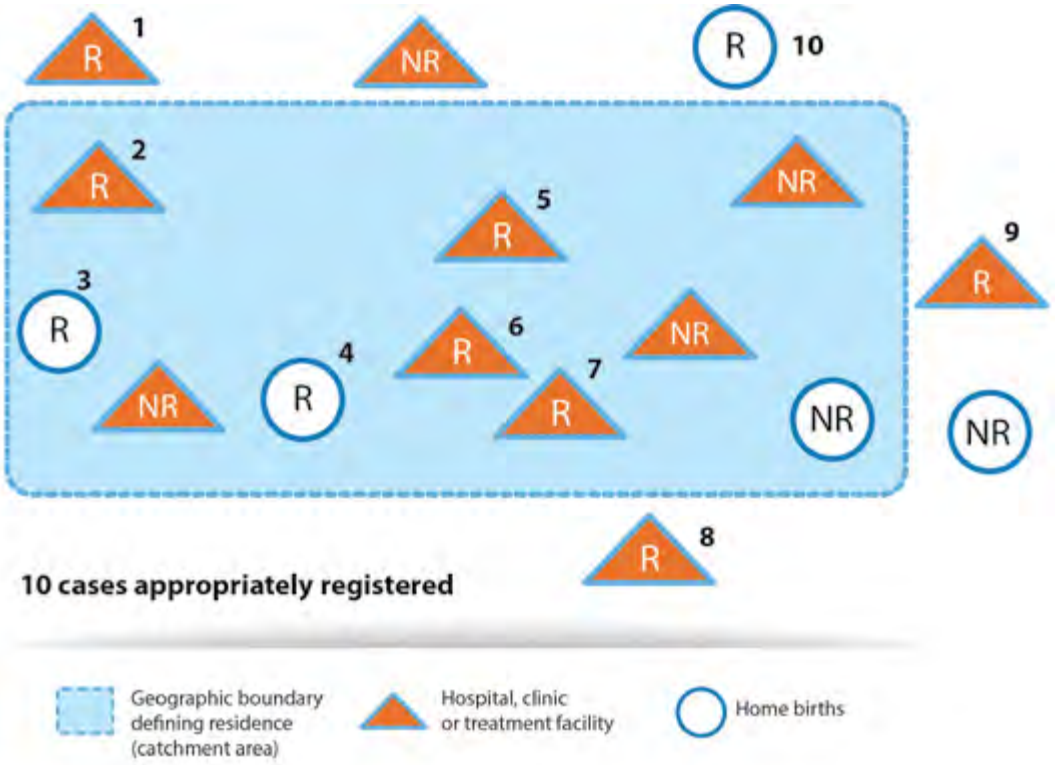
- Hospital-based programmes collect data from births that occur in pre-selected hospitals within a well-defined geographic area.
  - Fetuses and neonates delivered at home would not be included, even if identified later in life in participating hospitals.
  - Hospital-based programmes may be vulnerable to referral bias, since some hospitals may have particular characteristics.
    - Hospital births may only be common among families with higher socioeconomic status.
    - Hospital births may be more common among women who have been identified as being at high risk for a complicated birth.
  - Prevalence estimates may not be accurate if they rely solely on hospital-based programmes.
  - In countries where nearly all births take place in the hospital and all hospitals participate, those hospital-based programmes can closely approximate true prevalence.
- A country's selection between population-based and hospital-based surveillance programmes will depend on available resources and capacity. Both population-based and hospital-based programmes may have a short period of time to diagnose a condition in a neonate, since neonates are discharged within hours or days following birth. Congenital anomalies that are commonly identified after the neonatal period, such as congenital heart defects, are often not diagnosed in a hospital setting, unless the programmes have resources to conduct follow-up with children after discharge from the birth hospital.
- If a hospital-based programme is being implemented, selection of hospitals may include:
  - Hospitals with a willingness to participate
  - History of reporting other conditions to the ministry of health
  - Hospitals that have a log book in place with an indicator of the presence or absence of a congenital anomaly in the delivery ward and other sources (such as paediatric wards and neonatal wards), as appropriate
  - Hospitals with a high number of births

*Note*: It will be important to closely monitor the quality of reporting of participating hospitals.

## Begin Activity 3.1

- **Ask participants to refer to their workbooks, page 4, and ask them to complete the following activity.**
- **Show illustration (A) to the group without the numbers and the answers.**

**Illustration A**



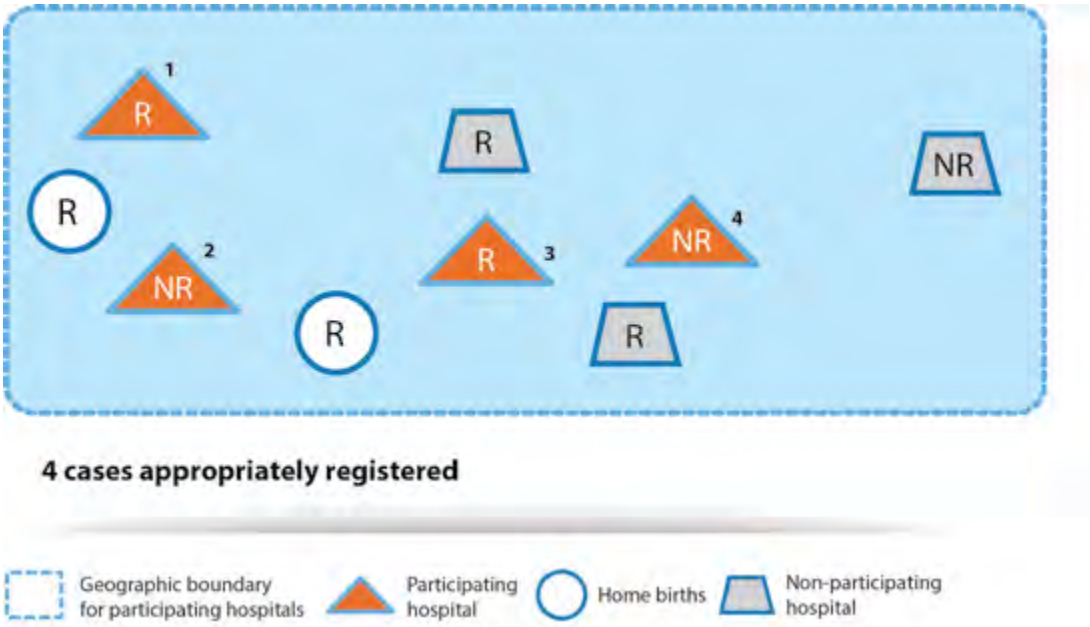

**R** = fetus or neonate with congenital anomaly whose mother is a resident.

**NR** = fetus or neonate with congenital anomaly whose mother is a non-resident.

- **Ask participants to complete the following questions (included in their workbooks, page 4). Allow 10 minutes and then discuss the responses as a group.** (Responses are in *italic*)
  - Does this figure represent a population-based or hospital-based surveillance programme? *(The figure represents a population-based programme.)*
  - What is the numerator (cases that should be registered) in this surveillance programme? *(10)*
  - Is maternal residence important for this type of surveillance? *(Yes it is; all neonates with congenital anomalies should be included if the mother is a resident.)*
  - Are home births with congenital anomalies counted in this type of surveillance? *(Yes, if the mother is a resident. No, if the mother is not a resident.)*

- **Show illustration (B) without the numbers and the answers to the group and ask them to complete the corresponding questions, tell them that the illustration and questions are in their workbooks, page 5.**

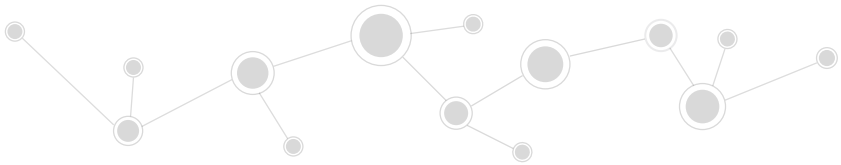
**Illustration B**




**R** = fetus or neonate with a congenital anomaly whose mother is a resident; included if the fetus or neonate is identified at a participating hospital.

**NR** = fetus or neonate with a congenital anomaly whose mother is a non-resident; included if the fetus or neonate is identified at a participating hospital.

- **Ask participants to complete the following questions. Allow 10 minutes and then discuss the responses as a group.** (Responses are in *italic*)
  - Does this figure represent a population-based or hospital-based surveillance programme? *(The figure represents a hospital-based programme.)*
  - What is the numerator (cases that should be registered) in this surveillance programme? *(4; births that occur in the four participating hospitals shown in the figure.)*
  - Is maternal residence important for this type of surveillance? *(No, it is not; neonates with congenital anomalies at participating hospitals should be included regardless of the mother's residency.)*
  - Are home births with congenital anomalies counted in this type of surveillance? *(No, they are not included; only births that occur in participating hospitals should be included.)*



## Case ascertainment

Expected time: 45 minutes

### Script/key points

- We will now spend some time discussing methods for case ascertainment.
- There are three methods for case ascertainment: active, passive and hybrid (a combination of passive and active). Each method has advantages and disadvantages.
- In active case ascertainment, surveillance personnel are hired and trained to abstract data from all data sources.
  - Abstractors regularly visit or have electronic access to participating institutions, such as hospitals and clinics.
  - Abstractors actively review multiple data sources such as log books, and medical, discharge and deaths records, to identify cases.
  - For those fetuses or neonates identified in the log books as having a congenital anomaly, abstractors should request maternal and infant medical records to record relevant information onto a reporting form.
    - Medical records need to contain relevant information in a format that can be readily identified and abstracted easily by the abstractors, who often have limited medical background.
  - Advantages
    - This method usually improves case detection and case reporting, and improves data quality because more extensive clinical detail is collected.
  - Disadvantages
    - This type of case ascertainment requires considerable surveillance programme resources and personnel.
    - The burden of work is placed on surveillance personnel.
- In passive case ascertainment, congenital anomalies are reported directly to the surveillance programme.
  - The information that is reported to the surveillance registry typically is not verified by direct abstraction of the medical record by surveillance personnel.
  - Advantages
    - This type of case ascertainment is less expensive because fewer surveillance programme resources and personnel are required.
  - Disadvantages
    - The burden of reporting falls on hospitals, clinics or other sources that may require time and effort from already busy staff.
    - Case detection and case reporting can be compromised because of the following:

- Not all cases are reported, leading to an underestimate of the number of cases
- Incomplete documentation, resulting in less detail on each case
- Less timely reporting, leading to a delay in analyses and communication
- Personnel may have varying levels of training and commitment, leading to inaccurate information
- Stimulated/incentivized reporting may result in overestimation of certain congenital anomalies
- Variation in reporting over time may generate spurious trends or hide real ones

- Hybrid case ascertainment uses a combination of passive and active reporting systems.
  - This method may use active case ascertainment to gather more detailed case information for specific congenital anomalies or to verify passive reporting for a percentage of all reported congenital anomalies, as a quality control measure. Hybrid ascertainment methods enable review of probable cases during follow-up and provide a definitive diagnosis, thereby reducing the number of births misclassified as cases.
  - For example:
    - A surveillance programme can use active ascertainment of neural tube defects to gather more detailed case information in a timely manner, while also using passive ascertainment of the other congenital anomalies under surveillance.
    - A programme can use passive reporting with active follow-up verification of certain congenital anomalies to verify the accuracy of data submitted and gather more data.
    - A programme can conduct active case ascertainment from some sources, such as birthing hospitals, and accept passive reporting from other sources, such as cytogenetic laboratories.
- Regardless of the method used, it is helpful for each participating hospital, clinic or participating site to identify a programme champion. This is likely to increase participation of data source units and services, and facilitate training of other health-care personnel and any new personnel. A champion is someone who advocates for the programme, and also takes a leading role in organizing, collecting data and overseeing the programme. A champion is usually a staff member at a site that is committed to the programme. This person is often a nurse or doctor. It is important to identify champions and keep them motivated and interested.

## Begin Group discussion 3.1

- **Have participants discuss which methodology may be appropriate for their country/countries, given their resources and capacity.**
- **Ask participants:**
  - How would you define a champion?
  - What would you say his or her role would be?
  - *Possible response:*
    - A person who will advocate for congenital anomalies surveillance, and motivate others to participate in these activities.
  - Can you think of a champion(s) from your country who will help support the development or expansion of a surveillance programme?
  - *Possible responses:*
    - *Doctors – e.g. paediatricians or neonatologists*
    - *Midwives*
    - *Nurses – e.g. from neonatal ward, surgery ward or paediatric ward*
    - *Technicians – e.g. from medical records*
  - How will you engage this champion or these champions?
  - *Possible responses:*
    - *Training sessions*
    - *Defined roles*
    - *Feedback*

**Allow time for discussion, and write responses on the flipchart.**

## Case-finding

Expected time: 1 hour

### Script/key points

- **Ask participants:** What are some potential data sources available for identifying cases? For example, in a hospital-based reporting system, which hospital units/ departments are reporting congenital anomalies? **Ask participants to provide responses aloud, and write responses on the flipchart.**
  - *Possible responses:*
    - *Delivery units*
    - *Paediatric, neurosurgery, orthopaedics units*
    - *Surgery units*
    - *Vital records (birth and death certificates)*
    - *Neonatal care units*
    - *Pathology units*

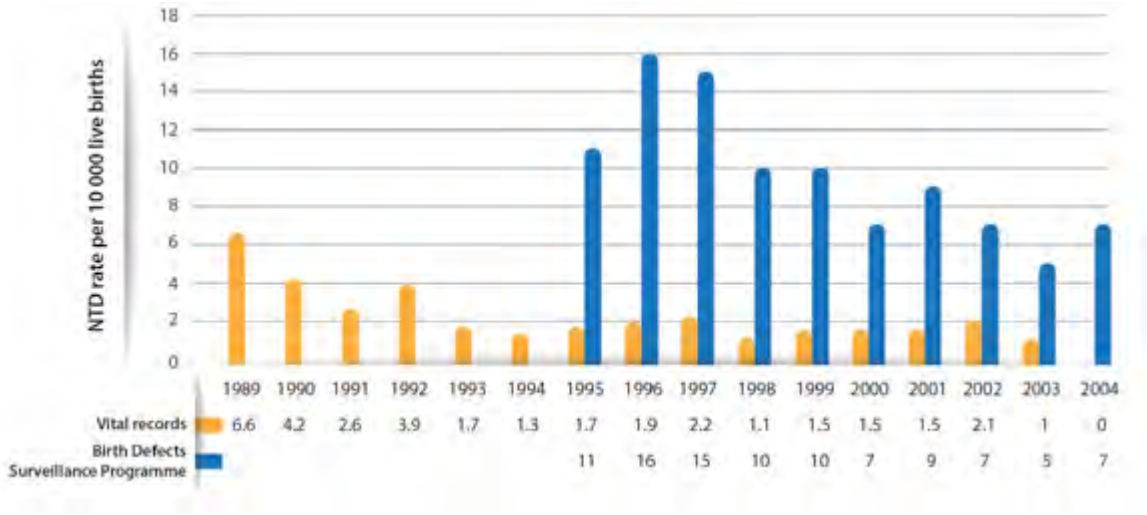
- Using multiple data sources, such as data from units other than the delivery unit, may improve data quality by increasing the amount and level of information available for diagnoses and case detection.

- **Ask participants:** What are some other potential data sources available for identifying congenital anomalies in a population-based reporting system? **Ask participants to provide responses aloud, and write responses on the flipchart.**
  - *Possible responses:*
    - *Surgical hospitals or any other hospital that provides treatment services*
    - *Treatment clinics*
    - *Rehabilitation clinics*
    - *Genetic laboratories*
    - *Clinics*
    - *Foundations and associations that help children born with congenital anomalies*
    - *Health insurance companies' databases*
    - *Vital records (birth and death certificates)*
    - *Health information systems*
    - *Hospital reporting systems*
    - *Pathology units*

## Begin Group discussion 3.2

- **Show participants Fig. 3.1. Ask participants:** What does this figure demonstrate?
- **Allow participants time to write responses. Encourage group discussion about responses before reviewing.**

**Fig. 3.1. Prevalence of neural tube defects by source**




NTD: neural tube defects.

*Source*: Birth Defects Surveillance Programme Puerto Rico Department of Health, and Auxiliary Secretariat for Planning and Development, San Juan, Puerto Rico.

- *Possible responses:*
  - *This example demonstrates how using multiple data sources increased the case detection compared to only using vital records.*
  - *It shows 16 years of vital records data and 10 years of data from multiple sources.*
  - *The data are for neural tube defects.*
  - *The birth prevalence of neural tube defects has declined over time.*
  - *Multiple sources identify more cases with neural tube defects.*
  - *Vital records usually include data at birth and multiple sources may include other age cut-off values that allow for identification of cases later in life.*

## Case inclusion

Expected time: 45 minutes

### Script/key points

- **Ask participants:** What types of congenital anomalies do you think should be included in a surveillance programme? **Ask participants to provide responses aloud, and write responses on the flipchart.**
  - *Possible responses:*
    - *Major anomalies*
      - *External major anomalies (e.g. neural tube defects)*
      - *Internal major anomalies (e.g. heart defects)*
    - *Minor anomalies*
      - *External minor anomalies (e.g. pre-auricular tags)*
    - *Both major and minor anomalies*
- **Ask participants:** Why did you choose these? **Ask participants to provide responses aloud, and write responses on the flipchart.**
  - *Possible responses:*
    - *Availability of technology*
    - *Availability of specialists*
    - *Capacity to diagnose*
    - *Complexity of the anomaly*
    - *Ability to prevent the congenital anomaly*
    - *Availability of evidence-based prevention strategies*
    - *Ability to collect high-quality data*
    - *Ability to monitor prevention interventions*
    - *Ability to show public health impact*
- Good ascertainment provides better data quality. Identifying the capacity of the programme to ascertain cases will help you choose how many and which congenital anomalies to include.

## Begin Group discussion 3.3

- **Have participants discuss which specific congenital anomalies they think they could include in a surveillance programme in their countries, and why they would include these.**
- **Provide the following considerations to the participants to help guide the discussion.**
  - Feasibility of prevention
    - Are there available evidence-based prevention strategies?
    - Can surveillance data assist in evaluating prevention strategies?
  - Availability of staff and staff capacity
    - Are there sufficient staff to conduct surveillance?
    - Is training available for surveillance staff?
  - Availability of testing
    - Are special tests necessary to confirm the diagnosis, for example karyotype?
  - Availability of equipment
    - Is there equipment available that is necessary to diagnose internal anomalies, for example sonograms, X-rays and echocardiograms?
  - Availability of specialists
    - Are there specialists available in the country to make diagnoses?
  - Cost
    - Are there funds available for surveillance?
- Some surveillance systems may not be appropriate for certain congenital anomalies. For example, those congenital anomalies typically identified after the neonatal period may not be captured in hospital-based systems unless there are resources to include children after discharge from the birth hospital.
- If a surveillance programme is specifically designed to evaluate a public health intervention such as folic acid fortification, then the priority may be to measure the related birth outcomes – for example, neural tube defects – as opposed to other unrelated anomalies.
- Incremental costs should be considered. If no extra costs would result from monitoring other anomalies in addition to neural tube defects, consider collecting data on other types of congenital anomalies.
  - Including more congenital anomalies in the surveillance programme will probably require additional resources.
- When starting a new congenital anomalies surveillance programme, consider starting with a small number of easily recognizable major external congenital anomalies with intervention or prevention potential and then expand to include additional congenital anomalies as the programme gains experience, creates awareness in participating facilities, and obtains more resources.

- The list of anomalies will vary, depending on capacity and resources, but typically includes major external anomalies, for example orofacial clefts, neural tube defects and limb deficiencies.
- Detecting many internal structural anomalies, such as unilateral kidney agenesis, requires imaging techniques or other procedures that may not be readily available.
- Additionally, even using the most advanced imaging techniques, the diagnosis of some internal structural anomalies, such as some congenital heart defects, can be very difficult.

## Inclusion criteria

Expected time: 1 hour

### Script/key points

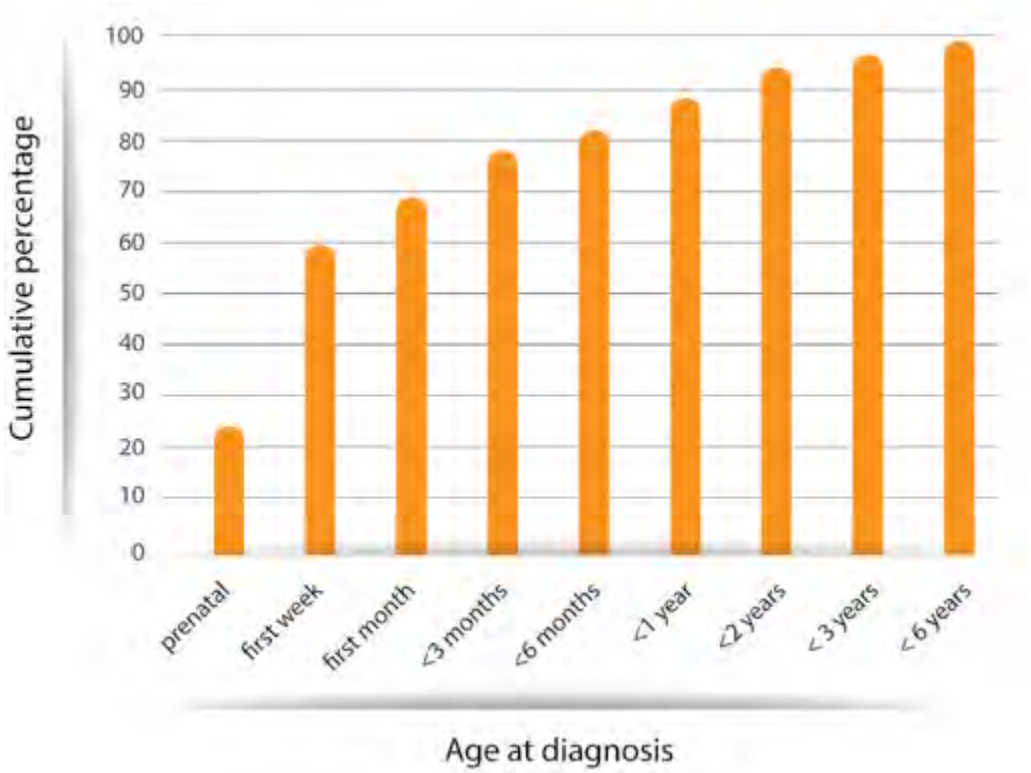
- We will now discuss inclusion and exclusion criteria.
- Inclusion criteria are the specific factors or characteristics that define whether a fetus or neonate with a congenital anomaly is included in the surveillance programme.
- Inclusion criteria are set by individual programmes and most likely will vary between programmes and countries.
- Exclusion criteria are the specific factors or characteristics that define whether a fetus or neonate should not be considered as a case in the surveillance programme.
- **Ask participants:** What are some examples of other inclusion criteria?
  - *Possible responses:*
    - *Age at diagnosis*
    - *Type of pregnancy outcome*
    - *Birth weight/gestational age at delivery*
    - *Maternal residence*
- **Ask participants:** Why is maternal residence important?
  - *Possible response:*
    - *To define the source population in which the cases occur*
- Now let's discuss age at diagnosis. Age at diagnosis is a critical component of the case definition. Many countries may have different age inclusion criteria.
  - The higher the age threshold, the greater the ability to identify additional anomalies. This is especially true for internal congenital anomalies that might not be evident at birth, such as heart defects.
  - The higher the age threshold, the greater a programme's expense because additional resources may be needed for follow-up.
  - Spontaneous abortions should not be included in the numerator or in the denominator. It will be hard to identify congenital anomalies in most spontaneous abortions and almost impossible to have a denominator for them.

- **Tell participants:** Please look at the following graph, showing an example of how age at diagnosis has been used. Researchers examined the age at diagnosis for all congenital anomalies reported to the Western Australian Birth Defects Registry from 2000 to 2001.

## Begin Group discussion 3.4

- **Show participants the following figure, which represents how researchers examined the age at diagnosis for all congenital anomalies reported to the Western Australia Birth Defects Registry from 2000 to 2001.**

**Fig. 3.2. Cumulative percentage of cases of major congenital anomalies by age at first diagnosis**




*Source*: reproduced by permission of the publisher from Bower et al., 2010 *(13)*.

- **Ask participants:** What does this graph demonstrate?
  - *Possible responses:*
    - *Nearly 60% of all major congenital anomalies reported to this registry programme are diagnosed during the first week, nearly 70% during the first month, nearly 90% during the first year, and nearly 100% by the sixth year of life.*
    - *Prenatal diagnosis is not able to identify all congenital anomalies.*
    - *The congenital anomalies that take longer to identify are probably not visible at birth.*
    - *It is important to notice that most congenital anomalies are captured in the first year of life.*
    - *This graph is for all congenital anomalies under surveillance and reported to the programme; if it only included external congenital anomalies, the graph would probably look very different.*

## Inclusion of pregnancy outcomes

Expected time: 30 minutes

### Script/key points

- Let's think about some possible pregnancy outcomes.
- **Ask participants:** What are some examples? **Allow participants to answer.**
  - *Possible responses:*
    - *Live births*
    - *Stillbirths*
    - *Spontaneous abortions/miscarriages*
    - *Elective terminations of pregnancy with fetal anomalies, also known as ETOPFA*
    - *Elective terminations for other reasons*
- If possible, surveillance programmes should aim to ascertain congenital anomalies among all pregnancy outcomes, with the exception of early spontaneous abortions.
  - Including as many types of pregnancy outcomes as possible improves case detection.
  - Inclusion of live births alone is a significant limitation that can lead to unreliable rates and trends, particularly for conditions with a high rate of loss prior to 28 weeks. An example of such a condition is anencephaly.
  - Inclusion of pregnancy outcomes is dependent on a country's ability and resources to detect all or most outcomes.
  - In some countries, it can be difficult to include stillbirths and ETOPFA.
  - If a country has the capacity to ascertain stillbirths, doing so can help provide a more accurate estimate of the prevalence of a condition like anencephaly.

## Begin Group discussion 3.5

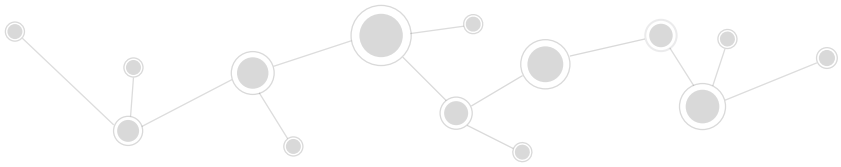
- **Ask the following questions to generate discussion. Write responses on the flipchart.**
  - How are stillbirths defined?
    - *Possible responses: ≥28 weeks of gestation, ≥1000 g if gestational age unknown. However, some countries may define stillbirths as ≥20–22 weeks of gestation or ≥500 g if the gestational age is unknown.*
  - Do you know how "stillbirth" is defined in your country?
  - Are terminations legal or practised in your country?
  - Are terminations allowed if there is a congenital anomaly?
  - If terminations are not legal, will women go to another country to terminate a pregnancy?
  - If autopsies are a common practice in your country, could this help in the diagnosis of congenital anomalies?
  - How would a surveillance programme that included stillbirths and terminations differ from a programme that only included live births?

## Begin Activity 3.2

- **Ask participants to refer to their workbooks, page 6.**
- **Have participants create inclusion and exclusion criteria for population-based or hospital-based surveillance programmes. Ask participants to keep in mind capacity and available data sources. Remind them that the inclusion and exclusion criteria will be different, depending on whether the programme is hospital based or population based. Allow 10 minutes and then discuss the responses as a group.**
  - *Possible responses:*
    - *Inclusion criteria:*
      - *Born with one of the congenital anomalies under surveillance*
      - *Age at diagnosis (up to 1 year of age)*
      - *Pregnancy outcome (live births, stillbirths and termination of pregnancies for fetal anomaly)*
      - *Gestational age (≥28 weeks, or ≥1000 g)*
      - *Maternal residence (born to resident mother)*
    - *Exclusion criteria:*
      - *Born without a congenital anomaly under surveillance*
      - *Older than 1 year of age*
      - *Miscarriage/spontaneous abortion*
      - *Less than 28 weeks of gestation, or <1000 g*
      - *Born to non-resident mother*

## Begin Group discussion 3.6

- **Ask participants the following questions. Allow participants time to write responses. Encourage group discussion about responses before reviewing.**
  - What defines a case?
    - *Response: A case is defined by the surveillance programme's inclusion criteria.*
  - 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.
    - *Response: No, the surveillance programme includes only live births. If the inclusion criteria of the surveillance programme considered stillbirths as cases, then the neonate would be included.*
  - 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.
    - *Response: It will depend on the guidelines of the hospital. If the hospital registers this neonate as being born at the hospital, then he/she would be included. If the inclusion criteria of the surveillance programme have this possibility as a case, then the neonate would be included.*
  - 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.
  - *Response: It will depend on the inclusion criteria of the surveillance programme. In a hospital-based surveillance programme, the neonate would not be included.*
  - 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.
    - *Response:*
      - *In a population-based surveillance programme, the neonate would not be included, because the mother is not from the defined geographic area. However, if the mother has been living in the country within the catchment area for a time period specified in the inclusion criteria, then the neonate would be included.*
      - *In a hospital-based programme, the neonate would be included unless the inclusion criteria state that only residents of the country should be registered.*
  - If a mother, born in another country, who has been living in your country for 3 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.



- *Response:*
  - *In a population-based surveillance programme, if, for example, residence is defined as the mother's primary address during the 3 months prior to pregnancy and the first trimester of pregnancy, the neonate would not be included, because the mother is not from the defined geographical area.*
  - *In a hospital-based programme, the neonate would be included, regardless of the length of time the mother has lived in the country. This is unless the inclusion criteria state that only residents of the country are registered.*

- 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 one month of age, would the infant be included? What if the child is identified at two years of age? Explain your response.
  - *Response: Since the criterion for inclusion is congenital anomalies identified during the first three days of life, the child would not be included. The response is the same if the congenital anomaly is identified at one month or two years of age.*
- How would the exclusion of stillbirths affect your prevalence estimate? For which congenital anomalies would it be important to include stillbirths?
  - *Response: If stillbirths are not included, some severe congenital anomalies will be underreported. For example, most fetuses with anencephaly are stillbirths. Therefore, the birth prevalence for anencephaly will not be accurate if only live births are counted.*
- 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.
  - *Response: The neonate would not be captured if the only data source is the delivery ward log. However, if there is a note in the protocol and inclusion criteria to also include all neonates with congenital anomalies from the obstetric operating theatre/room, then this neonate would be captured.*
- 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.
  - *Response: The neonate would not be included, because his/her weight is below 1000 g.*
- 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.
  - *Response: The fetus would not be included, because prenatal diagnosis was not confirmed at birth.*
- Explain how the inclusion and exclusion criteria can change the birth prevalence estimate.

- *Response:*
  - *Including live births, stillbirths and terminations will provide a more accurate prevalence of the congenital anomaly.*
  - *If the surveillance programme has resources and is able to have an age threshold of one year instead of the first three days of life, it will generate a more accurate prevalence of each congenital anomaly. If the surveillance programme can only capture births of 28 weeks of gestation or more, or 1000 g or more, many stillbirths with congenital anomalies will be missed.*

## Description formats for congenital anomalies

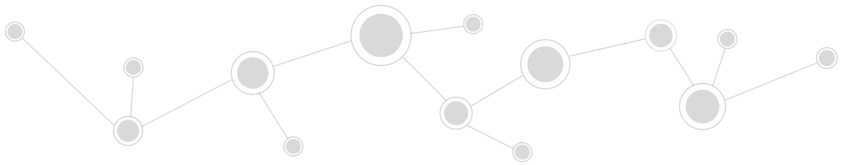
Expected time: 15 minutes

### Script/key points

- We will now discuss the methods for describing congenital anomalies in the data-collection process. Poor descriptions will negatively affect data quality.
- There are two methods for describing congenital anomalies in data-collection tools: verbatim descriptions and checkboxes.
  - Verbatim descriptions allow for more detail, which may be necessary for accurate diagnosis.
  - Checkboxes alone might limit data quality. It is more useful to include further options for categorizing congenital anomalies.
    - For example, if a country has the resources to collect data electronically, a checkbox could be useful as a first step, and then a window could open into a drop-down menu with more options to categorize the congenital anomaly.

### Begin Group discussion 3.7

- **Show the participants the examples of verbatim and checkbox formats.**
- **Ask participants the following question.** What do these examples demonstrate? **Allow participants time to write responses. Encourage group discussion about responses before reviewing.**
  - *Possible responses:*
    - *These examples demonstrate how using verbatim descriptions can add more thorough information about a diagnosis.*
    - *Using only the checkbox format requires a programme to rely solely on the training and expertise of the member of staff who checked the box, to complete the form.*
    - *A checkbox format might facilitate data management; however, detailed information that would help with a diagnosis may be missed.*



**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 |

## Core ascertainment variables

Expected time: 1.5 hours

### Script/key points

- Let's now discuss core ascertainment variables.
- The objectives and goals of a surveillance programme will drive what the programme will choose as its core ascertainment variables.
- Countries should assess the availability of core variables on existing data sources to determine what important information on congenital anomalies is already being collected for other analytical purposes.

### Begin Activity 3.3

- **Ask participants to refer to their workbooks, page 7.**
- **Review the table and discuss the suggested core ascertainment variables. Discuss how variables might differ, depending on the participant's country, and how they would be adapted. Direct participants to Appendices H and I in WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)* for additional variables to consider.**
- **Ask participants to write in the blank column in their workbook the reason a variable presented in the table below should be collected. Once they have completed the table, show participants the following table with possible responses for further discussion.**

| Category | Variable name | Why this variable should be collected (*responses in italics below)* |
|---|---|---|
| Report | Case record identification | *To retrieve a form, and ensure that it is not duplicated in the database* |
| | City, province, state, or territory | *To identify the prevalence of congenital anomalies by place of birth* |
| Father | Name(s) | *To retrieve information, link infant* |
| Mother | Name(s) | *To retrieve information, link infant* |
| | Mother's date of birth, or age if date of birth is not available | *To identify the distribution and prevalence of congenital anomalies by maternal age (maternal age is a risk factor for some congenital anomalies)* |
| | Total number of pregnancies | *To identify the distribution and prevalence of congenital anomalies by previous parity* |
| Infant | Date of birth | *To retrieve information, and to ascertain the distribution of congenital anomalies by month/year* |
| | Sex | *To identify the distribution of congenital anomalies by sex* |
| | Outcome at birth | *To identify the distribution and prevalence of congenital anomalies by pregnancy outcome. Prevalence can be calculated by live births with or without stillbirths and ETOPFA* |

- Each country can develop a fictitious record(s) based on the typical variables collected in the country, and participants can be asked to abstract the information into the data-collection tool (see Activity 3.4). For the activity, the facilitator can look at different charts from the hospitals, to create beforehand a "record" that can be used.

## Begin Activity 3.4

- **Ask participants to refer to their workbooks, page 8.**
- **Show the example of an abstraction form. Ask participants to consider the form, based on how they plan to collect data (paper-based or electronic collection) and which variables they 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: ○ physician ○ nurse ○ other (specify):
Contact information:

Version: January 2014

**Begin Group discussion 3.8**

- **Ask the following questions, to generate discussion:**
  - What demographic variables would be important to include in a surveillance programme for your country?
  - What are the estimated rates of consanguinity in your country?
  - Do you think it might be important to ask about consanguinity? Why or why not?
  - What race/ethnicities might be useful to capture?
  - Do you think it is important to collect information about ethnicity? Why or why not?
  - What data are already being collected from existing vital registries and hospital logbooks in your country, and are they available?

## Data-collection methods and tools

Expected time: 15 minutes

### Script/key points

- Collecting data requires appropriate data-collection tools, such as an abstraction form, for recording information.
- The two most commonly used collection methods are paper-based and electronic recording.
  - Paper-based methods are still widely used for vital registration, surveillance and research purposes.
    - Potential advantages:
      - May be cost-effective for low-resource settings
    - Potential disadvantages:
      - More prone to human error, since transcription is required
      - Requires more time to collect and transfer data
  - Electronic methods are improving as technology evolves. The ideal electronic collection tool should be able to collect data and provide data securely to a data management centre for storage and analysis.
    - Potential advantages:
      - Improves record accuracy
      - Allows quick data transfer to a data management centre
      - Identifies missing data in a timely manner
      - Ability to take and transfer digital photographs

- Potential disadvantages:
  - More vulnerable to theft
  - Can compromise privacy/security if data are not encrypted
  - Requires training on how to use an electronic system

- **Ask participants:** What country resources and training would be required for each?
- **Ask participants:** What do you think is the most appropriate method for your country's situation?

## Data collection and management

Expected time: 15 minutes

### Script/key points

- We are now going to spend some time discussing data collection and management.
- Valid and reliable data collection, storage, management and analysis are critical to congenital anomalies surveillance programmes.
- **Ask participants**: What are the benefits of a well-designed data system?
  - *Possible responses:*
    - *Promotes high-data quality and secure data storage*
    - *Provides systematic collection of data*
    - *Provides ability to link congenital anomalies data with other available information for surveillance, research and prevention purposes*
    - *Facilitates timely dissemination of prevalence estimates to those who need to know*
    - *Allows reliable comparisons with data from other congenital anomalies programmes*
    - *Allows evaluation of the information that is sent by participating sites to the central surveillance system*

## Data-management protocol

Expected time: 1 hour

### Script/key points

- We are now going to discuss the importance of having a protocol in place for data management.
- **Ask participants**: Why is establishing a data-management protocol important?
  - *Possible responses:*
    - *Improves data quality through standardization of data management*

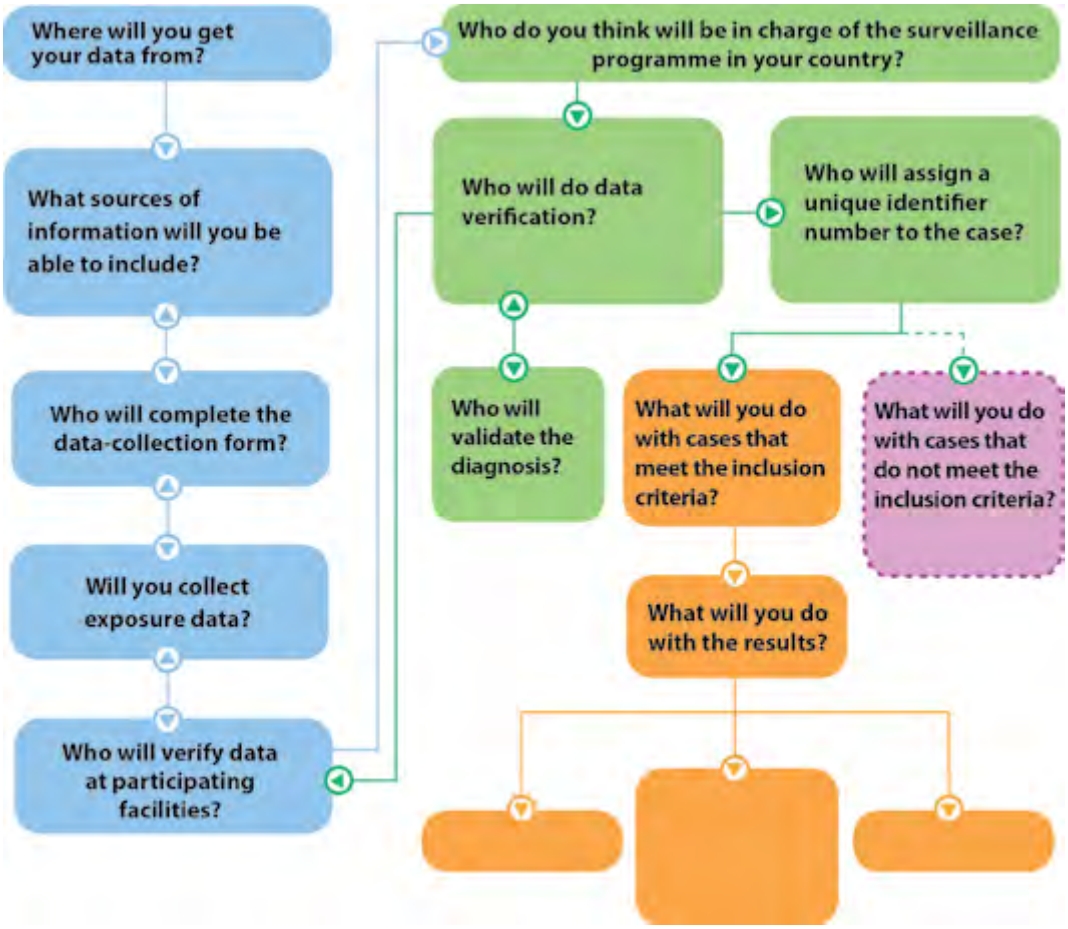
- *Creates procedural transparency*
- *Improves data transmission*
- *Ensures data confidentiality and security*
- *Defines the roles of data management in each step*

- It is important that all participating personnel are trained in the data-management protocol

- **Ask participants to refer to their workbooks, page 9. Review the questions in the flow chart and provide responses to them.**

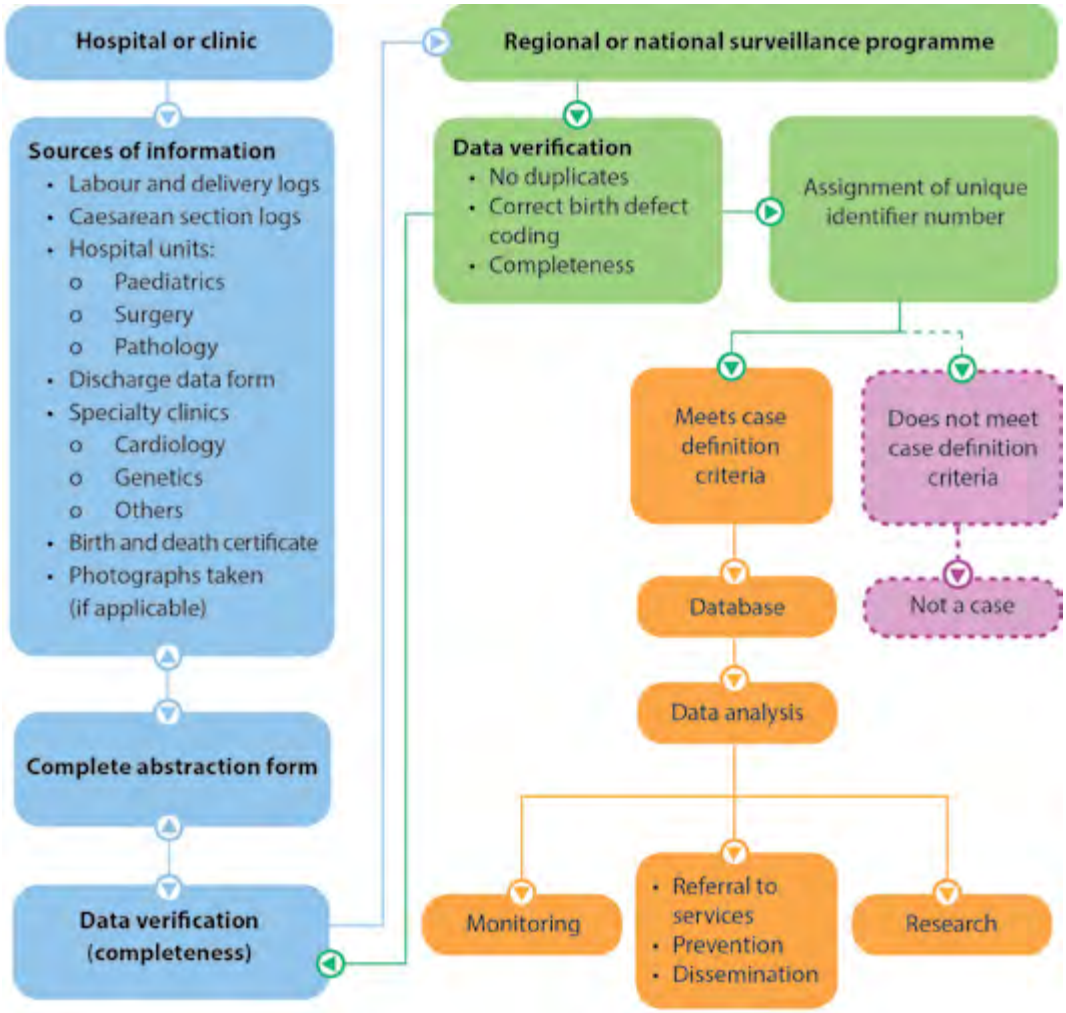
**Begin Activity 3.5**

- **Ask participants to complete the flow chart.**




- After participants complete their flow chart, show the following chart with possible responses and discuss participants' responses.

- *Possible response:*




- The protocol for data collection and management should include procedures for:
  - Identification of data sources where congenital anomalies can be identified
  - Identification and registration of congenital anomalies by health-care professionals in each data source
  - Training of personnel responsible for coding congenital anomalies according to the ICD-10 coding system
  - Taking photographs of a fetus or neonate with congenital anomalies, if appropriate for the setting
  - Verifying the information at the participating hospital site
  - Sending information to a regional or national-level surveillance programme

- It is important that the data collection and analysis are done in a systematic way by trained personnel, to ensure high-quality data and accurate interpretation.
  - Poor-quality data can lead to false conclusions about the occurrence of a congenital anomaly in a population, and weaken the credibility of the programme.
  - Inaccurate interpretation could have a substantial impact on the decision-making process of public health authorities.

- **Ask participants:** What are the most important attributes of high-quality data?
  - *Responses:*
    - *Completeness*
    - *Accuracy*
    - *Timeliness*
- Completeness refers to the extent to which data are comprehensive. For example, all cases at a given source in a specific timeframe have been identified, and all required data have been abstracted. Hospital audits and linkage of cases to other sources of data, such as vital records or specialized diagnostic centres, can help evaluate the completeness of case ascertainment.
- Accuracy refers to the extent to which data are exact, correct and valid. Approaches to help ensure data accuracy include:
  - Re-abstraction of information
  - Validity audits, such as identification of missed diagnoses or coding issues
  - Clinical reviews, such as verification of diagnoses, codes assigned, tests and procedures (i.e. two separate individuals input the same data and see if the database identified the duplicate)
  - Verification of data entry, such as customized programmes for range checks, automated fields, rejection of data that are known to be inaccurate, and routinely running data queries to identify duplicate entries and problems with variables
- Timeliness refers to the extent to which data are collected and analysed in a timely manner. It is measured by the time that elapses between the date of diagnosis and the date of abstraction; the date of abstraction and the date information is sent to the office; the date of arrival in the office to the date it is entered in the system; and the date of final product development and dissemination.
- Accuracy and timeliness also refer to how the data are reported to target audiences.

- **Ask participants:** What are some factors that could affect data quality in each of the steps of the protocol?
  - *Possible responses:*
    - *Missing values or empty data fields in the abstraction form*
    - *Duplication of cases*
    - *Errors in the diagnosis, description and/or coding of congenital anomalies*

## Data analysis

Expected time: 4 hours

### Script/key points

- Let's now talk about the typical measures used in surveillance of congenital anomalies; these measures include birth prevalence and trends.
- **Ask participants:** Does anyone know how to estimate birth prevalence? **Open for discussion, and then display the prevalence formula on screen. Discuss the difference between total prevalence, birth prevalence and live birth prevalence among participants (additional information can be found in Chapter 3 of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*).**
  - In a population-based surveillance programme, the total prevalence of congenital anomalies is calculated by aggregating the number of unduplicated cases (live births, stillbirths, and terminations) as the numerator, and the total number of live births and stillbirths in the source population as the denominator, for a specific catchment area and time period.
  - In hospital-based surveillance, the total prevalence of congenital anomalies is calculated by aggregating the number of unduplicated hospital cases (live births, stillbirths, and terminations) as the numerator, and the total number of live births and stillbirths from a participating hospital as the denominator, for a specific time period.
  - The prevalence of congenital anomalies is usually calculated and presented as prevalence per 10 000 births. Prevalence can be calculated for all congenital anomalies, specific individual congenital anomalies, or groups of anomalies.
  - Prevalence cannot be calculated with only the number of cases – the numerator data – without having information about the denominator.
  - The following expression is used to calculate the birth prevalence of congenital anomalies, with the assumption that both live births and fetal deaths are being captured:

    Birth prevalence = $a/b \times 10\,000$

    $a$ = Number of live births and fetal deaths (stillbirths) with a specific congenital anomaly (e.g. spina bifida) counted among the source population in a given year

    $b$ = Number of live births and fetal deaths (stillbirths), during the same year

1. Live birth prevalence of congenital anomalies $= \frac{\text{live birth cases}}{\text{total live births}} \times 10\,000$

2. Birth prevalence of congenital anomalies $= \frac{\text{live birth cases + fetal death (stillbirths) cases}}{\text{total live births + fetal deaths (stillbirths)}} \times 10\,000$

3. Total prevalence of congenital anomalies $= \frac{\text{live birth cases + fetal death (stillbirths) cases + ETOPFA cases}}{\text{total live births + total fetal deaths (stillbirths) + total ETOPFA}} \times 10\,000$

ETOPFA = elective termination of pregnancy for fetal anomaly.

- The numerator includes live births and known fetal deaths (stillbirths) with congenital anomalies, and pregnancy terminations with congenital anomalies (if these data are available). The denominator comprises only live births and fetal deaths (stillbirths; if these data are available), because it is practically impossible to assess the total number of pregnancy losses. Because the number of pregnancy losses is relatively small, compared with the number of live births, their exclusion has little effect on the prevalence estimate. Spontaneous abortions (also called miscarriages) are not included in the numerator or in the denominator because it is practically impossible to assess the total number of spontaneous abortions.

- Note that terminations are not included in the denominator. **Ask participants why they think terminations do not necessarily need to go in the denominator, and write responses on the flipchart**.
  - *Possible responses:*
    - *Hard to ascertain and usually not well reported in official demographic statistics*
    - *Likely that the total number is small and, compared to the total number of births, their exclusion has little impact on the overall estimate*
- For more information on how to calculate total prevalence, live birth prevalence and birth prevalence, please refer to Chapter 3 of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*.

## Begin Activity 3.6

- **Divide participants into groups. Ask participants to read the following case-study and to calculate the prevalence of neural tube defects. Tell them that they have the information in their workbooks, page 10.**

### Case-study 1: Cases of neural tube defects by type of ascertainment, United States of America (USA), 2004–2006

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 2010, data for 21 congenital anomalies from 2004–2006 were presented as national congenital anomalies prevalence estimates. The data presented in Table 3.1 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.

**Table 3.1. Cases of neural tube defects by type of ascertainment, USA, 2004–2006**

| | Number of cases | | | |
|---|---|---|---|---|
| Neural tube defects | Active ascertainment (11 programmes)[a] | Hybrid ascertainment (6 programmes)[b] | Passive ascertainment (7 programmes)[c] | National |
| Anencephaly | 697 | 211 | 192 | 1100 |
| Spina bifida | 1162 | 561 | 820 | 2543 |
| Encephalocele | 261 | 125 | 184 | 570 |
| Total neural tube defects | 2120 | 897 | 1196 | 4213 |

*Source*: Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE et al. Updated national birth prevalence estimates for selected congenital anomalies in the United States 2004–2006. Birth Defects Res A Clin Mol Teratol. 2010; 88:1008–16.


Data from programmes with active, hybrid or passive ascertainment.
[a] Number of live births in the active ascertainment programmes: 3 120 605.
[b] Number of live births in the hybrid ascertainment programmes: 2 075 973.
[c] Number of live births in the passive ascertainment programmes: 2 145 287.

- **Ask participants** to answer the questions in their workbooks, page 11:
  - 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.

- *Response:*

**Table 3.2. Prevalence of neural tube defects by type of ascertainment , USA, 2004–2006**

| Neural tube defects | Active ascertainment (11 programmes)[a] | | Hybrid ascertainment (6 programmes)[b] | | Passive ascertainment (7 programmes)[c] | | National | |
|---|---|---|---|---|---|---|---|---|
| | Cases | Prevalence | Cases | Prevalence | Cases | Prevalence | Cases | Prevalence |
| Anencephaly | 697 | 2.23 | 211 | 1.02 | 192 | 0.89 | 1100 | 1.50 |
| Spina bifida | 1162 | 3.72 | 561 | 2.70 | 820 | 3.82 | 2543 | 3.46 |
| Encephalocele | 261 | 0.84 | 125 | 0.60 | 184 | 0.86 | 570 | 0.78 |
| Total neural tube defects | 2120 | 6.79 | 897 | 4.32 | 1196 | 5.57 | 4213 | 5.74 |

*Source*: Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE et al. Updated national birth prevalence estimates for selected congenital anomalies in the United States 2004–2006. Birth Defects Res A Clin Mol Teratol. 2010; 88:1008–16.


Data are numbers of surveillance systems with active, hybrid or passive ascertainment.
[a] Number of live births in the active ascertainment programmes: 3 120 605.
[b] Number of live births in the hybrid ascertainment programmes: 2 075 973.
[c] Number of live births in the passive ascertainment programmes: 2 145 287.

- **Ask participants:** Describe the differences and similarities in prevalence by ascertainment method, and provide some reasons for why differences might exist.
  - *Possible responses:*
    - *The prevalence of specific defects varied by ascertainment method*
    - *The prevalence of anencephaly varied considerably by ascertainment method*
  - *Possible reasons for differences include:*
    - *Some programmes may include elective terminations*
    - *Some programmes may include stillbirths in the numerator, denominator or both*
    - *Some programmes may have conducted specialized prenatal ascertainment*
    - *The prevalence of encephalocele was higher among the passive ascertainment method than among the hybrid or active ascertainment methods*

- **Ask participants:** What are some possible reasons why the three ascertainment methods have different prevalence estimates for spina bifida?
  - *Possible responses:*
    - *Misclassification of cases at birth*
    - *Reporting problems/congenital anomalies not reported – could say biased reporting, i.e. underreporting, overreporting or selective reporting*
    - *Hybrid ascertainment methods are able to assess probable cases during follow-up and provide a definitive diagnosis*

- **Tell participants:** We are now going to discuss trends. Trends in the context of congenital anomalies surveillance are:
  - Used to provide information for needs assessments, programme planning, programme evaluation and policy development activities
  - Used to generate tables and graphs for prevalences over relatively long periods of time
    - Small numbers can introduce a large variation in yearly rates. When examining trends for small areas, small populations, or a narrow range of time, it may be necessary to combine several years of information.

## Begin Activity 3.7

- **Divide participants into groups of 3 or 4 participants. Ask participants to read the scenario below. Ask participants to go to page 12 of their workbooks.**

**Case-study 2: Pre- and post-fortification birth prevalence of neural tube defects in the USA, 1999–2007**

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 (1995–1997) and following the folic acid fortification mandate (1998–2010). The estimated annual prevalence of neural tube defects for nine hospitals in the USA during these time periods is presented in Table 3.3.

**Table 3.3. Prevalence of neural tube defects in the USA per 10 000 live births by race/ethnicity (1995–2007)**

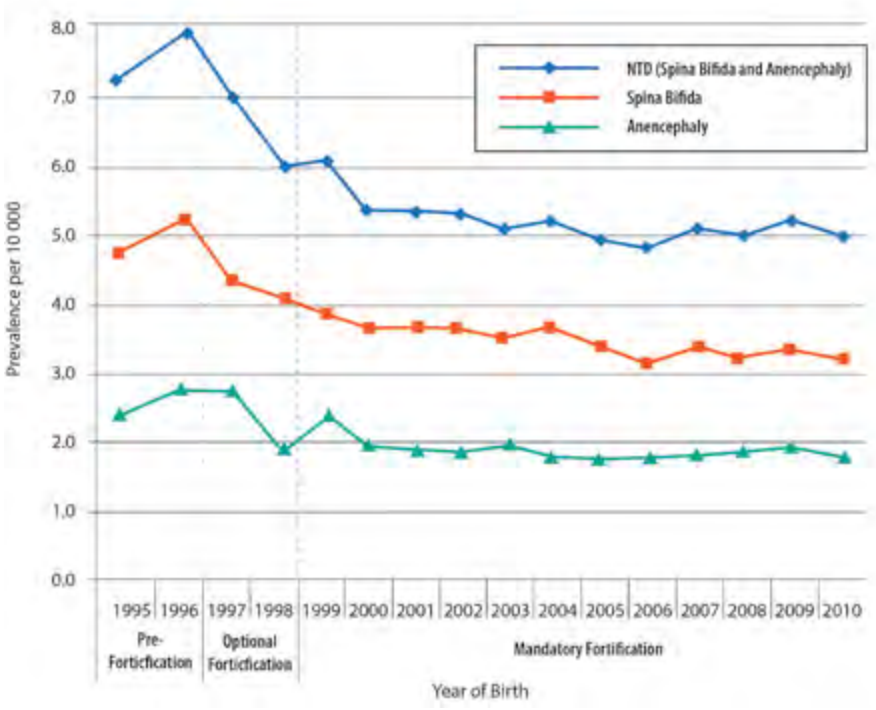
| Race/ ethnicity | Year | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 1995 | 1996 | 1997 | 1998 | 1999 | 2000 | 2001 | 2002 | 2003 | 2004 | 2005 | 2006 | 2007 |
| Hispanic | 9.20 | 10.84 | 9.69 | 7.37 | 7.83 | 6.45 | 6.63 | 6.98 | 6.95 | 6.63 | 6.27 | 5.69 | 6.04 |
| Black | 4.89 | 5.75 | 3.59 | 4.78 | 4.80 | 4.49 | 4.81 | 5.16 | 4.17 | 3.68 | 3.89 | 3.37 | 3.74 |
| Caucasian | 7.1 | 7.8 | 6.7 | 5.5 | 5.5 | 5.3 | 5.1 | 4.6 | 4.6 | 5.2 | 4.6 | 4.9 | 5.3 |

*Source*: CDC Grand Rounds: additional opportunities to prevent neural tube defects with folic acid fortification. MMWR Morb Mortal Wkly Rep. 2010;59(31):980–4.

- **Ask participants:**
  - 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?
  - *Possible response:*
    - *Folic acid fortification of staple foods is most likely responsible for the majority of the decline in prevalence of neural tube defects. The observable decline in the prevalence of neural tube defects is probably due to fortification.*

- *Note*: **If participants have a computer and access to Excel, ask them to make a graph with the data provided:**
  - **Remind participants** about the:
    - Need for a meaningful scale on the *Y* axis, to improve understanding and the use of a timescale on the *X* axis to show trends
    - Importance of axis labelling
    - Use of highlighting effects or factors of interest, such as year in which fortification started in the USA (voluntary and mandatory)
    - Need for clear and descriptive titles
- 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 such as national vital records, 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. Fig. 3.3 shows secular trend data for the prevalence of neural tube defects in the USA by race/ethnicity.

**Fig. 3.3. Prevalence of neural tube defects (per 10 000 births) by race/ethnicity, USA, 1995–2007**




*Source*: National Birth Defects Prevention Network. Neural Tube Defect Ascertainment Project 2010 (http://www.nbdpn.org/docs/NTD_Fact_Sheet_11-13_for_website.pdf).

- **Ask participants:** 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?
  - *Possible response: There is a decline in the prevalence of neural tube defects.*

- **Ask participants:**
  - When was this change first evident?
    - *Possible response: The change in prevalence started in the year 1997. After that point, the decrease in prevalence of neural tube defects accelerated through 2004, followed by a levelling off of the prevalence.*
  - What are some possible reasons for some of the changes observed in the prevalence of neural tube defects?
    - *Possible responses:*
      - *The introduction of folic acid fortification of staple foods*
      - *The prevalence of neural tube defects was already declining and it is only a continuation of such decline, possibly due to other unmeasured factors*
      - *Improved surveillance, more accurate data, fewer misclassifications*
      - *Changes in ascertainment*
  - What are some factors that could impact the prevalence of a health condition?
    - *Possible responses:*
      - *Population changes due to migration*
      - *Improved diagnostic procedures*
      - *Enhanced reporting techniques*
      - *Changes in the surveillance system or methods*
      - *Changes in prevalence of other risk factors for the condition*
      - *Changes in intervention*

## Data dissemination

Expected time: 1.5 hours

### Script/key points

- One of the most important steps in public health surveillance is the distribution or dissemination of relevant findings to appropriate audiences in a timely manner.
  - Possible audiences include partners, stakeholders, health-care providers, policy-makers and the public.
  - It is important to remember who your target audiences are when preparing data for dissemination.
    - What message or messages do you want to convey?
    - What do your target audiences need to know?
    - What data presentation format will be best understood?

- **Ask participants:** What do you think are some of the uses of public health surveillance data? **Allow participants to answer and discuss.**
  - *Possible responses:*
    - *Identifying congenital anomalies trends and geographic-specific rates*
    - *Planning, implementing and assisting with the evaluation of interventions or screening*
    - *Educating policy-makers, government officials and the public*
    - *Educating clinical/public health practitioners, nongovernmental organizations and the public*
    - *Identifying children with special needs and referring them to services*
- **Ask participants:** What would you include in a report designed for public health professionals and health-care providers? **Allow participants to answer.**
  - *Possible responses:*
    - *Analyses and interpretation of public health surveillance results*
    - *Recommendations for prevention activities stemming from the results*
    - *Suggestions for how the public health professionals and health-care providers can become involved*
- **Ask participants:** What would you include in a report designed for participating providers and institutions? **Allow participants to answer and discuss.**
  - *Possible responses:*
    - *Ways to improve reporting*
    - *Comparison of rates between geographic areas or populations, or participating institutions if the programme is hospital based*
    - *Detailed information related to programme progress in participating hospitals and health-care systems*
    - *Overall programme progress*
- Other important points to keep in mind when creating an output for surveillance data are the timeline for dissemination, the usefulness of disseminated information, and the channel for data dissemination. It is important to remember the audiences to which these reports are addressed, and reports should be tailored accordingly. Examples of channels are written reports, the Internet, media, or a combination of these.

## Begin Activity 3.8

- **Ask participants** to answer the questions in their workbooks, page 14:
- **Divide participants into small groups. Assign a target audience (groups 1–4 below) to each group. Using the sample surveillance data, have groups discuss how they would communicate and disseminate the surveillance data to their assigned group. After groups have had enough time to complete the activity, discuss results aloud as a larger group.**

### Target audience

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

## Begin Activity 3.9

- **Ask participants** to answer the questions in their workbooks, page 15:
- **Divide participants into small groups. Assign a target audience (groups 1–4 below) to each group. Using the sample surveillance data and the activity description, have groups draft an advocacy letter to their assigned target audience, requesting support for a local congenital anomalies surveillance programme.**

### Activity description

You are a group of paediatricians working in a large maternity facility in your country. You are seeing many babies (see Table 3.4) 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, what data will be collected and how it will be presented to make the case to your 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)

### Table 3.4. Birth prevalence of congenital anomalies by race/ethnicity

| | Prevalence of anomalies 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) |

## Evaluation questions 3

Expected time: 30 minutes

Correct answers are presented in **bold.**

1. Define population-based and hospital-based surveillance programmes.

**Answer:** Population-based programmes collect data from the entire resident population in a defined geographic area. Hospital-based programmes collect data from births that occur in preselected hospitals in a province, country or region.

2. Why are prevalence estimates drawn from hospital-based surveillance programmes less likely to be accurate?

**Answer:** Prevalence estimates drawn from hospital-based programmes only provide the estimate for the participating hospitals and cannot be generalized to a broader population. There is an exception, however; in countries where nearly all births take place in the hospital and all hospitals in a state, country or region participate, and where there is no selective inflow of cases from outside the state/country/region, those hospital-based programmes can accurately represent true prevalence.

3. What are some factors to consider when deciding whether to implement a population-based or hospital-based surveillance programme in a country?

**Answer:** Available resources AND/OR capacity

4. Which of the following is NOT a type of case ascertainment method?

    a. Active

    b. **Independent**

    c. Passive

    d. Hybrid

5. Which of the following is NOT a characteristic of passive case ascertainment?

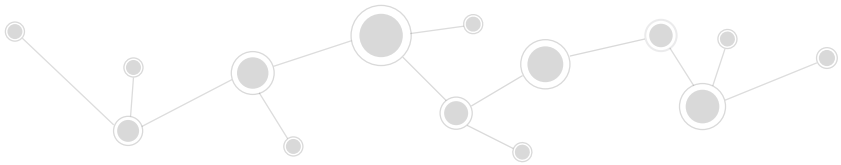
    a. The burden of work is placed on the data source staff.

    b. The data are not verified by surveillance staff, resulting in lower data quality and the potential for less accuracy.

    c. **This method can be resource and personnel intensive but usually results in improved data quality.**

6. True or **false**: Using a single data source may improve data quality.

7. **True** or false: New, smaller programmes should consider starting with a small number of easily recognizable, major external congenital anomalies and then expand to include additional anomalies as the programme gains experience, creates awareness and is assigned more resources.



8. List some differences between the two methods for describing congenital anomalies in the data-collection process.

**Answer:** Verbatim descriptions allow for more detail, which may be necessary for accurate diagnosis and during the case-management process. Checkbox descriptions are predetermined categories of selected congenital anomalies. Checkbox descriptions alone may be insufficient for high data quality. It is more useful to include further options for categorizing anomalies.

9. **True** or false: Inclusion of live births alone is a significant limitation that can lead to unreliable rates and trends, particularly for conditions with a high rate of loss prior to 28 weeks.

10. Define stillbirth according to WHO.

**Answer:** Fetal deaths at ≥28 weeks of gestation or weighing ≥1000 g if gestational age is unknown.

11. Which is an advantage of paper-based collection methods?

    a. **They may be cost-effective for low-resource settings.**

    b. They may be more prone to human error, since transcribing is required.

    c. They may require more time to collect and transmit data.

12. True or **false**: Incidence is usually used to describe the occurrence of congenital anomalies.

13. Define prevalence.

**Answer:** Prevalence is a measure of the total number of existing cases of a disease for a given point in time or period, and in a given population, regardless of whether or not they are new cases.

14. **True** or false: The three top attributes of data quality are timeliness, accuracy and completeness.

# INTRODUCTION TO SURVEILLANCE OF SELECTED CONGENITAL ANOMALIES

MODULE 4

Expected time: 1.5 hours

## Objectives

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

- describe basic congenital anomaly characteristics;
- describe features and subtypes of selected congenital anomalies;
- recognize and differentiate the various types of spina bifida;
- recognize omphalocele and differentiate it from gastroschisis;
- recognize and differentiate the various types of limb reduction defects (limb deficiencies).

*Note*: This module is linked to Chapter 4 of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*.

## External major congenital anomalies for monitoring

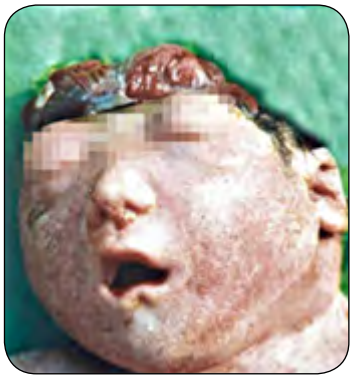
Expected time: 1 hour

### Script/key points

- We are now going to turn our discussion to a selection of external major congenital anomalies.
- 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.
- Show *A video guide to a stepwise surface examination of newborns* at http://www.who.int/tdr/publications/videos/completed-productions/en/
- Please refer to Chapter 4 in WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)* for details on selected major congenital anomalies.

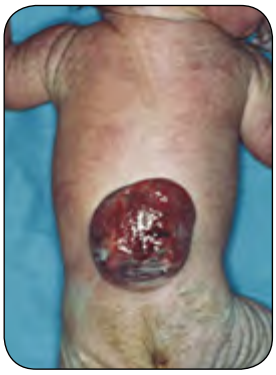
## Begin Activity 4.1

- **Ask participants** to answer the questions in their workbooks, page 16:
- **Have participants view and compare photos of anencephaly, encephalocele and spina bifida, and describe the differences. After enough time has been given to complete the activity, discuss responses aloud as a larger group.**



**Anencephaly**

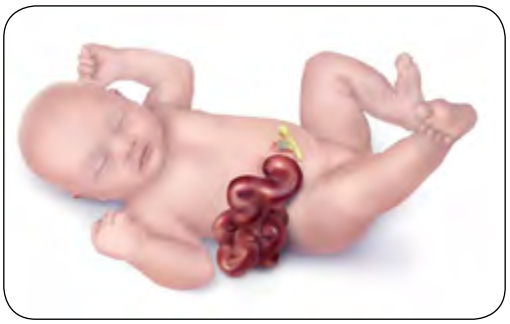
**Encephalocele**



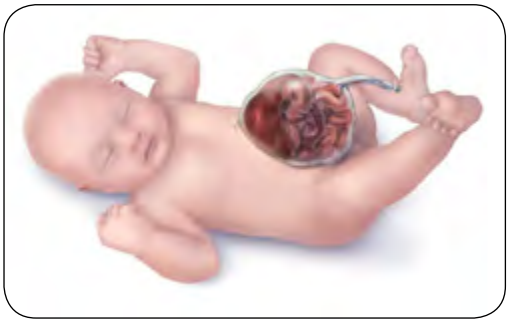
**Spina bifida**

- *Responses:*
  - *Anencephaly is characterized by either total or partial absence of the brain, together with total or partial absence of the cranial vault and the covering skin.*
  - *An encephalocele is a pedunculated or sessile cystic lesion protruding through a defect in the skull.*
  - *Spina bifida is a general term used to describe a neural tube defect of the spine, in which part of the meninges or spinal cord, or both, protrudes through an opening in the vertebral column.*

- **Have participants identify the diagnoses for each of the following congenital anomalies. After enough time has been given to complete the activity, discuss the differences between gastroschisis and omphalocele aloud as a larger group.**



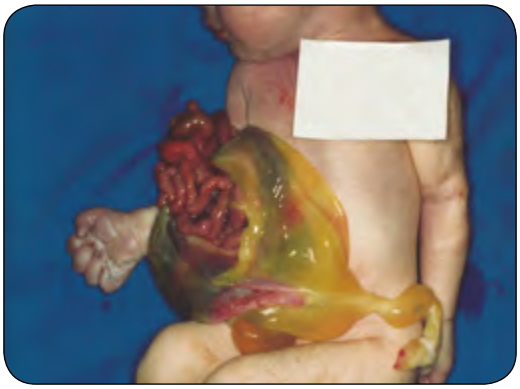
**Gastroschisis**



**Omphalocele**

- *Responses:*
  - *Gastroschisis Q79.3; omphalocele: Q79.2*
  - *In gastroschisis, the abdominal opening is positioned lateral to the umbilical cord insertion, usually to the right.*
  - *In omphalocele, the abdominal contents are herniated through an enlarged umbilical ring and the umbilical cord is inserted in the distal part of the membrane covering the defect. It is important to note that omphalocele and gastroschisis may not be easily differentiated when the membrane covering the omphalocele has ruptured.*

- **Have participants view the following photograph (on page 17 of their workbooks). Ask participants:** Do you think the baby has gastroschisis or omphalocele? **Have them explain their responses aloud as a group.**



- *Response:*
  - *Omphalocele can be seen in the photograph, but the membrane is ruptured.*

- **Have participants identify the diagnoses for each of the following congenital anomalies (on page 17 of their workbooks). Tell participants:** Hydrocephalus cannot be seen in all the photos; therefore, use a code for spina bifida either with or without hydrocephalus. **After enough time has been given to complete the activity, discuss responses aloud as a larger group.**

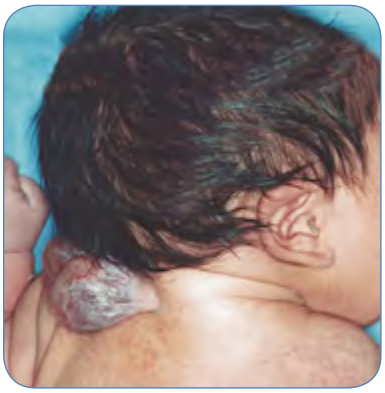


**Photo A**

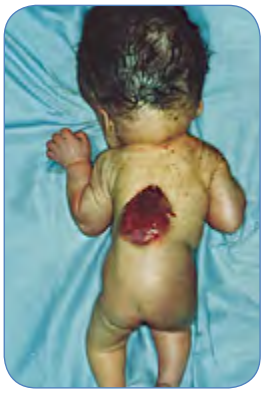


**Photo B**

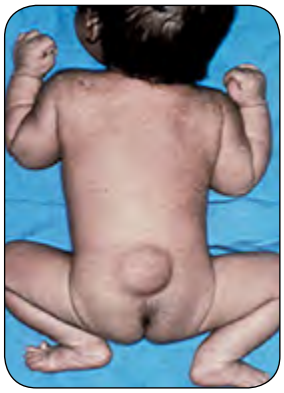


**Photo C**

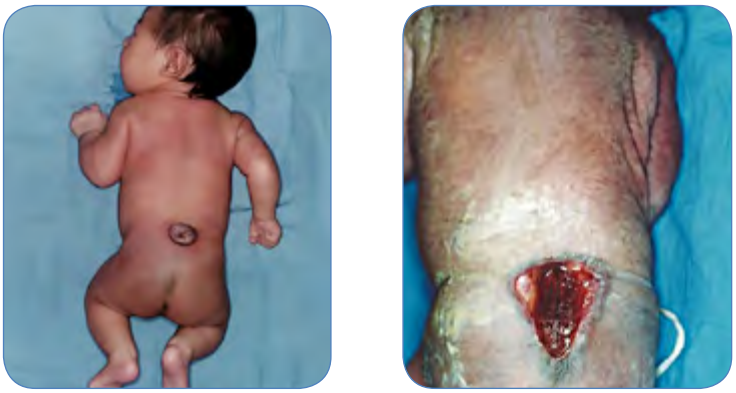


**Photo D**



**Photo E**

- *Responses:*
  - *Photo A: Cervical spina bifida with or without hydrocephalus – Q05.0 or Q05.5*
  - *Photo B: Thoracic spina bifida with or without hydrocephalus – Q05.1 or Q05.6*
  - *Photo C: Lumbar spina bifida with or without hydrocephalus – Q05.2 or Q05.7*
  - *Photo D: Lumbar spina bifida with or without hydrocephalus – Q05.2 or Q05.7*
  - *Photo E: Lumbar spina bifida with or without hydrocephalus – Q05.2 or Q05.7*

**Coding**

| | |
|---|---|
| Q05.0 | Cervical spina bifida with hydrocephalus |
| Q05.1 | Thoracic spina bifida with hydrocephalus |
| Q05.2 | Lumbar spina bifida with hydrocephalus; 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; Lumbosacral spina bifida without hydrocephalus, not otherwise specified |
| Q05.8 | Sacral spina bifida without hydrocephalus |
| Q05.9 | Spina bifida, unspecified |

## Evaluation questions 4

Expected time: 30 minutes

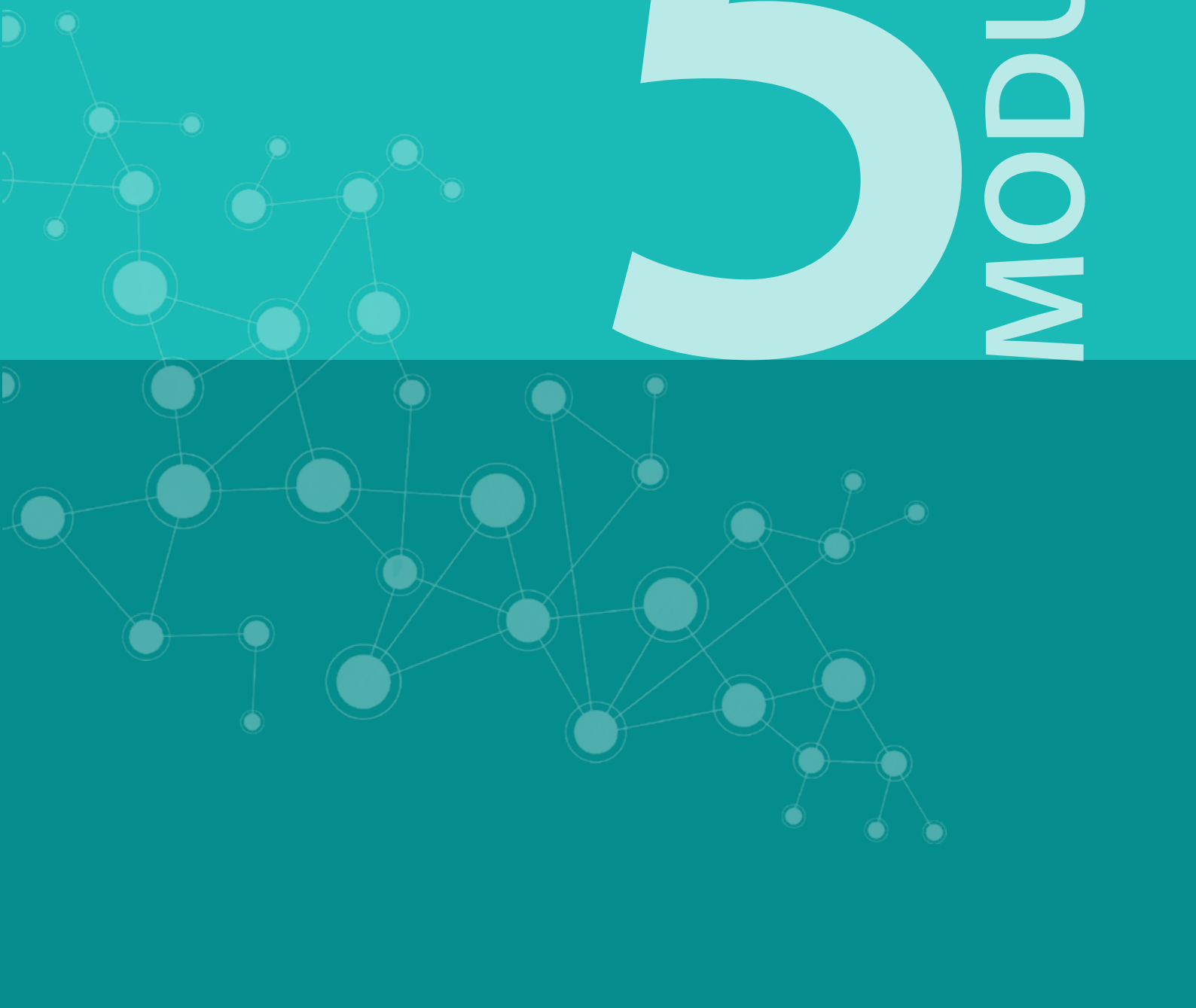
Correct answers are presented in **bold**.

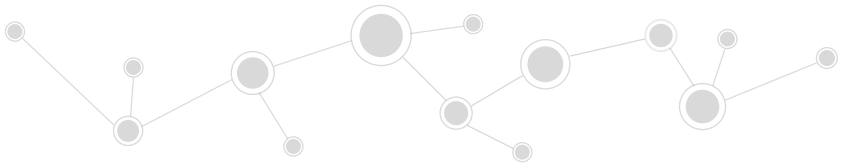
1. Which of the following is preferred when collecting congenital anomalies surveillance data?
    a. **High-quality data on a smaller number of diagnoses**
    b. Low-quality data on all of the diagnoses
2. Which of the following is NOT considered a neural tube defect?
    a. Anencephaly
    b. Craniorachischisis
    c. **Gastroschisis**
    d. Encephalocele
    e. Spina bifida
3. Which of the following describe a type of anencephaly?
    a. **Holoanencephaly**
    b. Iniencephaly
    c. Encephalocele
    d. a and c
    e. All of the above
4. Which of the following is a sessile cystic lesion protruding through a defect in the skull that may contain herniated meninges and brain tissue and is commonly located in the occipital region, except in South-East Asia, where it is commonly anterior?
    a. Iniencephaly
    b. **Encephalocele**
    c. Spina bifida
5. How do the clinical presentations of omphalocele and gastroschisis differ?

**Response:** In gastroschisis, the abdominal opening is positioned lateral to the umbilical cord insertion, while in omphalocele, the abdominal contents are herniated through an enlarged umbilical ring and the umbilical cord is inserted in the distal part of the membrane covering the anomaly. However, omphalocele membranes can sometimes rupture prior to or during delivery, resulting in an anomaly that may be difficult to distinguish from gastroschisis.

# INTRODUCTION TO CODING

MODULE 5





Expected time: 5 hours

## 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.

*Note*: This module is linked to Chapter 5 of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*.

- **Display the coding process flowchart while presenting this session**

## The International Classification of Diseases

Expected time: 30 minutes

### Script/key points

- We are now going to discuss how congenital anomalies are coded utilizing the *International statistical classification of diseases and related health problems*, tenth edition, also known as the ICD-10, and the Royal College of Paediatrics and Child Health (RCPCH) adaptation.
- The ICD-10 is developed and maintained by WHO, and is considered the international standard diagnostic classification system.
- The most recent version of the ICD-10 is available on the WHO website *(3)* (http://apps.who.int/classifications/icd10).
- It is widely used in many countries as a classification system for diseases.
- It is useful in assisting with analysis and assessment of the health situation of population groups and for monitoring the incidence and prevalence of diseases and other health conditions.
- ICD-10 codes are listed in alpha-numeric order and are described in detail. Classification of structural congenital anomalies is found in Chapter XVII: Congenital malformations, deformations and chromosomal abnormalities (Q00–Q99). Classification of haemolytic anaemias (thalassemia and sickle cell disorders) is found in Chapter III: Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism (D50–D89).

- The ICD-10 codes lack specificity for uniquely coding some congenital anomalies and most genetic syndromes. Therefore, some congenital anomalies surveillance programmes use their own local modification of the ICD-10 that includes additional codes for some specific congenital anomalies not found in the ICD-10, and add an extra digit to allow for more detailed coding of some anomalies and specificity of diagnoses.
  - As a result of this lack of specificity of ICD-10 codes, the RCPCH developed an adaptation of the ICD-10. Please refer to Chapter 5 of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4)*, for an example of this adaptation of the ICD-10.
  - This adaptation is most commonly used by programmes carrying out public health surveillance of congenital anomalies.
  - Also, when the ICD-10 code is not specific enough, for example, "Q01.8 Encephalocele of other sites" (**write on the flipchart**), then using the classification developed by the RCPCH could be beneficial. **Write the following examples on the flipchart:** "Q01.80 Parietal encephalocele", "Q01.81Orbital encephalocele", "Q01.82 Nasal encephalocele", "Q01.83 Nasopharyngeal encephalocele".

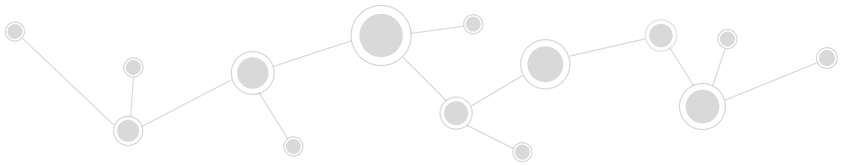
- If possible, the use of codes for nonspecific diagnoses should be avoided. For example, try to avoid a code such as "Q01.9 Encephalocele, unspecified"; however, sometimes you do not have a choice.

## Certainty of diagnosis

Expected time: 30 minutes

### Script/key points

- We are now going to discuss certainty of diagnosis.
- The certainty of diagnosis could vary for live births versus stillbirths and for prenatal versus postnatal assessments.
- With pregnancy terminations, the prenatal diagnosis may not be verified for many reasons, including the method of termination, the condition of the specimen, or lack of post-termination examination or autopsy.
- Programmes interested in more detailed information on inclusion of prenatal diagnosis in congenital anomalies surveillance can find useful and practical suggestions and tips in the guidelines developed by the National Birth Defects Prevention Network in the USA. Guidelines are included under the resources section of your workbook.
- Among neonates who die shortly after birth, the diagnosis could also cause difficulties if certain examinations such as X-rays or an autopsy are not done.
- It is beneficial to code possible diagnoses differently from confirmed diagnoses (for analytic purposes).



- This can be done by using a separate field on the congenital anomalies abstraction form to include this information.
- An extra digit can also be added to the ICD-10-RCPCH codes.

  - **Ask participants:** Give one example of a congenital anomaly with a possible diagnosis.
- *Possible responses:*
  - *Hydrocephalus suggested by prenatal ultrasound and no postnatal confirmation is done*
  - *Cleft lip prenatally diagnosed without postnatal confirmation*
  - *Clinical diagnosis of holoprosencephaly without imagining studies or postmortem examination*

## Personnel responsible for diagnosis and coding

Expected time: 15 minutes

### Script/key points

- Although coding of congenital anomalies can happen in the hospital or clinic, the final code will always be at the central registry where final review and verification of all codes reported by participating sites occurs. However, it is important to also train the hospital staff responsible for diagnosis and coding of congenital anomalies.
- If coding is done at the hospital, someone who is knowledgeable about congenital anomalies should also review and confirm the diagnoses and assign the codes.
  - They will be reviewed and verified at the central registry level.
    - Not every site will have 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 of multiple congenital anomalies

Expected time: 15 minutes

Script/key points

- Approximately 75% of babies with major congenital anomalies present as isolated anomalies, and the remaining 25% have more than one major anomaly.
- Neonates with one or more major anomalies may also have one or more minor anomalies.
  - For more details about the types of congenital anomalies according to clinical presentation, please refer to Appendix C of WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4).*

    - A 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 very important for an accurate diagnosis and, therefore, an accurate coding of the congenital anomaly.

## Considerations for coding congenital anomalies

Expected time: 3 hours

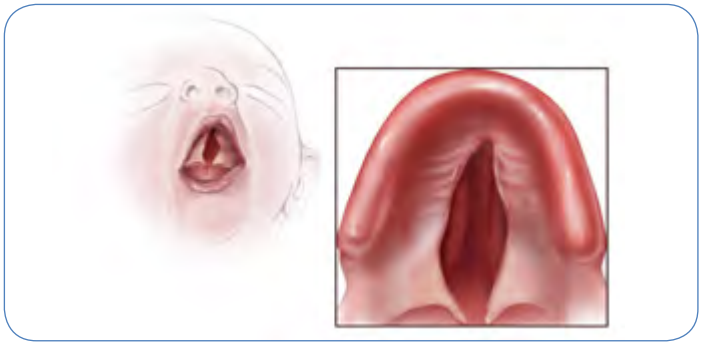
### Script/key points

- It is critical to ensure that coding of the recorded diagnostic information is done correctly and accurately. Correct and accurate coding is:
  - Central to the process of generating valid and reliable information within a congenital anomalies surveillance system
  - Achieved by following a standardized coding system such as the ICD-10 or the more detailed ICD-10-RCPCH
- Similarly, it is important to obtain the best possible clinical descriptions, so that a 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 can supplement the 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.

## Begin Activity 5.1

- **Ask participants to refer to their workbooks, page 18.** Have them identify the diagnoses for each of the following congenital anomalies (on page 18 of their workbooks), and code each case. After enough time has been given to complete the activity, discuss responses aloud as a larger group.

**Baby 1**



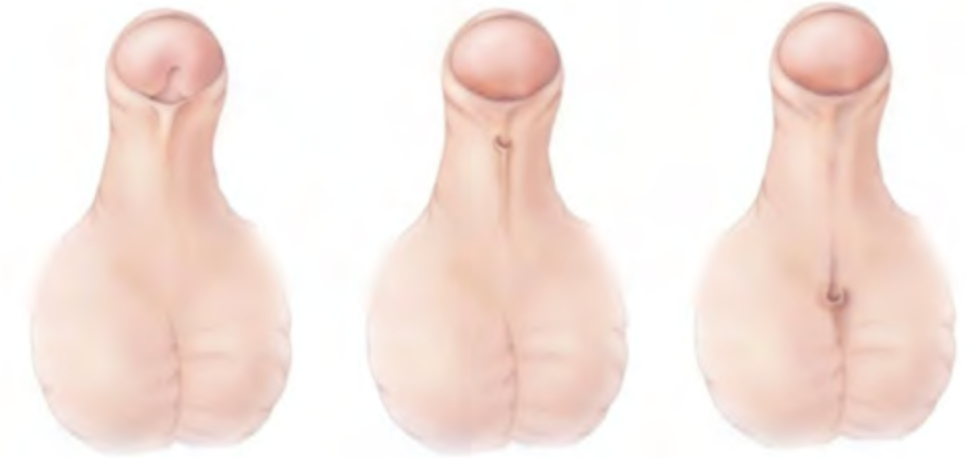
**Baby 2**

*Coding: ICD-10 or RCPCH*

| | |
|---|---|
| **Cleft palate** | Q35.1, Q35.3, Q35.5, Q35.59, Q35.9, Q87.0 |
| **Cleft lip** | Q36.0, Q36.9, Q36.90, Q36.99 |
| **Cleft palate with cleft lip** | Q37.0, Q37.10, Q37.19, Q37.2, Q37.3, Q37.4, Q37.5, Q37.59, Q37.8, Q37.9, Q37.99 |

- *Responses:*
  - *Baby 1: Unilateral cleft lip – Q36.90*
  - *Baby 2: Cleft palate – Q35.5*

Types of hypospadias



**Subcoronal** **Midshaft** **Penoscrotal**

***Coding: ICD-10 or RCPCH***

| | |
|---|---|
| **Q54.0** | Hypospadias, balanic<br>Coronal<br>Glanular |
| **Q54.1** | Hypospadias, penile |
| **Q54.2** | Hypospadias, penoscrotal |
| **Q54.3** | Hypospadias, perineal |
| **Q54.8** | Other hypospadias |
| **Q54.9** | Hypospadias, unspecified |

- *Responses:*
  - *Subcoronal: Hypospadias, penile – Q54.1*
  - *Midshaft: Hypospadias, penile – Q54.1*
  - *Penoscrotal: Hypospdias, penoscrotal – Q54.2*

- **Ask participants:** What is the correct diagnosis and ICD-10-RCPCH code when both anencephaly and an open spine are present and contiguous?
  - *Response: The correct diagnosis is craniorachischisis. The ICD-10-RCPCH code is Q00.1. Craniorachischisis refers to the presence of anencephaly with a contiguous spine defect without meninges covering the neural tissue (rachischisis). It may be limited to the cervical region or affect the entire spine.*

- **Ask participants:** How do you code if both anencephaly and spina bifida are present, but are not contiguous?
  - *Response: In cases in which anencephaly and spina bifida are present, but are not contiguous, both should be coded; however, when the malformations are counted, only anencephaly should be counted.*

- **Ask participants:** Do you use ICD-10 codes in your country? Who uses them, and for what purpose?

## Begin Activity 5.2

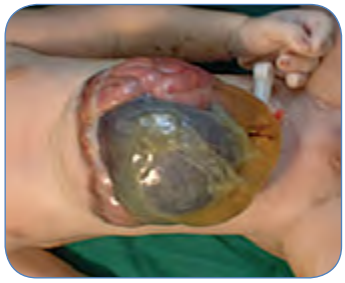
- Instructions:

**Ask participants to refer to their workbooks, page 19.** A total of 20 cases are included in the activity.

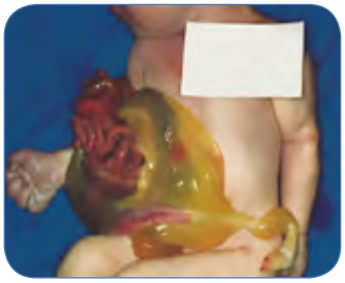
- Break out into smaller groups (no more than 5 people per group).
- Each group will receive a set of pictures labelled with numbers.
- Write the number of the photo and describe it on the answer sheet.
  **DO NOT write down the ICD-10 or ICD-10-RCPCH code**.
- Exchange answer sheets (but not photos) 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.

*Instructor note*: All picture sets should have different pictures.

The descriptions for each case are provided at the end of this *Facilitator's guide,* in "Description and coding answers".

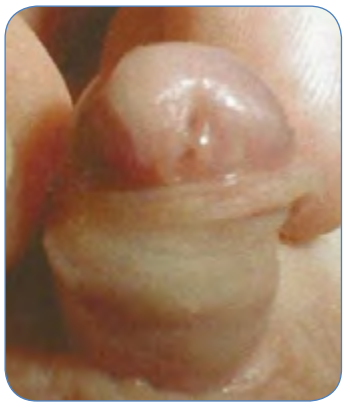


1

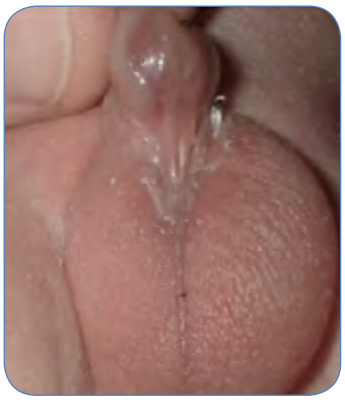


2

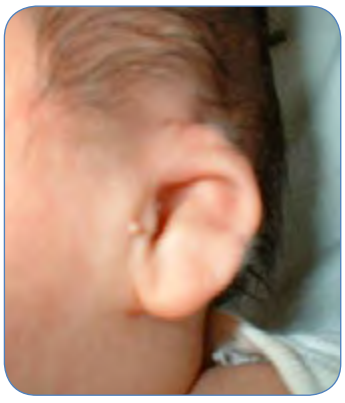
3



4



5



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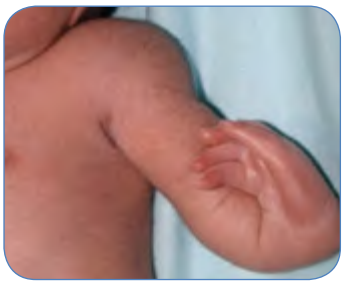
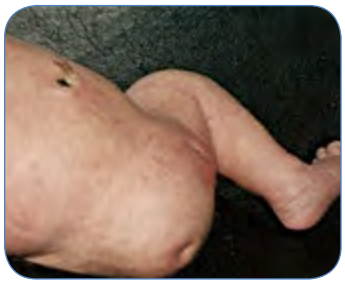
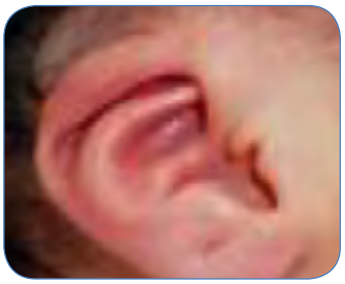


9



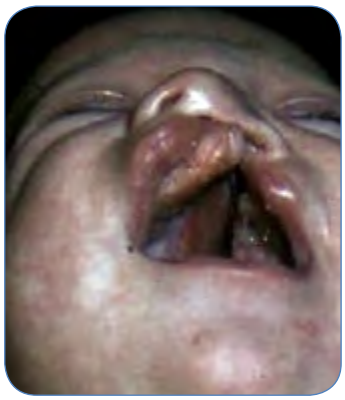
10

11-A

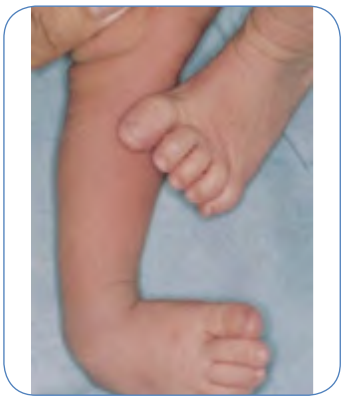


11-B

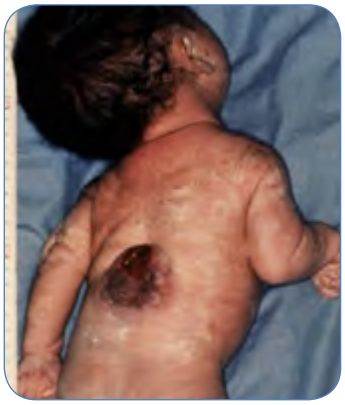
11-C



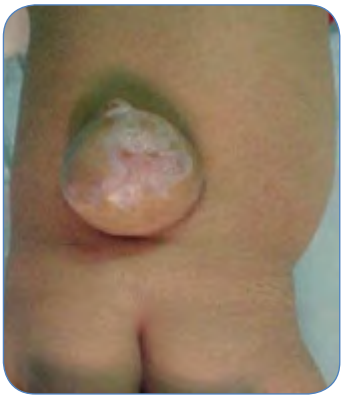
12



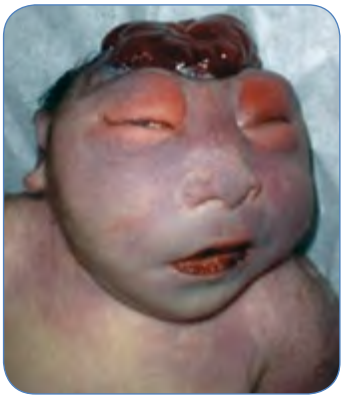
13



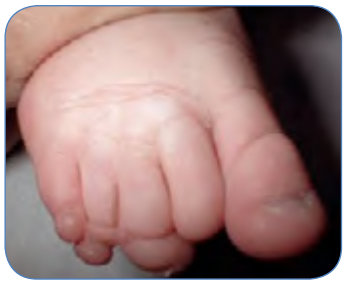
14



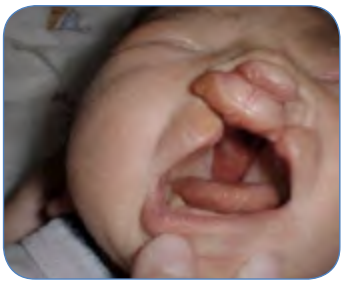
15



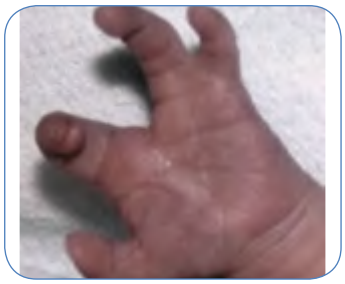
16



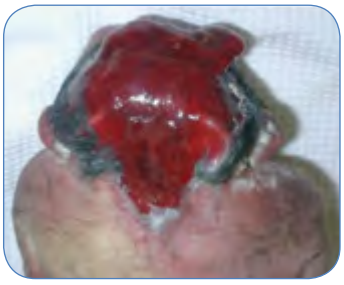
17



18



19



20

## Begin Activity 5.3

- **Ask participants** to answer the questions in their workbooks, page 20:
- **Have participants assign an ICD-10 or ICD-10-RCPCH code or codes, based on the available clinical description of the different fetuses or infants with congenital anomalies. After enough time has been given to complete the activity, discuss aloud as a larger group.**

*Note*: ICD-10 or ICD-10-RCPCH codes are listed

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 1**<br><br>Spina bifida with LS meningocele; massive hydrocephalus<br><br>■ *Response*<br>› *In this case, "LS" is used to abbreviate "lumbosacral". Although the description might suggest two anomalies (spina bifida and hydrocephalus), hydrocephalus is common among children with spina bifida and it is considered a consequence of spina bifida, the primary major congenital anomaly in this case. There are specific codes for "spina bifida with hydrocephalus" in the ICD-10.*<br><br>› *The ICD-10 code for lumbosacral spina bifida with hydrocephalus is Q05.2.*<br><br>*Note*: This case would not be included in analyses of hydrocephalus as a primary anomaly. | 1. Q05.2 |
| **CASE 2**<br><br>Frontal encephalocele; clubbing of left foot<br><br>■ *Response*<br>› *The ICD-10 code for "frontal encephalocele" is Q01.0.*<br><br>› *The ICD-10 code for "clubbing of left foot" is Q66.8.* | 1. Q01.0<br><br>2. Q66.8 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 3**<br><br>Cleft lip and palate; omphalocele<br><br>■ *Response*<br>› *It is not specified whether the soft palate, hard palate or both are affected, and no information is provided regarding the laterality (sidedness) of the cleft lip. The ICD-10-RCPCH adaptation code for "cleft lip and palate" is Q37.99 (Cleft palate with cleft lip, unspecified).*<br>› *The ICD-10 code for "omphalocele" is Q79.2.*<br><br>*Note*: For cleft palate, it is uncommon to have the detailed description available (whether the soft or hard palate is affected), unless the description is provided as a result of a surgical repair. | 1.Q37.99<br><br>2. Q79.2 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 4**<br><br>Facial cleft; amniotic band evidence on face<br><br>■ *Response*<br>› *Since "amniotic band" is present on the face, this is the most likely cause of the facial cleft. Facial cleft is different from cleft lip.*<br>› *The ICD-10 code for "facial cleft" is Q18.8 (Other specified congenital malformations of face and neck).*<br>› *If the surveillance programme is focusing on recording cases with cleft lip and cleft palate, then it is not necessary to code this anomaly, and the case can be excluded from surveillance. However, if one would like to include all types of congenital anomalies and assign a code, then the code for cases with known or probable amniotic band/constriction band presence is Q79.80 (Congenital constriction bands), which is the ICD-10-RCPCH adaptation of ICD-10 code Q79.8 (Other congenital malformations of musculoskeletal system).*<br><br>*Note*: ICBDSR recommends using Q79.80 to identify the presence of an amniotic band. Cases with amniotic bands should be coded using the codes for the specific congenital anomalies as well as the Q79.80 amniotic band code. This anomaly will be excluded from analysis of cleft lip and palate.<br><br>It is on the **exclusion** list as noted in WHO/CDC/ICBDSR *Birth defects surveillance: a manual for programme managers (4).* | 1. Q18.8<br><br>2. Q79.80 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 5**<br><br>Small encephalocele in the parietal area; cleft palate NOS; fused toes NOS<br><br>■ *Response*<br>› *The abbreviation "NOS" stands for "not otherwise specified".*<br>› *The ICD-10 code for "encephalocele in the parietal area" is Q01.8 (Encephalocele of other sites) or the more specific ICD-10-RCPCH adaptation code Q01.80 (Parietal encephalocele).*<br>› *The ICD-10 code for "cleft palate NOS" is Q35.9 (Cleft palate, unspecified) or the ICD-10-RCPCH adaptation code Q35.99 (Cleft palate, unspecified).*<br>› *The ICD-10 code for "fused toes" is Q70.2.*<br><br>*Note*: Although "NOS" is a valid term in the ICD-10, it should be used only when there is no possibility of obtaining a better description for a specific congenital anomaly. For cleft palate, it is uncommon to have the detailed description available (whether the soft or hard palate is affected), unless the description is provided as a result of a surgical repair. | 1. Q01.8/Q01.80<br><br>2. Q35.9/Q35.99<br><br>3. Q70.2 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 6**<br><br>Anencephaly; heart defect NOS; spinal anomalies NOS; lower extremity abnormal development<br><br>■ *Response*<br>› *The abbreviation "NOS" stands for "not otherwise specified".*<br>› *The ICD-10 code for "anencephaly" is Q00.0.*<br>› *The ICD-10 code for "heart defect NOS" is Q24.9.*<br>› *The ICD-10 code for "spinal anomalies NOS" is Other congenital malformations of spine – Q76.4.*<br>› *The ICD-10 code for "lower extremity abnormal development" is Q74.9 (Unspecified congenital malformation of limbs).*<br><br>*Note*: Although "NOS" is a valid term in the ICD-10, it should be used only when there is no possibility of obtaining a better description for a specific congenital anomaly. | 1. Q00.0<br><br>2. Q24.9<br><br>3. Q76.4<br><br>4. Q74.9 |
| **CASE 7**<br><br>Hypospadias, penoscrotal; unilateral absent middle phalanx on foot (no further description)<br><br>■ *Response*<br>› *The ICD-10 code for "hypospadias, penoscrotal" is Q54.2.*<br>› *The ICD-10 code for "unilateral absent middle phalanx on foot" is Q72.3 (Congenital absence of foot and toe(s)) or the ICD-10-RCPCH adaptation code Q72.30 (Congenital absence of toe(s)).* | 1. Q54.2<br><br>2. Q72.3/ Q72.30 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 8**<br><br>Transposition of the great arteries with intact ventricular septum (D-TGA); bilateral cleft lip and palate<br><br>■ *Response*<br>› *The ICD-10 code for "transposition of the great arteries with intact ventricular septum (D-TGA)" is Q20.3 (Discordant ventriculoarterial connection). Transposition of great vessels complete is listed under this code.*<br>› *The ICD-10 code for "bilateral cleft lip and palate" is Q37.8 (Unspecified cleft palate with cleft lip, bilateral).*<br><br>*Note*: For cleft palate, it is uncommon to have the detailed description available (whether the soft or hard palate is affected), unless the description is provided as a result of a surgical repair. | 1. Q20.3<br><br>2. Q37.8 |
| **CASE 9**<br><br>Occipital encephalocele; subcoronal hypospadias; bilateral club feet<br><br>■ *Response*<br>› *The ICD-10 code for "occipital encephalocele" is Q01.2.*<br>› *The ICD-10 code for "subcoronal hypospadias" is Q54.1.*<br>› *The ICD-10 code for "bilateral club feet is Q66.8 (Clubfoot NOS).* | 1. Q01.2<br><br>2. Q54.1<br><br>3. Q66.8 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 10**<br><br>Cleft palate; micrognathia; low set ears; posteriorly rotated ears; excess nuchal skin posteriorly; bilateral 5th finger clinodactyly; missing middle phalanx on finger; moderate to severe right hydronephrosis with thinning of the renal cortex<br><br>■ *Response*<br>› *The ICD-10 code for "cleft palate" is Q35.9 (Cleft palate, unspecified).*<br>› *The ICD-10 code for "missing middle phalanx on finger" is Q71.3 (Congenital absence of hand and finger(s)) or the ICD-10-RCPCH adaptation code Q71.30 (Congenital absence of finger(s)).*<br>› *The ICD-10 code for "hydronephrosis" is Q62.0 (Congenital hydronephrosis).*<br><br>The other malformations listed are considered minor anomalies. Coding them is optional. If coded:<br>› *The ICD-10 code for "micrognathia" is Q75.8 or K07.00 (although not in chapter Q).*<br>› *The ICD-10 code for "low set ears" is Q17.4.*<br>› *The ICD-10 code for "posteriorly rotated ears" is Q17.4.*<br>› *The ICD-10 code for "excess nuchal skin posteriorly" is Q18.3*<br>› *The ICD-10 code for "bilateral 5th finger clinodactyly" is Q68.1 or the more specific ICD-10-RCPCH adaptation code Q68.10.* | 1. Q35.9<br>2. Q75.8<br>3. Q17.4<br>4. Q17.4<br>5. Q18.3<br>6. Q68.1<br>7. Q71.3<br>8. Q62.0 |
| **CASE 11**<br><br>Craniorachischisis<br><br>■ *Response*<br>› *The ICD-10 code for "craniorachischisis" is Q00.1.* | 1. Q00.1 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 12**<br><br>Cleft lip NOS; spina bifida NOS; ear tags<br><br>■ *Response*<br>› *The abbreviation "NOS" means "not otherwise specified".*<br>› *The ICD-10 code for "cleft lip NOS" is Q36.9.*<br>› *The ICD-10 code for "spina bifida NOS" is Q05.9.*<br>› *Ear tags are considered minor anomalies; therefore, coding them is optional. If coded, the ICD-10 code for "ear tags" is Q17.0 (Preauricular appendage or tag).*<br><br>*Note*: Although "NOS" is a valid term in the ICD-10, it should be used only when there is no possibility of obtaining a better description for a specific congenital anomaly. | 1. Q36.9<br><br>2. Q05.9<br><br>3. Q17.0 |
| **CASE 13**<br><br>Anencephaly; absence of digits NOS; malformed feet NOS<br><br>■ *Response*<br>› *The abbreviation "NOS" means "not otherwise specified".*<br>› *The ICD-10 code for "anencephaly" is Q00.0.*<br>› *The ICD-10 code for "absence of digits NOS" is Q73.8 or the ICD-10-RCPCH adaptation code Q73.80 (Absent digits, unspecified).*<br>› *The ICD-10 code for "malformed feet NOS" is Q66.9 (Congenital deformity of feet, unspecified).*<br><br>*Note*: Although "NOS" is a valid term in the ICD-10, it should be used only when there is no possibility of obtaining a better description for a specific congenital anomaly. | 1. Q00.0<br><br>2. Q73.8/Q73.80<br><br>3. Q66.9 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 14**<br><br>Myelomeningocele, T3–T4 open; epicanthal folds; high arch palate; hypoplastic nipples<br><br>■ *Response*<br>› *The ICD-10 code for "myelomeningocele" is Q05.6 (Thoracic spina bifida without hydrocephalus). T3–T4 indicates the level of lesion in the thoracic region (the spine is open at the T3–T4 vertebrae).*<br><br>*The other malformations listed are considered minor anomalies. Coding them is optional. If coded:*<br><br>› *The ICD-10 code for "epicanthal folds" is Q10.3.*<br><br>› *The ICD-10 code for "high arch palate" is Q38.50.*<br><br>› *The ICD-10 code for "hypoplastic nipples" is Q83.8.*<br><br>*Note*: Since it is not mentioned or specified whether hydrocephalus is present or not, it is an assumption of "spina bifida without hydrocephalus". It is possible to use the ICD-10 code Q05.9 (Spina bifida, unspecified) but by using this code, the specificity for lesion level would not be captured. It is recommended that the congenital anomalies surveillance programme includes information in its protocol on how to code spina bifida when hydrocephalus is not mentioned or described in the medical records. | 1. Q05.6/Q05.9<br><br>2. Q10.3<br><br>3. Q38.50<br><br>4. Q83.8 |
| **CASE 15**<br><br>Hypoplastic left heart syndrome (HLHS); spina bifida occulta<br><br>■ *Response*<br>› *The ICD-10 code for "hypoplastic left heart syndrome" is Q23.4.*<br><br>› *"Spina bifida occulta" (Q76.0) is considered a minor anomaly.* | 1. Q23.4<br><br>2. Q76.0 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 16**<br><br>Unilateral (right side) cleft lip with cleft hard palate; bilateral talipes equinovarus<br><br>■ *Response*<br>› *The ICD-10 code for "unilateral (right side) cleft lip with cleft hard palate" is Q37.1 or the ICD-10-RCPCH adaptation code Q37.10 (Cleft hard palate with cleft lip, specified as unilateral).*<br>› *The ICD-10 code for "bilateral talipes equinovarus" is Q66.0.* | 1. Q37.1/Q37.10<br><br>2. Q66.0 |
| **CASE 17**<br><br>Left radial hypoplasia; transposition of the great arteries; secundum ASD, 3–4 mm<br><br>■ *Response*<br>› *The ICD-10 code for "left radial hypoplasia" is Q71.4.*<br>› *The ICD-10 code for "transposition of the great arteries" is Q20.3 (Discordant ventriculoarterial connection).*<br>› *The ICD-10 code for "secundum ASD" is Q21.1.* | 1. Q71.4<br><br>2. Q20.3<br><br>3. Q21.1 |
| **CASE 18**<br><br>Gastroschisis – large and intact pink intestine outside abdominal wall; large hiatal hernia; very narrow malrotated bowel<br><br>■ *Response*<br>› *The ICD-10 code for "gastroschisis" is Q79.3.*<br>› *The ICD-10 code for "large hiatal hernia" is Q40.1 (Congenital hiatus hernia).*<br>› *The ICD-10 code for "malrotated bowel" is Q43.9.* | 1. Q79.3<br><br>2. Q40.1<br><br>3. Q43.9 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 19**<br><br>Urethral meatus opens in the shaft of the penis; tetralogy of Fallot with massive ASD ostium secundum type<br><br>■ *Response*<br>› *ASD (atrial septal defect) is not part of tetralogy of Fallot, so needs to be coded separately. The ICD-10 code for "ASD" is Q21.1.*<br>› *The ICD-10 code for "tetralogy of Fallot" is Q21.3.*<br>› *The ICD-10 code for "urethral meatus opens in the shaft of the penis" is Q54.1 (Hypospadias, penile).*<br><br>*Note*: Tetralogy of Fallot (TOF) is a single entity consisting of four heart malformations: overriding aorta, pulmonary valve stenosis, ventricular septal defect (VSD) and right ventricular hypertrophy. | 1. Q21.1<br><br>2. Q21.3<br><br>3. Q54.1 |
| **CASE 20**<br><br>Holoprosencephaly; cleft lip bilateral<br><br>■ *Response*<br>› *The ICD-10 code for "holoprosencephaly" is Q04.2.*<br>› *The ICD-10 code for "cleft lip bilateral" is Q36.0.*<br><br>*Note*: Cleft lip is included but this case would not be counted in studies of clefts. | 1. Q04.2<br><br>2. Q36.0 |
| **CASE 21**<br><br>Gastroschisis with most of the abdominal contents expelled through abdominal wall defect; split hand<br><br>■ *Response*<br>› *The ICD-10 code for "gastroschisis" is Q79.3.*<br>› *The ICD-10 code for "split hand" is Q71.6.* | 1. Q79.3<br><br>2. Q71.6 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 22**<br><br>Absent right foot; hypoplasia of femur and tibia right leg; 3 toes missing on left foot; club right hand<br><br>■ *Response*<br>› *The ICD-10 code for "absent right foot" is Q72.3 (Congenital absence of foot and toe(s)). This code includes the "3 toes missing on left foot" diagnosis. However, using the more specific ICD-10-RCPCH adaptation, the code for the missing toes would be Q72.30, which could be added as an additional code since the toes are missing on the left foot, keeping the ICD-10 code Q72.3 for the missing right foot.*<br><br>*Although, the clinical description "hypoplasia of femur and tibia right leg" may sound like one anomaly affecting the right leg, there are different codes for femoral and tibial hypoplasia.*<br><br>› *The ICD-10 code for "hypoplasia of femur" is Q72.4.*<br><br>› *The ICD-10 code for "hypoplasia of tibia" is Q72.5.*<br><br>› *The ICD-10 code for "club right hand" is Q71.4.* | 1. Q72.3/Q72.30<br><br>2. Q72.4<br><br>3. Q72.5<br><br>4. Q71.4 |
| **CASE 23**<br><br>Spina bifida, cervical without hydrocephalus<br><br>■ *Response*<br>› *The ICD-10 code for "spina bifida, cervical without hydrocephalus" is Q05.5.* | 1. Q05.5 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 24**<br><br>Cleft soft palate; tetralogy of Fallot; spina bifida, sacral with hydrocephalus; oligodactyly on foot<br><br>■ *Response*<br>› *The ICD-10 code for "cleft soft palate" is Q35.3.*<br>› *The ICD-10 code for "tetralogy of Fallot" is Q21.3.*<br>› *The ICD-10 code for "spina bifida sacral" is Q05.3 (Sacral spina bifida with hydrocephalus).*<br>› *The ICD-10 code for "oligodactyly on foot" is Q72.3 (Congenital absence of foot and toes) or the ICD-10-RCPCH adaptation code Q72.30 (Congenital absence or hypoplasia of toe(s) with remainder of foot intact).*<br><br>*Note*: Tetralogy of Fallot (TOF) is a single entity consisting of four heart malformations: overriding aorta, pulmonary valve stenosis, VSD and right ventricular hypertrophy | 1. Q35.3<br>2. Q21.3<br>3. Q05.3<br>4. Q72.3/Q72.30 |
| **CASE 25**<br><br>Tibial hypoplasia, right; ulnar hypoplasia, right<br><br>■ *Response*<br>› *The ICD-10 code for "ulnar hypoplasia, right" is Q71.5.*<br>› *The ICD-10 code for "tibial hypoplasia, right" is Q72.5.* | 1. Q71.5<br>2. Q72.5 |
| **CASE 26**<br><br>Pierre Robin sequence<br><br>■ *Response*<br>› *The ICD-10 code for "Pierre Robin sequence" is the ICD-10-RCPCH adaptation code Q87.08.* | 1. Q87.08 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 27**<br><br>Anencephaly infant with gross abnormalities; bilateral cleft lip; cleft palate<br><br>■ *Response*<br>› *The ICD-10 code for "anencephaly" is Q00.0 (Anencephaly).*<br>› *The "gross abnormalities" description is vague, and coding is optional. If coded, the ICD-10 code is Q89.9 (Congenital malformation, unspecified).*<br>› *Although, the description may suggest two anomalies (cleft lip and cleft palate), there is a specific ICD-10 code to assign to cleft palate with bilateral cleft lip. Because the type of cleft palate is not specified, the ICD-10 code is Q37.8 (Unspecified cleft palate with bilateral cleft lip).*<br><br>*Note*: Avoid using the Q89.9 ICD-10 code if possible because it does not provide any specificity, and it has very minimal value in congenital anomalies surveillance. | 1. Q00.0<br><br>2. Q89.9<br><br>3. Q37.8 |
| **CASE 28**<br><br>Iniencephaly; complete amelia of upper limb<br><br>■ *Response*<br>› *The ICD-10 code for "iniencephaly" is Q00.2.*<br>› *The ICD-10 code for "complete amelia of upper limb" is Q71.0.* | 1. Q00.2<br><br>2. Q71.0 |
| **CASE 29**<br><br>Short limbs (possible achondroplasia)<br><br>■ *Response*<br>› *The ICD-10 code for "achondroplasia" would be Q77.4. However, the diagnosis is not definite. Generalized limb shortening including skeletal dysplasias are on the exclusion list in WHO/CDC/ICBDSR* Birth defects surveillance: a manual for programme managers (4). | 1. Q77.4 |

| Cases – clinical description and response key for facilitator | ICD-10 or ICD-10-RCPCH codes |
|---|---|
| **CASE 30**<br><br>Amelia upper and lower limbs<br><br>■ *Response*<br>› *There are two ICD-10 codes to be assigned. One is for "amelia of upper limbs": Q71.0 (Congenital complete absence of upper limb(s), amelia); the other is for "amelia of lower limbs": Q72.0 (Congenital complete absence of lower limb(s), amelia).* | 1. Q71.0<br><br>2. Q72.0 |

## Evaluation questions 5

Expected time: 30 minutes

Correct answers are presented in **bold**.

1. It is important to understand and follow a _____ coding system, in order to accurately and consistently _____ and _____ the various types of congenital anomalies.

**Answers:** standardized, classify, code

2. What type of supplemental information can help a reviewer assign a proper code to a case?

**Answer:** a detailed description of the congenital anomaly, or copies or excerpts of clinical reports (e.g. surgery, imaging, autopsy report); photographs

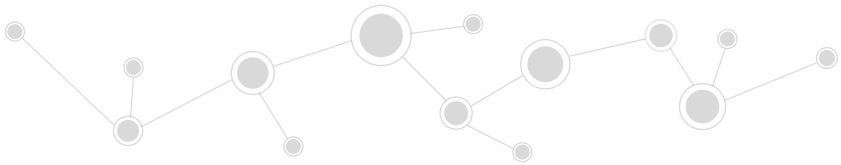
3. What is the international standard diagnostic classification system?

**Answer:** *International statistical classification of diseases and related health problems*, tenth edition (ICD-10)

4. What are some ways in which a surveillance programme can work around the ICD-10 coding system's lack of specificity for certain congenital anomalies?

**Possible answers:**

- Use its own local modification that includes additional codes
- Add an extra digit for more detailed coding
- Refer to the RCPCH's adaptation of the ICD-10



5. **True** or false: Final coding will always be at the central registry.
6. Which of the following does NOT cause difficulties when coding congenital anomalies?
   a. Prenatal diagnosis
   b. Live births where the neonate dies shortly after birth
   c. **Confirmed diagnosis**
   d. Possible diagnosis
7. **True** or false: Capturing major anomalies should be prioritized over capturing minor anomalies on the data-collection form.
8. **True** or false: The ICD-10 is developed and maintained by WHO.

## References


1. World Health Organization. Congenital anomalies. Fact sheet No 370. October 2012 (http://www.who.int/mediacentre/factsheets/fs370/en/index.html, accessed 29 April 2015).

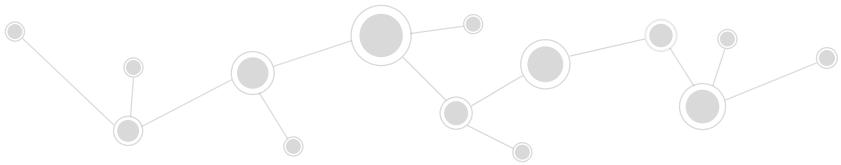
2. Resolution WHA63.17. Birth defects. In: Sixty-third World Health Assembly, Geneva, 17–21 May 2010. Geneva: World Health Organization; 2010 (http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_R17-en.pdf, accessed 29 April 2015).

3. International statistical classification of diseases and related health problems, 10th revision. Geneva: World Health organization; 2015 (http://apps.who.int/classifications/icd10/browse/2015/en, accessed 24 February 2015).

4. World Health Organization, National Center on Birth Defects and Developmental Disabilities from the United States Centers for Disease Control and Prevention (CDC), International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR). Birth defects surveillance: a manual for programme managers. Geneva: World Health Organization; 2014 (http://www.cdc.gov/ncbddd/birthdefectscount/documents/bd-surveillance-manual.pdf, accessed 10 February 2015).

5. World Health Organization, National Center on Birth Defects and Developmental Disabilities from the United States Centers for Disease Control and Prevention (CDC), International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR). Birth defects surveillance: atlas of selected congenital anomalies. Geneva: World Health Organization; 2014 (http://apps.who.int/iris/bitstream/10665/127941/1/9789241564762_eng.pdf?ua=1, accessed 10 February 2015).

6. CDC Foundation. What is public health? (http://www.cdcfoundation.org/content/what-public-health, accessed 24 February 2015).



## Annex 1

**Congenital anomalies surveillance: tentative programme**

| Time | Item | Presenter |
|---|---|---|
| ***Day 1*** | | |
| 08:00–08:30 | • Registration | All |
| 08:30–09:00 | • Welcome and introductions | |
| 09:00–09:15 | • Overview of workshop | |
| 09:15–09:30 | • National overview of neural tube defects | |
| 09:30–09:45 | • Congenital anomalies in the country | |
| 09:45–10:45 | • Global overview of congenital anomalies | |
| 10:45–11:15 | *Coffee and tea break; group photograph* | |
| 11:15–12:00 | • Folic acid fortification and neural tube defects prevention | |
| 12:00–13:00 | • Congenital anomalies: prevention, prioritization, and modifiable risk factors | |
| 13:00–14:00 | *Lunch* | |
| 14:00–14:30 | • Congenital anomalies surveillance: an overview<br>• Discussion | |
| 14:30–15:00 | • Planning activities and tools: logic models | |
| 15:00–16:30 | • Logic model activity | |
| 16:30–17:00 | • Review and plans for Day 2 | All |

| Time | Item | Presenter |
|---|---|---|
| ***Day 2*** | | |
| 08:00–09:00 | • Welcome and review of Day 1 | |
| 09:00–10:30 | • Legislation, privacy and confidentiality<br>*Activity* | |
| | • Partnerships<br>*Activity* | |
| 10:30–11:00 | *Coffee and tea break* | |
| 11:00–11:30 | • Population coverage | |
| 11:30–12:15 | • Case ascertainment<br>*Group discussion* | |
| 12:15–13:15 | • Case inclusion<br>*Group discussion* | |
| 13:15–14:15 | *Lunch* | |
| 14:15–14:45 | • Case-finding and description formats<br>*Group discussion* | |
| 14:45–15:45 | • Data dissemination<br>*Presentation* | |
| 15:45–16:15 | *Coffee and tea break* | |
| 16:15–16:30 | • Local speaker invited | |
| 16:30–17:00 | • Review and plans for Day 3 | All |

| Time | Item | Presenter |
|---|---|---|
| ***Day 3*** | | |
| 08:00–08:30 | • Welcome and review of Day 2 | |
| 08:30–09:00 | • Core ascertainment variables<br>*Activity*<br>*Group discussion* | |
| 09:00–10:30 | • Data collection and management | |
| 10:30–11:00 | *Coffee and tea break* | |
| 11:00–12:00 | • Data analysis | |
| 12:00–13:30 | *Lunch* | |
| 13:30–14:00 | • Data analysis<br>*Activity* | |
| 14:00–15:00 | • Data dissemination<br>*Activity* | |
| 15:00–16:00 | • Clinical review of congenital anomalies<br>*Activity* | |
| 16:00–16:30 | *Coffee and tea break* | |
| 16:30–17:00 | • Presentation from coding authority in country | |
| 17:00–17:30 | • Review and plans for Day 4 | All |

| Time | Item | Presenter |
|---|---|---|
| ***Day 4*** | | |
| 08:00–08:30 | • Welcome and review of Day 3 | |
| 08:30–10:00 | • Overview of congenital anomalies coding | |
| 10:00–10:30 | *Coffee and tea break* | |
| 10:30–12:30 | • Database presentation | |
| 12:30-13:15 | *Lunch* | |
| 13:15–15:00 | • Database activity | |
| 15:00 | • Review and plans for Day 5 | All |

| Time | Item | Presenter |
|---|---|---|
| ***Day 5*** | | |
| 08:00-08:30 | • Welcome and review of Day 4 | |
| 08:30–10:00 | • Overview of congenital anomalies coding<br>*Activity* | |
| 10:00–10:30 | *Coffee and tea break* | |
| 10:30–12:30 | • Development of country plan | All |
| 12:30–13:15 | *Lunch* | |
| 13:15–13:45 | • Discussion of country plan or next steps | |
| 13:45–14:45 | • Closing remarks and evaluation of programme | All |
| 15:00 | *Departure* | |

## Annex 2

### Pre-course participant assessment of country status

Congenital anomalies surveillance pre-course assessment

*Note:* Participants should receive this assessment prior to the workshop, and bring it with them to the workshop for discussion.

Please bring responses to the following questions with you to the workshop and be prepared to discuss the current situation in your country related to congenital anomalies surveillance.

1. In your country, is there a vital registry system?

_____

2. In your country, is there any information collected on live births?

_____

3. In your country, is there any information collected on fetal deaths/stillbirths?

_____

4. In your country, is there any information collected on terminations of pregnancy?

_____

5. In your country, is there any surveillance system collecting information about the occurrence of birth defects?

_____

6. In your country, is there any surveillance system collecting information about the occurrence of neural tube defects such as anencephaly and/or spina bifida?

_____

   a. If there is a surveillance system collecting information about the occurrence of neural tube defects, how are the data used?

   _____

7. Within your country, what is the estimated percentage of births that occur in hospitals?

_____

   a. What are the primary reasons why not all births occur in hospitals?

   _____

   _____

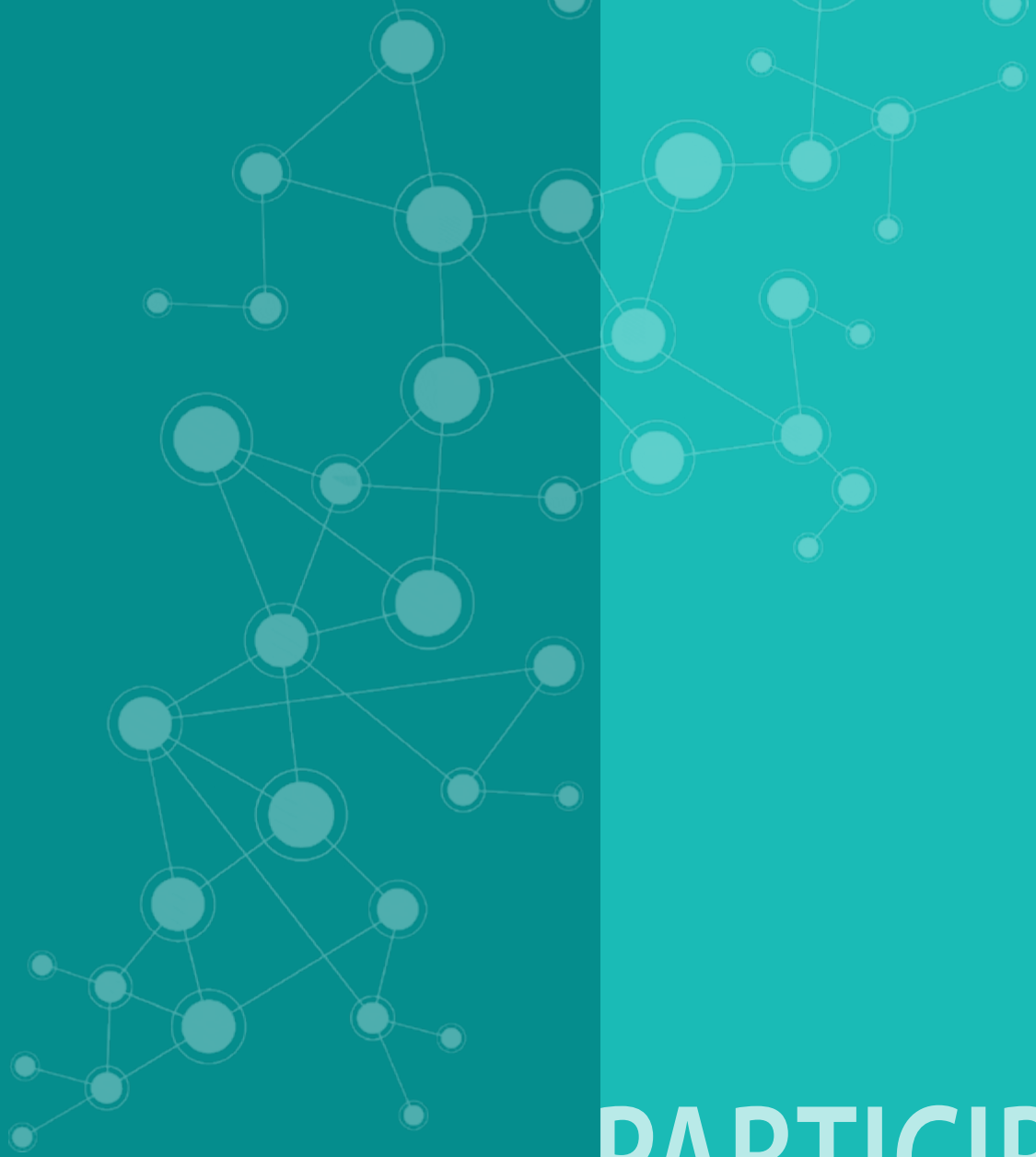
   _____

8. What are the leading causes of neonatal and infant mortality in your country?

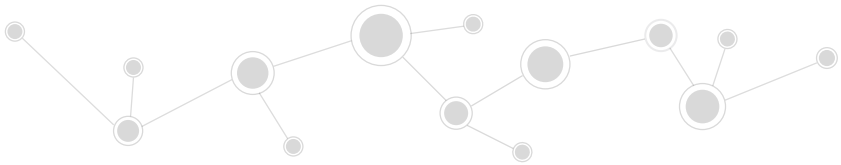
9. What kind of legislation, if any, exists in your country for the reporting of health conditions? What are some health conditions that require mandatory reporting?

10. What kind of laws, if any, are in place in your country related to privacy and confidentiality of patient data?

11. Is informed consent required in your country before being able to share patient information?



# PARTICIPANT WORKBOOK



## 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-term 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: |

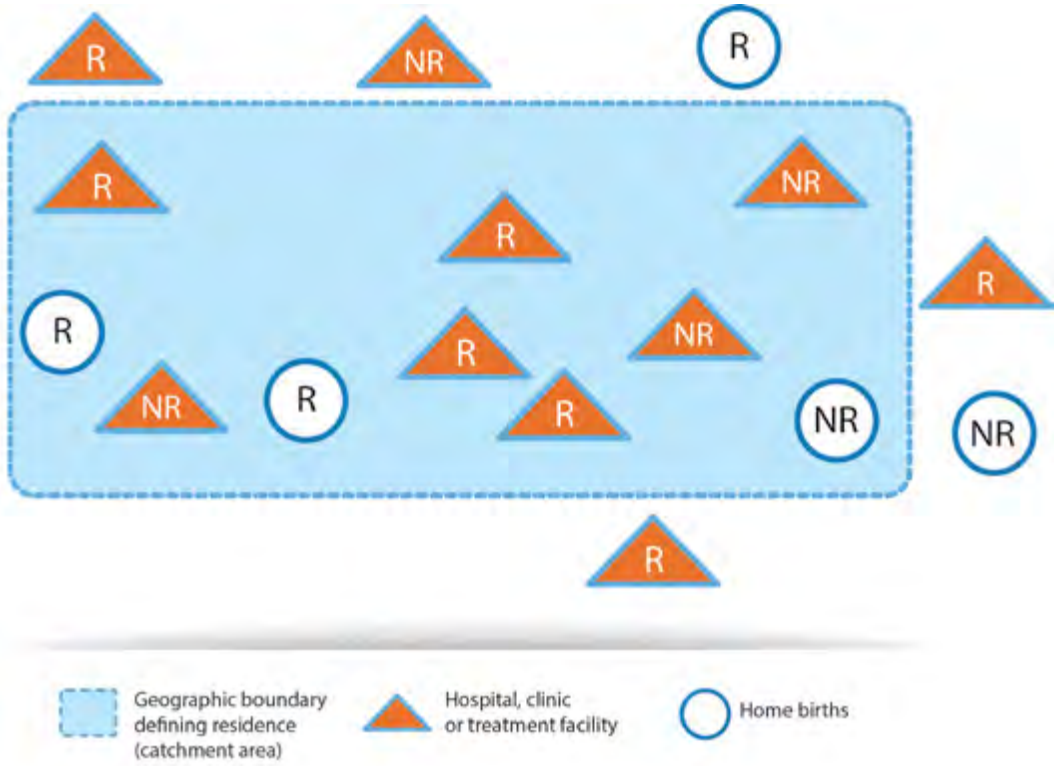
## Activity 2.2

- 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 | | | |

**Activity 3.1**

- Study the figure below.

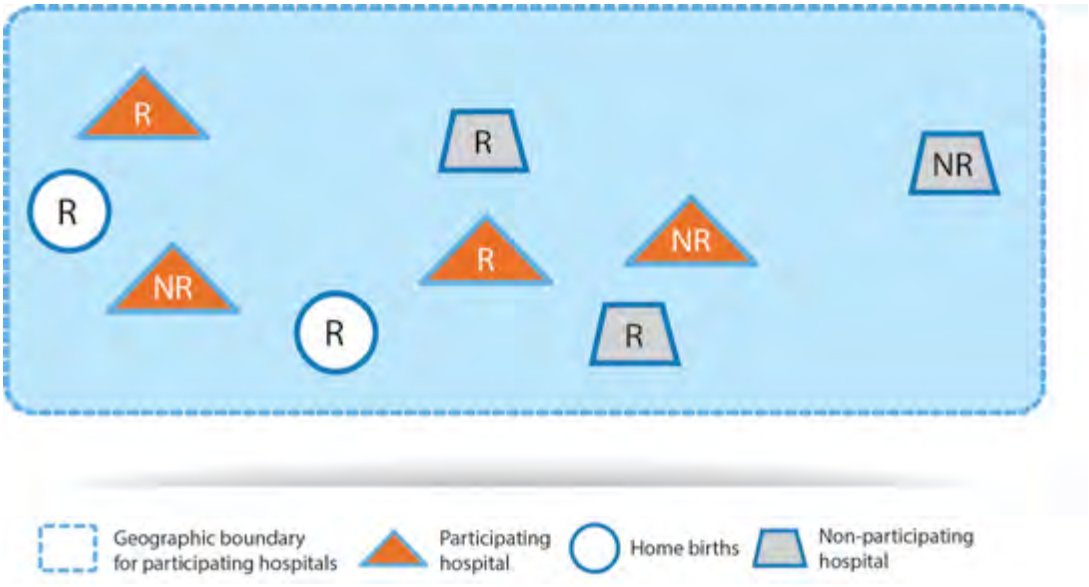



**R** = fetus or neonate with congenital anomaly whose mother is a *resident*.
**NR** = fetus or neonate with congenital anomaly whose mother is a *non-resident*.

- 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 3.1 continued**

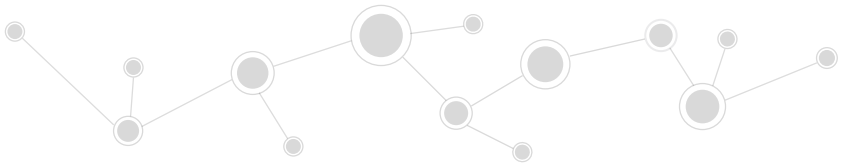
- Study the figure below.




**R** = fetus/neonate with a birth defect whose mother is a *resident* .

**NR** = fetus/neonate with a birth defect whose mother is a *non-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**

- 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 based or population based.

**Activity 3.3**

- Review the table below and consider the suggested core ascertainment variables.
- Complete the blank column in the table with the reason each variable should be presented.

| 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 3.4

- 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:

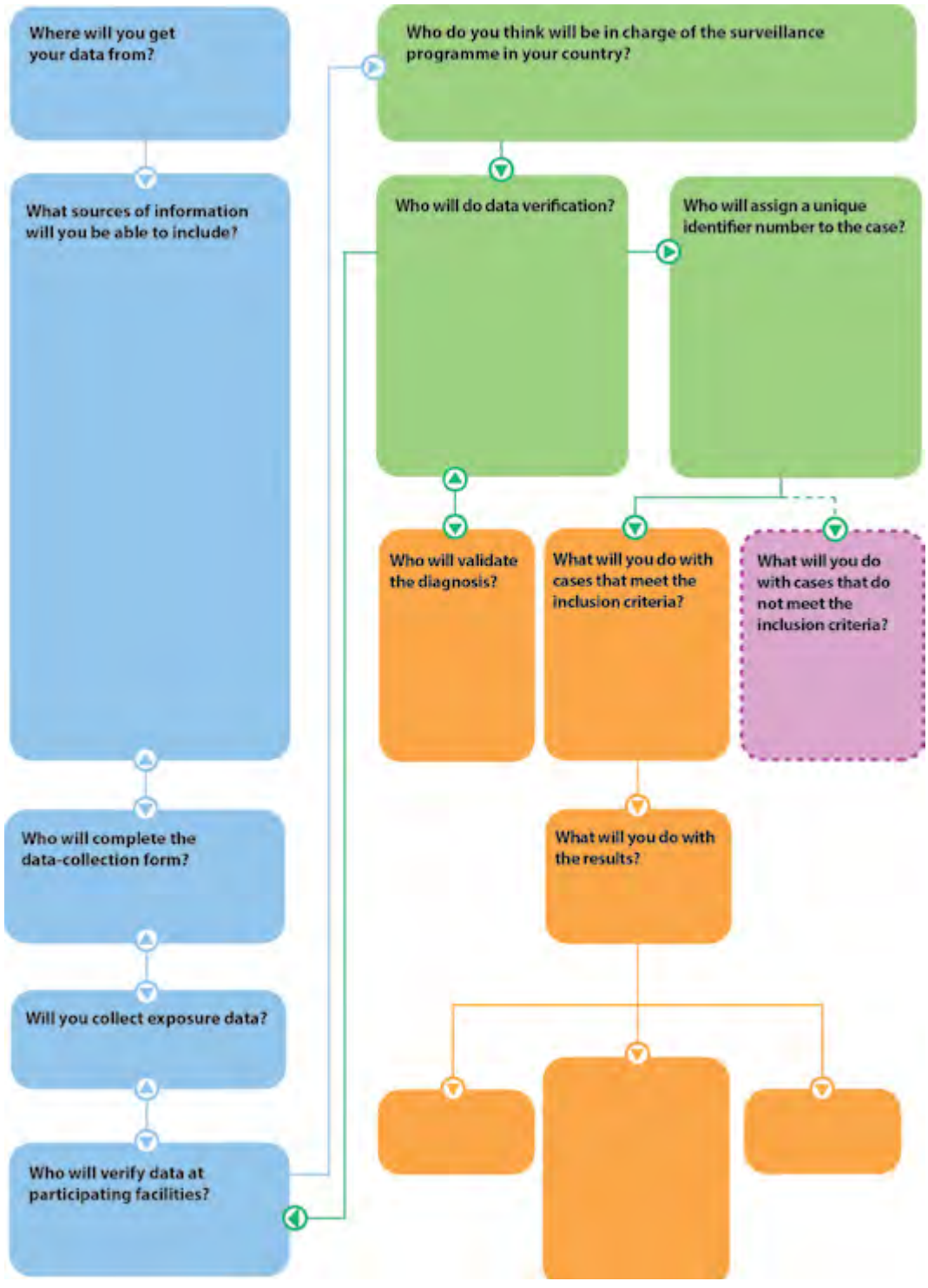
* C = Confirmed diagnosis
P = Possible diagnosis

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

Version: January 2014

**Activity 3.5**

- Complete the flow chart below.



## Activity 3.6

- Read the case-study below.

**Case-study: Cases of neural tube defects by type of ascertainment, United States of America (USA), 2004–2006**

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 2010, data for 21 congenital anomalies from 2004–2006 were presented as national congenital anomalies prevalence estimates. 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, 2004–2006**

| | Number of cases | | | |
|---|---|---|---|---|
| **Neural tube defects** | **Active ascertainment (11 programmes)[a]** | **Hybrid ascertainment (6 programmes)[b]** | **Passive ascertainment (7 programmes)[c]** | **National** |
| Anencephaly | 697 | 211 | 192 | 1100 |
| Spina bifida | 1162 | 561 | 820 | 2543 |
| Encephalocele | 261 | 125 | 184 | 570 |
| Total neural tube defects | 2120 | 897 | 1196 | 4213 |

*Source*: Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE et al. Updated national birth prevalence estimates for selected congenital anomalies in the United States 2004–2006. Birth Defects Res A Clin Mol Teratol. 2010; 88:1008–16.

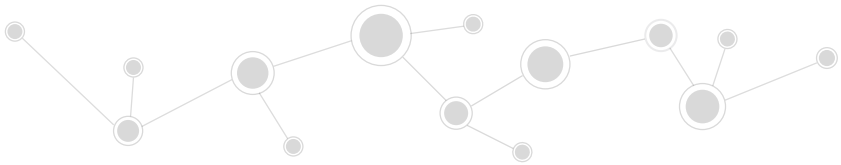

Data from programmes with active, hybrid or passive ascertainment.
[a] Number of live births in the active ascertainment programmes: 3 120 605.
[b] Number of live births in the hybrid ascertainment programmes: 2 075 973.
[c] Number of live births in the passive ascertainment programmes: 2 145 287.

- 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, 2004–2006**

| | Active ascertainment (11 programmes) | | Hybrid ascertainment (6 programmes) | | Passive ascertainment (7 programmes) | | National | |
|---|---|---|---|---|---|---|---|---|
| **Neural tube defects** | **Cases** | **Prevalence** | **Cases** | **Prevalence** | **Cases** | **Prevalence** | **Cases** | **Prevalence** |
| Anencephaly | 697 | | 211 | | 192 | | 1100 | |
| Spina bifida | 1162 | | 561 | | 820 | | 2543 | |
| Encephalocele | 261 | | 125 | | 184 | | 570 | |
| Total neural tube defects | 2120 | | 897 | | 1196 | | 4213 | |

- 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 3.7**

- Read the case-study below.

**Case-study: Pre- and post-fortification birth prevalence of neural tube defects in the USA, 1999–2007**

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 (1995–1997) and following the folic acid fortification mandate (1998–2010). The estimated annual prevalence of neural tube defects for nine hospitals 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 (1995–2007)**

| Race/ethnicity | Year | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | 1995 | 1996 | 1997 | 1998 | 1999 | 2000 | 2001 | 2002 | 2003 | 2004 | 2005 | 2006 | 2007 |
| Hispanic | 9.20 | 10.84 | 9.69 | 7.37 | 7.83 | 6.45 | 6.63 | 6.98 | 6.95 | 6.63 | 6.27 | 5.69 | 6.04 |
| Black | 4.89 | 5.75 | 3.59 | 4.78 | 4.80 | 4.49 | 4.81 | 5.16 | 4.17 | 3.68 | 3.89 | 3.37 | 3.74 |
| Caucasian | 7.1 | 7.8 | 6.7 | 5.5 | 5.5 | 5.3 | 5.1 | 4.6 | 4.6 | 5.2 | 4.6 | 4.9 | 5.3 |

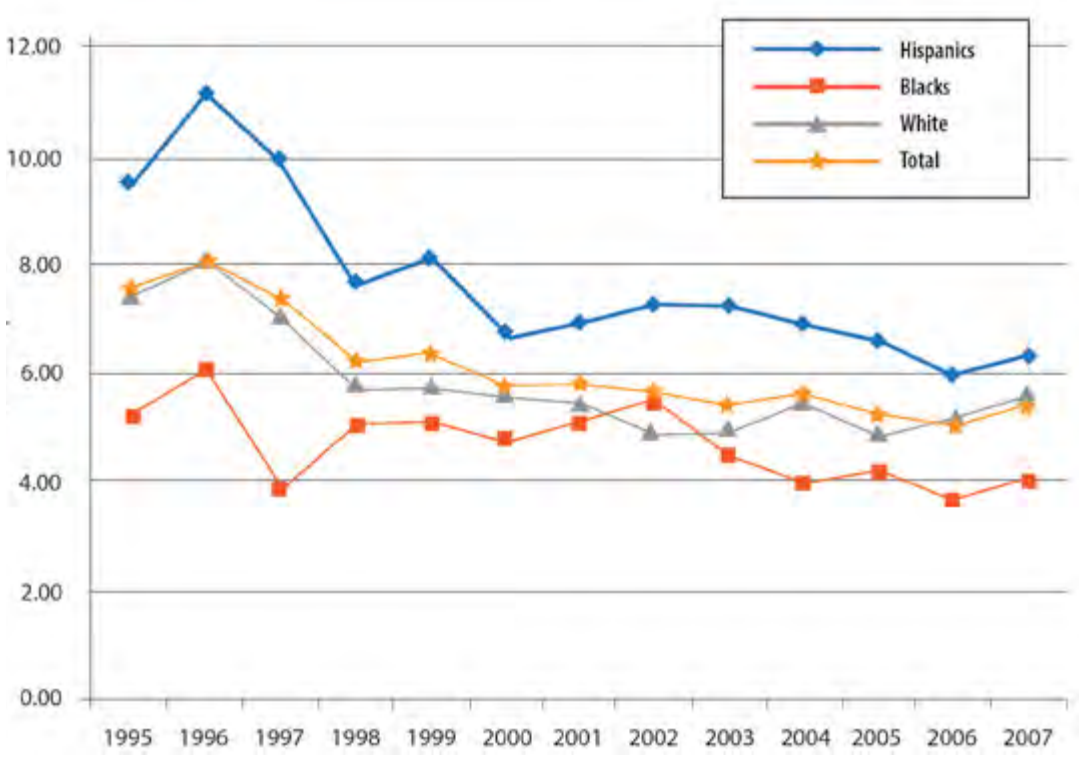
*Source:* CDC Grand Rounds: additional opportunities to prevent neural tube defects with folic acid fortification. MMWR Morb Mortal Wkly Rep. 2010;59(31):980–4.

- 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, 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.

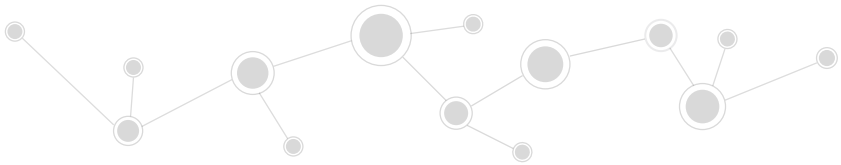
Typically in public health, trend data are presented as population-based rates. These data are accessed from large database systems such as national vital records, and show how rates change over relatively long periods of time, e.g. 10 years or more. Trend data can be visually presented through tables 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 race/ethnicity, United States, 1995–2010**




*Source:* National Birth Defects Prevention Network. Neural Tube Defect Ascertainment Project 2010 (http://www.nbdpn.org/current/2010pdf/NTD%20fact%20sheet%2001-10%20for%20website.pdf).

- 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 3.8**

- 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 3.9

- You are a group of paediatricians working in a large maternity facility in your country. You are seeing many babies (see table) 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, what 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.

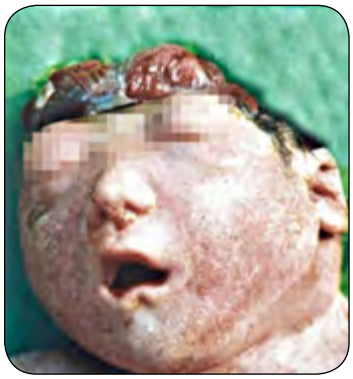
- **Target audience**
  - Groups 1 and 2: Ministry of health (government agency)
  - Groups 3 and 4: Clinic/public health practitioners (from other maternity facilities within the country)

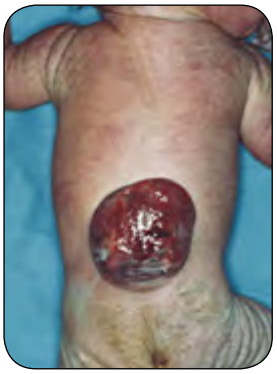
### Birth prevalence of congenital anomalies by race/ethnicity

| | Prevalence of anomalies 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) |

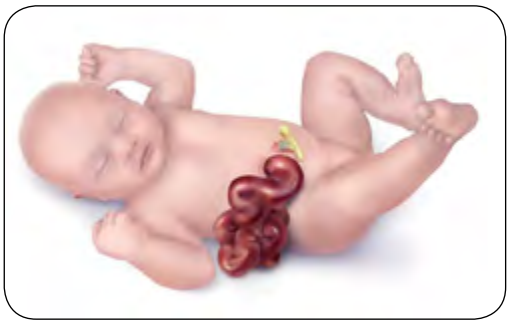
**Activity 4.1**

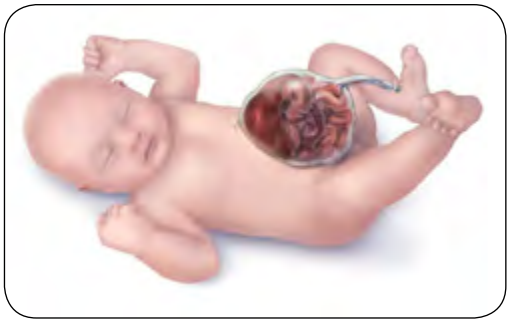
- Look at the three photos of congenital anomalies below and describe the differences.



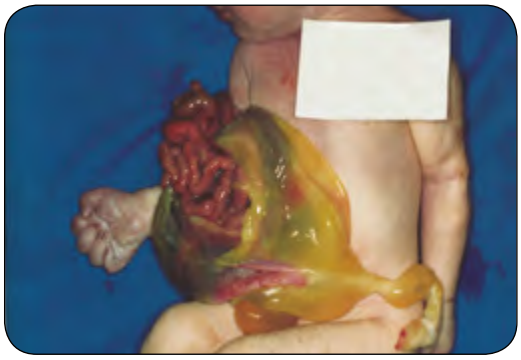


- Look at the two illustrations of congenital anomalies below and describe the differences.





- Look at the following photo. Do you think the baby has gastroschisis or omphalocele?



- Identify the diagnoses for each of the congenital anomalies below.

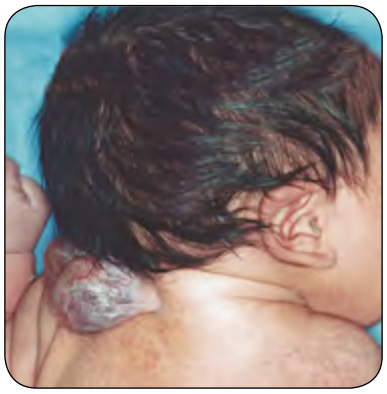


**Photo A**

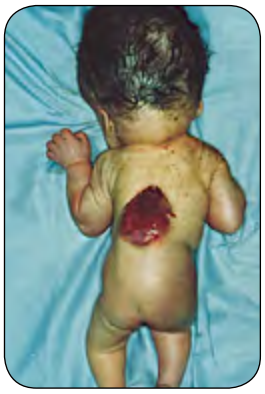


**Photo B**

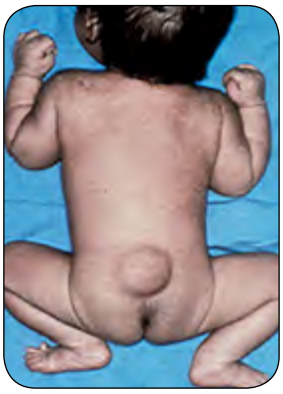


**Photo C**

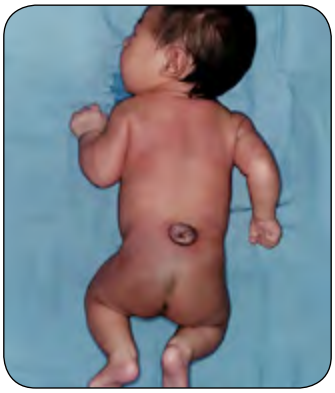


**Photo D**

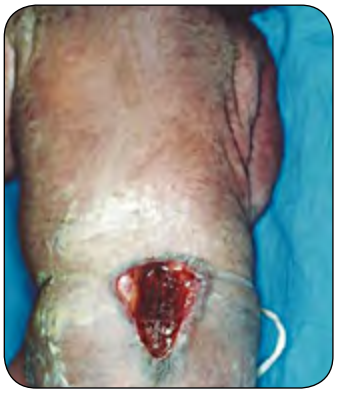
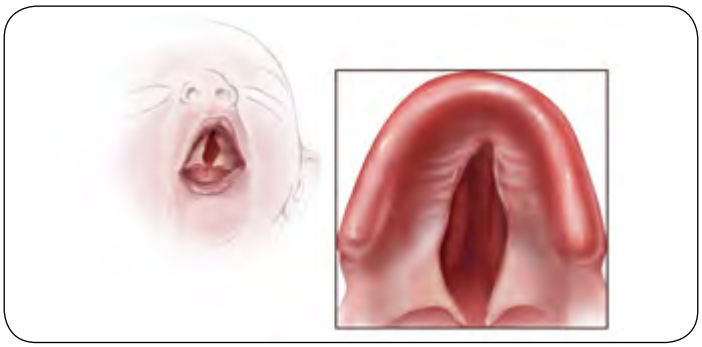


**Photo E**

**Activity 5.1**

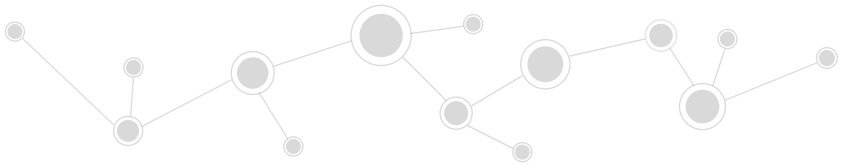
- Identify the diagnoses for each of the following congenital anomalies, and code each case.



**Activity 5.2**

A total of 20 cases are included in the 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.



**Activity 5.3**

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 meningocele; massive hydrocephalus.
- **Case 2**

  Frontal encephalocele; clubbing of left foot
- **Case 3**

  Cleft lip and palate; omphalocele
- **Case 4**

  Facial cleft; amniotic band evidence on face
- **Case 5**

  Small encephalocele in the parietal area; cleft palate NOS; fused toes NOS

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

- **Case 6**

  Anencephaly; heart defect NOS; spinal anomalies NOS; lower extremity abnormal development
- **Case 7**

  Hypospadias, penoscrotal; unilateral absent middle phalanx on foot (no further description)
- **Case 8**

  Transposition of the great arteries with intact ventricular septum (D-TGA); bilateral cleft lip and palate
- **Case 9**

  Occipital encephalocele; subcoronla hypospadias; bilateral club feet
- **Case 10**

  Cleft palate; micrognathia; low set ears; posteriorly rotated ears; excess nuchal skin posteriorly; bilateral 5th finger clinodactyly; missing middle phalanx on finger; moderate to severe right hydronephrosis with thinning of the renal cortex
- **Case 11**

  Craniorachischisis
- **Case 12**

  Cleft lip NOS; spina bifida NOS; ear tags
- **Case 13**

  Anencephaly; absence of digits NOS; malformed feet NOS
- **Case 14**

  Myelomeningocele, T3–T4 open; epicanthal folds; high arch palate; hypoplastic nipples

- **Case 15**

  Hypoplastic left heart syndrome (HLHS); spina bifida occulta
- **Case 16**

  Unilateral (right side) cleft lip with cleft hard palate; bilateral talipes equinovarus
- **Case 17**

  Left radial hypoplasia; transposition of the great arteries, secundum ASD, 3–4 mm
- **Case 18**

  Gastroschisis – large and intact pink intestine outside abdominal wall; large hiatal hernia; very narrow malrotated bowel
- **Case 19**

  Urethral meatus opens in the shaft of the penis; tetralogy of Fallot with massive ASD ostium secundum type
- **Case 20**

  Holoprosencephaly; cleft lip bilateral
- **Case 21**

  Gastroschisis with most of the abdominal contents expelled through abdominal wall defect; split-hand
- **Case 22**

  Absent right foot; hypoplasia of femur and tibia right leg; 3 toes missing on left foot; club right hand
- **Case 23**

  Spina bifida, cervical without hydrocephalus
- **Case 24**

  Cleft soft palate; tetralogy of Fallot; spina bifida, sacral with hydrocephalus; oligodactyly on foot
- **Case 25**

  Tibial hypoplasia, right; ulnar hypoplasia, right
- **Case 26**

  Pierre Robin sequence
- **Case 27**

  Anencephaly infant with gross abnormalities; bilateral cleft lip; cleft palate
- **Case 28**

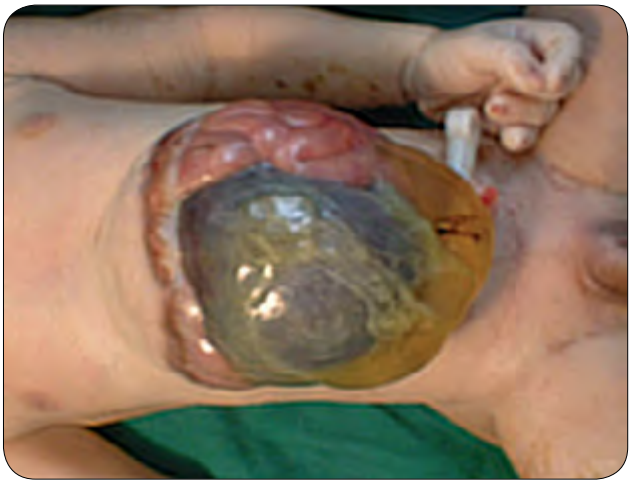
  Iniencephaly; complete amelia of upper limb
- **Case 29**

  Short limbs (possible achondroplasia)
- **Case 30**

  Amelia upper and lower limbs

# DESCRIPTION AND CODING **ANSWERS**

## CASE 1



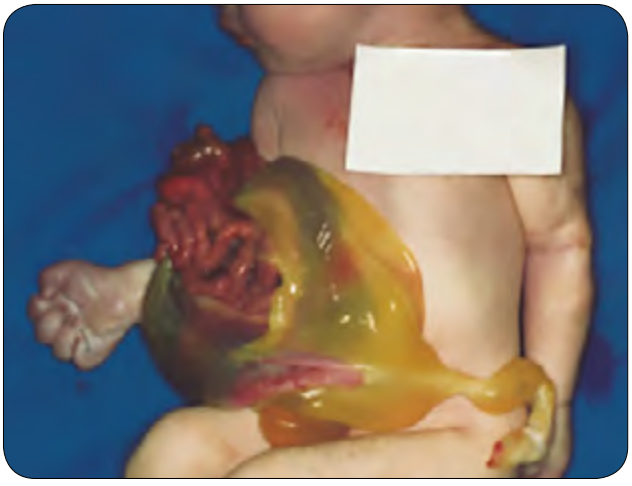
Large abdominal midline defect.

Some organs outside abdomen throughout the overstretched umbilical cord, the umbilical cord membrane covers the leaked intestine.

Omphalocele **Q79.2**

*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

## CASE 2



Very large abdominal midline defect.

Greater part of intestine outside abdomen through the overstretched umbilical cord (terminal part visible at lower left).

The translucent membrane is ruptured and covers only part of the herniated abdominal content.

Make sure you document the size (in cm).

Omphalocele **Q79.2**

*Source*: *Birth defects surveillance: atlas of selected congenital anomalies (5)*

## CASE 3

Small anterior abdominal defect on the right of the umbilical cord.

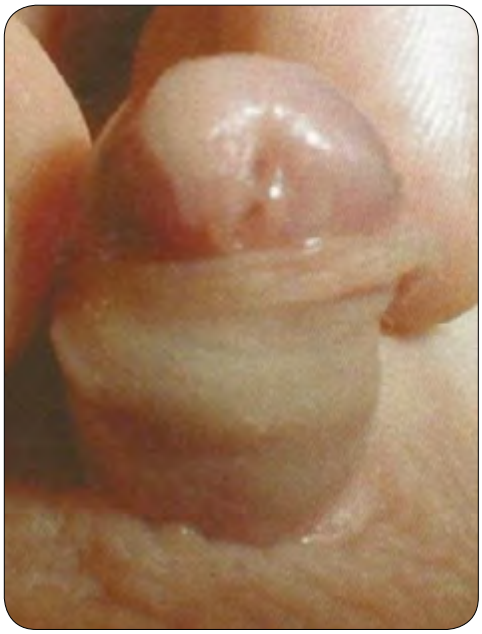
Most of small intestine outside of the abdomen through the abdominal opening.

The intestine outside of the body is not covered by any membrane.

Gastroschisis **Q79.3**

*Source*: *Birth defects surveillance: atlas of selected congenital anomalies (5)*

## CASE 4



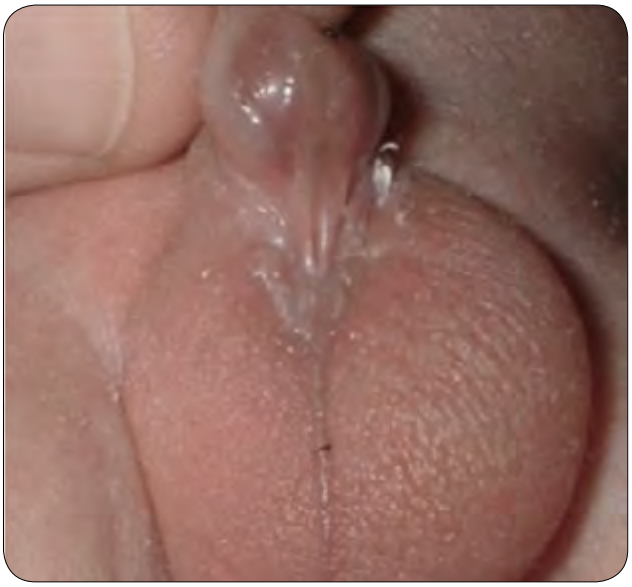
The external urethral meatus is not at the tip of the penis as it should be, but is on the ventral part of the glans.

Chordee not present (descended testes?)

Glan(d)ular first degree hypospadias **Q54.0**

*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

## CASE 5



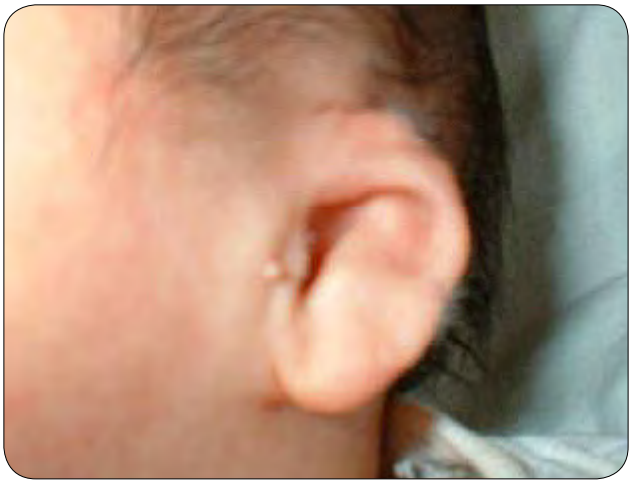
The external urethral meatus is not at the tip of penis but below the bases of the shat, just between the scrotal sacs.

Descended testes.

Penoscrotal third degree hypospadias **Q54.2**

*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

## CASE 6



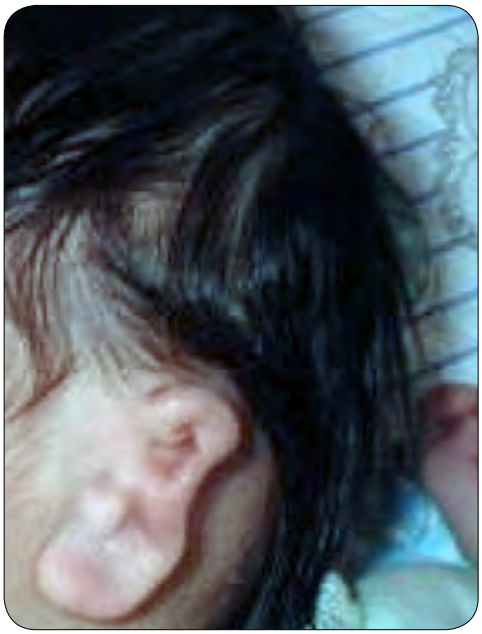
The left external ear is smaller than normal, the shape is simplified and some anatomical components are missing or abnormal. Upper portion of helix is missing.

Preauricular tags or pits present. Make sure you document the size (in cm).

Microtia type II **Q17.22**

*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

## CASE 7



The right external ear is replaced by vertical rudiments of soft tissue (peanut shape) that do not conform to recognized ear components.

Microtia type III **Q17.23**

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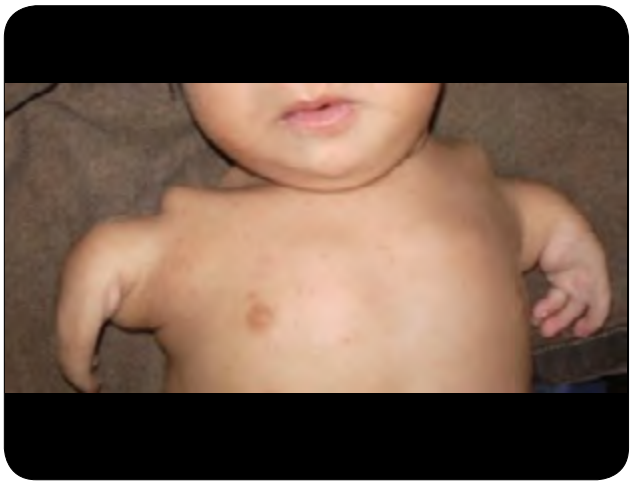
## CASE 8



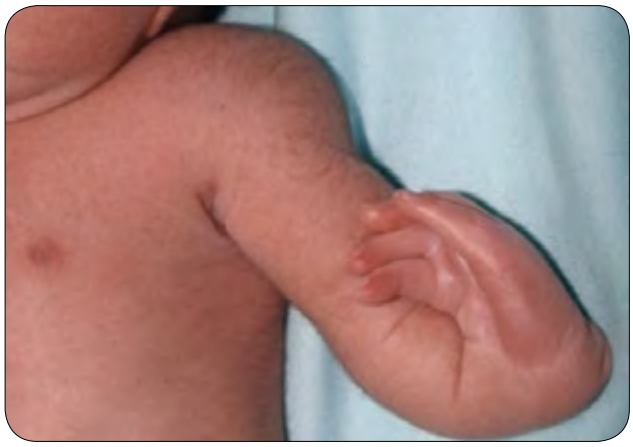
Bilateral and nearly symmetric defects of upper limbs.

Both defects are characterized by almost complete absence of arm and forearm, the hands are present but malformed with missing fingers.

Transverse intercalary upper limb defect (true phocomelia) **Q71.1**

*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

## CASE 9



Limb deficiency involving left forearm and hand.

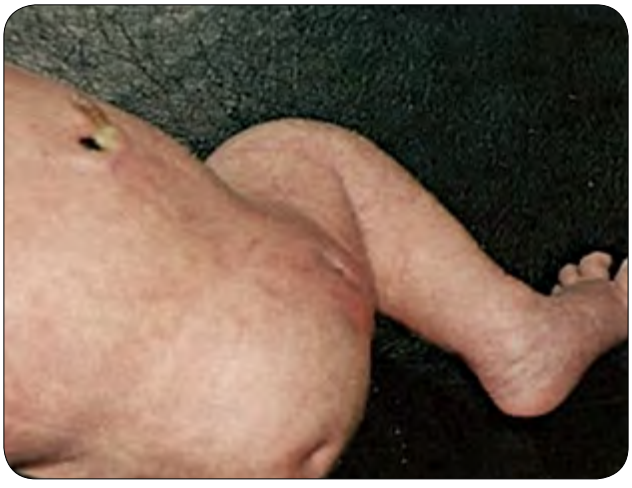
The forearm is shorter than normal, with radial deviation of the hand.

The hand shows only four fingers and the thumb is missing (replaced by the second finger).

Radial (preaxial) deficiency **Q71.4**

*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

## CASE 10

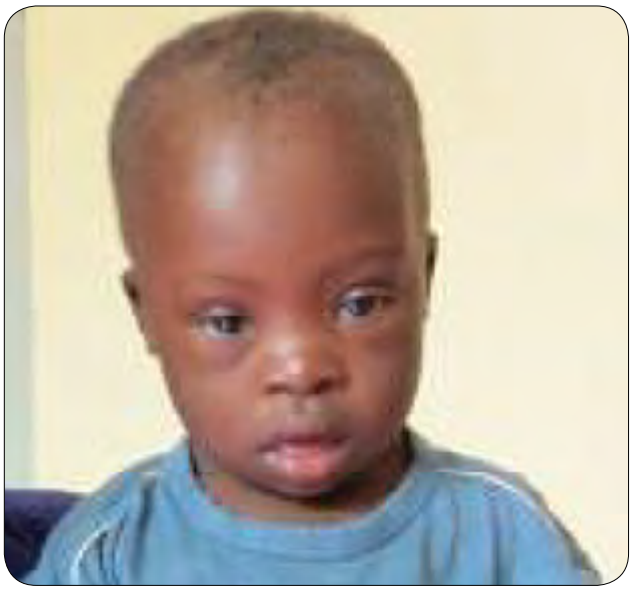


Complete absence of right lower limb.

Transverse lower limb deficiency **Q72.0**

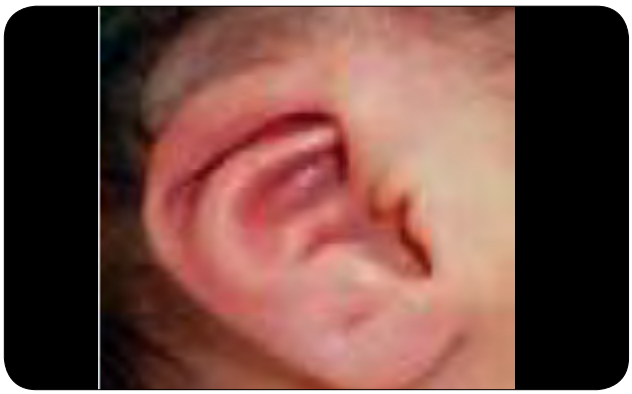
*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

## CASE 11



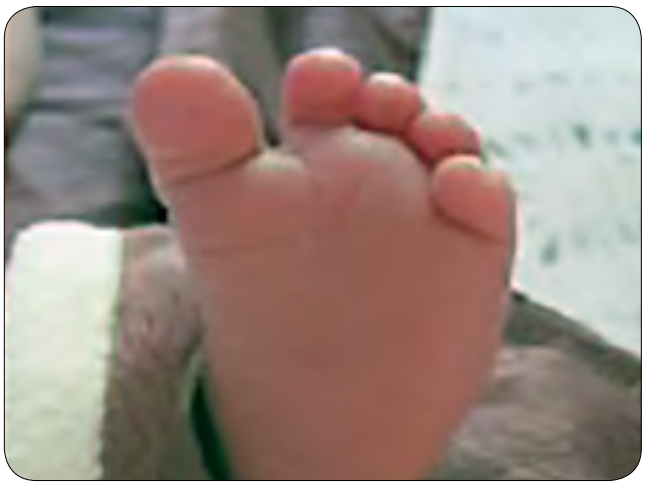
Distinctive facial traits: upslanting palpebral fissures (Q10.3), flat nasal bridge (Q30.8).

Down syndrome **Q90.9**



Overfolded helix right ear. Make sure you measure the ear length (in cm).

Misshapen ear **Q17.3**

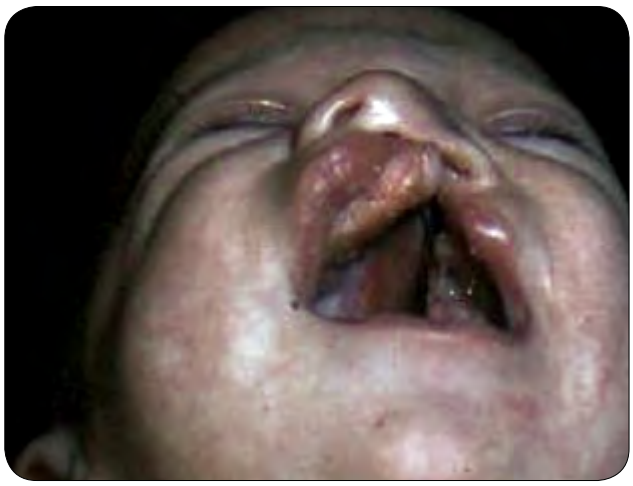


Wide gap between 1st and 2nd toe of left foot. Sandal gap.

Congenital deformity of foot **Q66.8**

*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

## CASE 12



Unilateral left clefting involving lip, nose, and palate.

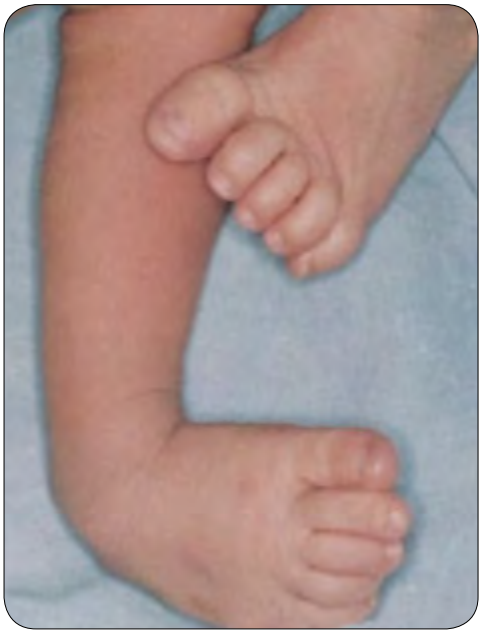
Total schisis of lip, deformed nose, cleft palate (hard and soft).

No pits of lower lip.

Cleft lip and palate unilateral **Q37.10**

*Source: Birth defects surveillance: atlas of selected congenital anomalies (5)*

## CASE 13



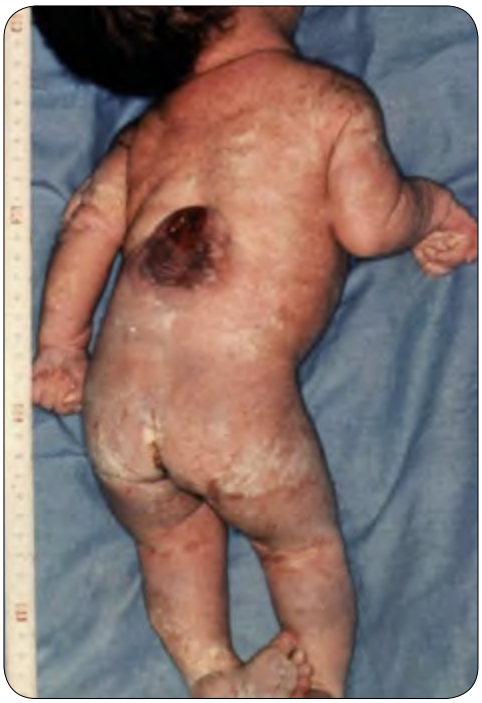
Rigid (non-reducible) deformity involving right foot.

Plantar flexion (talus pointing down), deviation of heel (calcaneus) and forefoot (inward), foot rests on outer side (upward rotation).

Talipes equinovarus **Q66.0**

*Source: Birth defects surveillance: atlas of selected congenital anomalies (5)*

## CASE 14



Midline spinal defect, lower thorax.

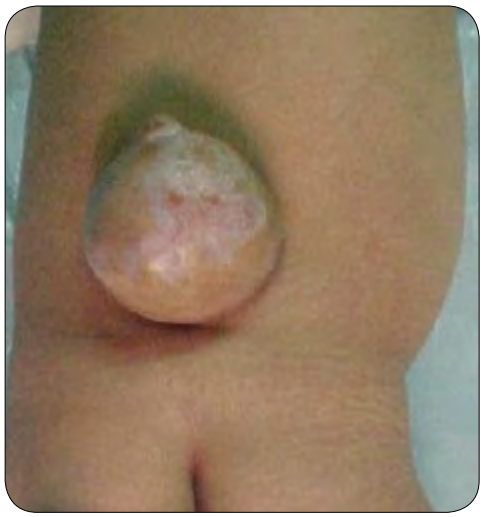
Rounded mass of degenerated spinal cord not covered by membrane. Possible hydrocephalus (head circumference 39 cm), bilateral talipes equinovarus.

Thoracic spina bifida with hydrocephalus
**Q 05.1**

Talipes equinovarus
**Q66.0**

*Source*: *Birth defects surveillance: atlas of selected congenital anomalies (5)*

## CASE 15



Midline spinal defect, lumbar region.

Rounded mass covered by partially atrophic skin.

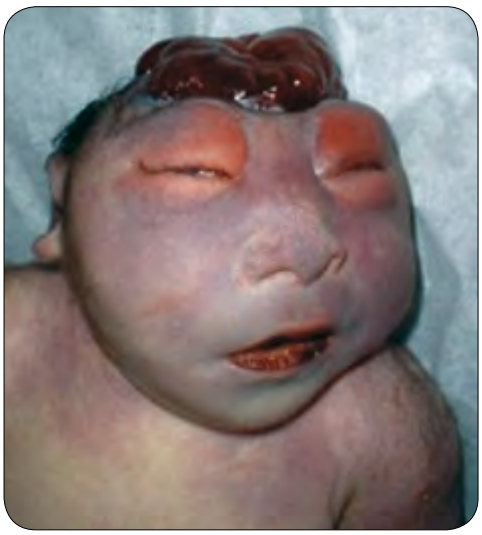
Hydrocephalus not mentioned (talipes equinovarus present/ absent, bilateral/unilateral).

Lumbar spina bifida without hydrocephalus
**Q05.7**

*Source*: Reproduced with permission of Estudio Colaborativo Latino Americano de Malformaciones Congénitas (ECLAMC)

*Note*: Spina bifida without mention of hydrocephalus does not have a specific code. We should assume that there is not hydrocephalus. The code should be changed if hydrocephalus is discovered later.

## CASE 16



Defect of cranial vault, partially degenerated brain tissue visible within the cranial bone defect. Severe swelling of palpebrae, large nose.

Mero-anencephaly (incomplete) **Q00.01**

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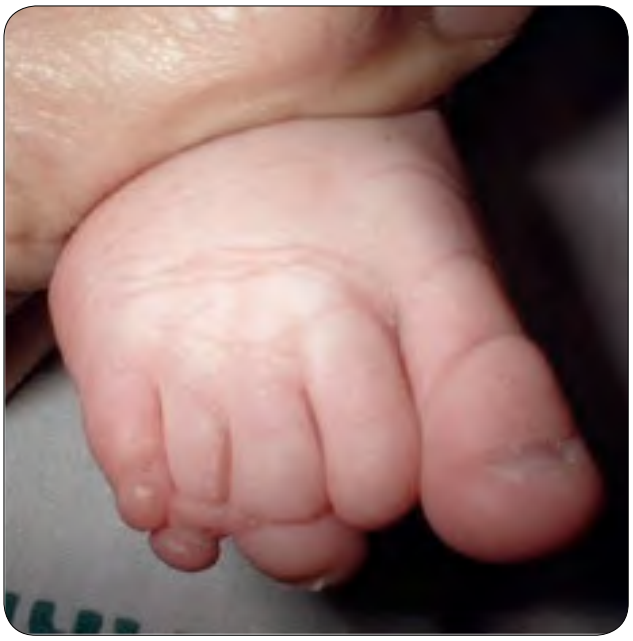
## CASE 17



Deficiency involving all digits of right foot, with absence of terminal parts of digits. Nubbins present.

Ring constriction and fibrotic band noted on 3rd, 4th and 5th digits.

(To be confirmed by X-rays)

Partial absence of toes (terminal transverse deficiency) **Q72.30**

Ring constriction **Q84.81**

Constriction band **Q79.80**

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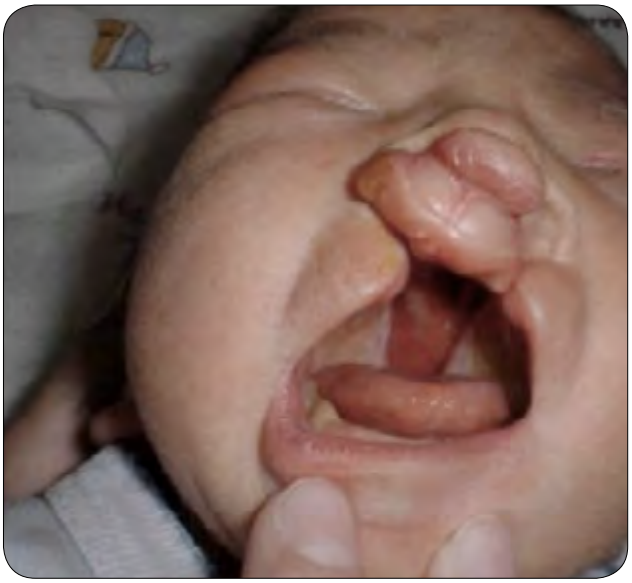
## CASE 18



Severe defect of upper lip.

Bilateral schisis of upper lip with midline part of the lip present.

No lower lip pits.

Cleft palate.

Bilateral cleft lip **Q36.0**

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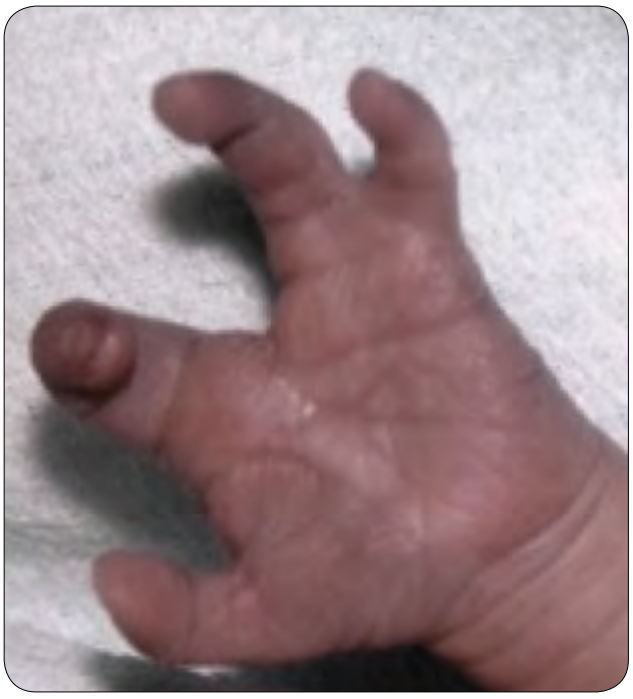
## CASE 19



Limb deficiency involving left hand.

Absent 3rd, 4th and 5th fingers, partially fused at the first phalanx level.

Absent third finger **Q71.30**

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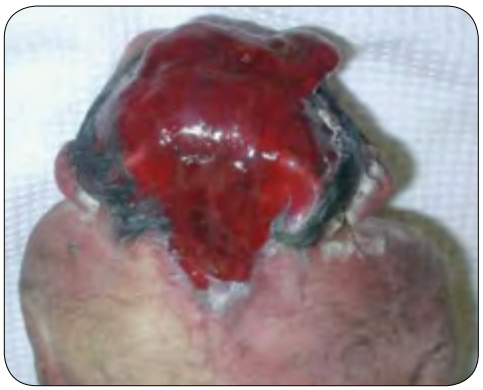
## CASE 20



Severe defect of cranial vault, parietal and occipital region and cervical spine: partially degenerated brain tissue outside the cranial and cervical spinal defect.

Craniorachischisis
**Q00.1**

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**For more information, please contact:**

**National Center on Birth Defects and Developmental Disabilities**
**Centers for Disease Control and Prevention**
1600 Clifton Rd, MS-E86, Atlanta, GA 30333, United States of America
Fax: +1 404 498 3550
http://www.cdc.gov/ncbddd/birthdefects/index.html

**International Clearinghouse for Birth Defects Surveillance and Research**
Via Carlo Mirabello 14, 00192 Roma, Italy
Fax +39 06 270 1904
Email: icbd@icbd.org
www.icbdsr.org

**Department of Nutrition for Health and Development**
World Health Organization
Avenue Appia 20, CH-1211 Geneva 27, Switzerland
Fax: +41 22 791 4156
Email: nutrition@who.int
www.who.int/nutrition

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