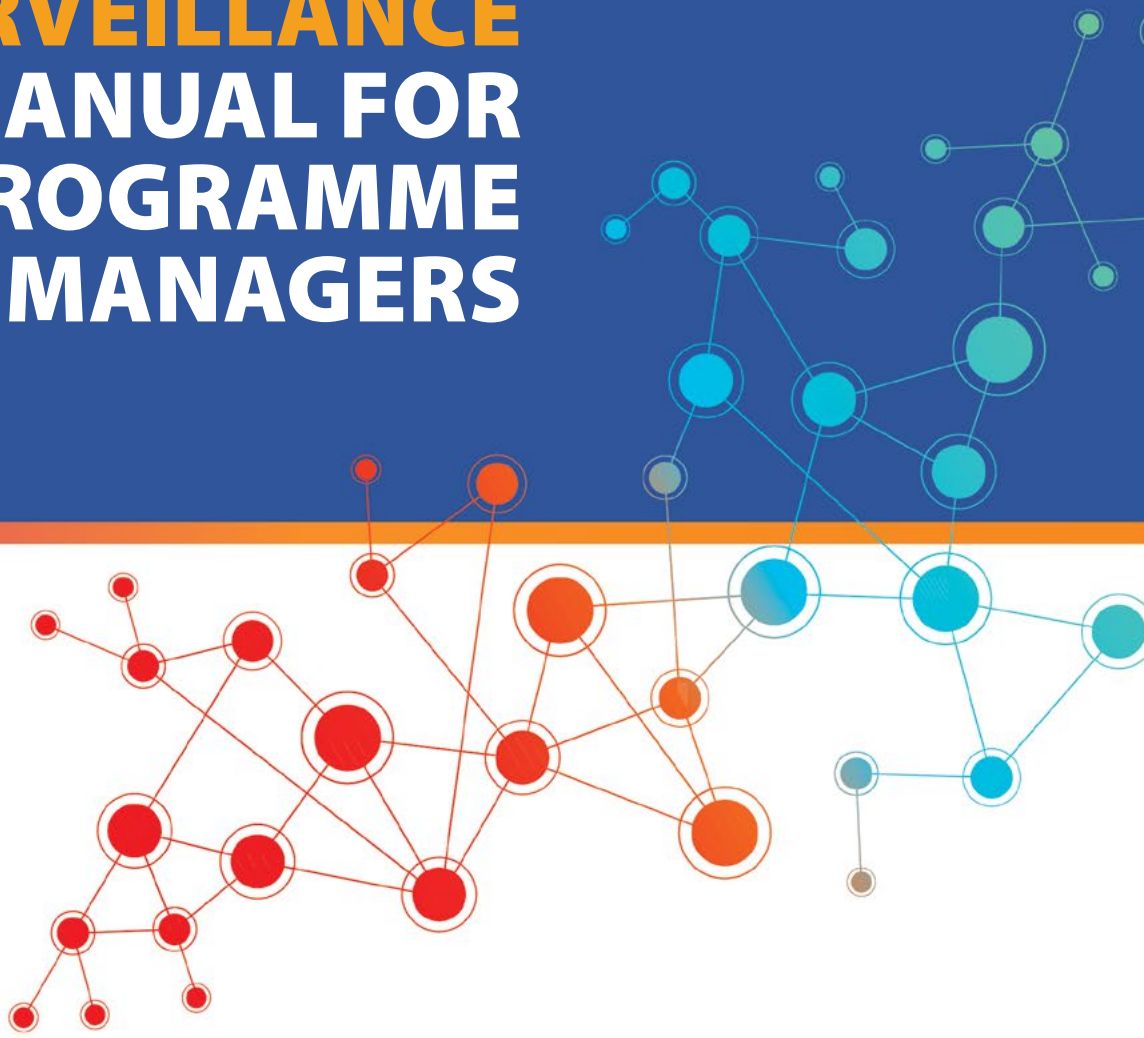
BIRTH DEFECTS SURVEILLANCE
A MANUAL FOR PROGRAMME MANAGERS



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BIRTH DEFECTS SURVEILLANCE

A MANUAL FOR PROGRAMME MANAGERS

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Abbreviations

CDC	United States Centers for Disease Control and Prevention
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
ICD-10	*International statistical classification of diseases and related health problems*, 10th revision
NBDPN	National Birth Defects Prevention Network
NCBDDD	National Center on Birth Defects and Developmental Disabilities
NGO	nongovernmental organization
NOS	not otherwise specified
RCPCH	Royal College of Paediatrics and Child Health
USA	United States of America
WHO	World Health Organization

Objectives of the manual

Congenital anomalies, also known as birth defects, are structural or functional abnormalities, including metabolic disorders, which are present from birth. Congenital anomalies are a diverse group of disorders of prenatal origin, which can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens or micronutrient malnutrition.

This manual is intended to serve as a tool for the development, implementation and ongoing improvement of a congenital anomalies surveillance programme, particularly for countries with limited resources. The focus of the manual is on population-based and hospital-based surveillance programmes. Some countries may not find it feasible to begin with the development of a population-based programme. Therefore, the manual covers the methodology needed for the development of both population-based and hospital-based surveillance programmes. Further, although many births in predominantly low- and middle-income countries occur outside of hospitals, some countries with limited resources may choose to start with a hospital-based surveillance programme and expand it later into one that is population based. Any country wishing to expand its current hospital-based programme into a population-based programme, or to begin the initial development of a population-based system, should find this manual helpful in reaching its goal.

This manual provides selected examples of congenital anomalies (see Appendix A). Typically, these anomalies are severe enough that they would probably be captured during the first few days following birth. Also, because of their severity and frequency, these selected conditions have significant public health impact, and for some there is a potential for primary prevention. Nevertheless, these are just suggestions, and countries can choose to monitor a subset of these conditions or add other congenital anomalies to meet their needs. In particular, this manual will help the reader to:

- describe the purpose and importance of public health surveillance of congenital anomalies;
- describe the use of logic models for planning and evaluation of a surveillance programme;
- understand how to present data to policy-makers;
- identify an initial list of congenital anomalies to consider for monitoring;
- describe the tools needed to ascertain and code identified cases;
- describe the processes for managing and analysing data;
- understand how to calculate the prevalence of congenital anomalies.

Surveillance of congenital anomalies should be ongoing and should involve a systematic review of birth outcomes to determine the presence of congenital anomalies. If countries have the capacity to identify risk factors associated with congenital anomalies such as maternal exposures (e.g., use of medications during the first trimester), a pregnancy registry or a case–control study can be implemented to allow for the collection of exposure data during pregnancy.

This manual is intended to facilitate the collection of essential information for the purpose of assessing the burden of congenital anomalies. It must be noted that the manual does not present specific information on how to collect risk factor information or how to manage a neonate born with congenital anomalies.

1. Surveillance of congenital anomalies

Introduction

Congenital anomalies are defined as abnormalities of body structure or function that are present at birth and are of prenatal origin *(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 manual, the term "congenital anomalies" will be used throughout.

According to the World Health Organization (WHO) in 2010, an estimated 270 000 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 purpose of congenital anomalies surveillance

Public health surveillance is defined as the 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, 4)*. Surveillance allows for the planning, implementation and evaluation of health strategies, and the integration of data into the decision-making process to help prevent adverse health conditions.

The ultimate purpose of a surveillance programme is to prevent adverse health conditions and their complications. Surveillance data, once collected, are critical for determination of whether a programme is having any effect, evaluation of whether new strategies are necessary, as well as detection of problem areas and intended populations that require more intensive intervention and follow-up.

The objectives of a surveillance programme for congenital anomalies are to:

- monitor trends in the prevalence of different types of congenital anomalies among a defined population;
- detect clusters of congenital anomalies (outbreaks);
- refer affected infants to appropriate services in a timely manner;

- disseminate findings and interpretations to appropriate partner organizations and government agencies, in a timely fashion;
- provide a basis for epidemiologic research (including risk factors) and prevention programmes;
- allow evaluation of prevention programmes.

Surveillance of congenital anomalies has been used for one or more of the following purposes:

- to measure the burden of congenital anomalies and identify high-risk populations;
- to identify disparities in prevalence and outcomes by factors such as race or ethnicity, maternal age, socioeconomic level or geographic region;
- to assess the effects of prenatal screening and diagnosis and other changes in diagnostic technologies on birth prevalence;
- to describe short-term and long-term outcomes of children with congenital anomalies, and to 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 to plan for needed services among the affected population;
- to guide the planning, implementation and evaluation of programmes to help prevent congenital anomalies *(4)* and to minimize complications and adverse outcomes among those affected by congenital anomalies;
- to assess any additional risk and the nature of adverse outcomes (including congenital anomalies) for fetuses and infants exposed to medicines during pregnancy, to improve management and to inform national and global public health policies *(5)*.

Types of surveillance programmes

Surveillance programmes can be population based or hospital/facility based and can use active or passive case ascertainment, or can be a hybrid of the two. More information about types of programmes and case ascertainment can be found in Chapter 3.

Population-based congenital anomalies surveillance programmes capture birth outcomes with congenital anomalies that occur among a population that is resident in a defined geographical area. Hospital- or facility-based congenital anomalies surveillance programmes capture birth outcomes with congenital anomalies that occur in selected facilities. Sentinel congenital anomalies surveillance programmes are generally set up in one or a few facilities/hospitals, to obtain rapid estimates of the occurrence of an adverse birth outcome.

Congenital anomalies: definitions

Congenital anomalies comprise a wide range of abnormalities of body structure or function that are present at birth and are of prenatal origin. For efficiency and practicality,

the focus is commonly on *major* structural anomalies. These are defined as structural changes that have significant medical, social or cosmetic consequences for the affected individual, and typically require medical intervention. Examples include cleft lip and spina bifida. Major structural anomalies are the conditions that account for most of the deaths, morbidity and disability related to congenital anomalies (see Box 1.1). In contrast, minor congenital anomalies, although more prevalent among the population, are structural changes that pose no significant health problem in the neonatal period and tend to have limited social or cosmetic consequences for the affected individual. Examples include single palmar crease and clinodactyly. Major anomalies are sometimes associated with minor anomalies, which might be objective (e.g. preauricular tags) or more subjective (e.g. low-set ears). Box 1.2 represents selected external minor congenital anomalies frequently captured by different surveillance systems, but only when associated with any of the major anomalies under surveillance. For a more detailed listing of minor anomalies, please refer to Appendix B.

Box 1.1. Selected external major congenital anomalies

Anencephaly

Cleft lip

Cleft palate

Cleft palate with cleft lip

Craniorachischisis

Encephalocele

Exomphalos/omphalocele

Gastroschisis

Hypospadias

Iniencephaly

Reduction defects of upper and lower limbs

Spina bifida

Talipes equinovarus/clubfoot

Box 1.2. Selected external minor congenital anomalies

- Absent nails
- Accessory tragus
- Anterior anus (ectopic anus)
- Auricular tag or pit
- Bifid uvula or cleft uvula
- Branchial tag or pit
- Camptodactyly
- Cup ear
- Cutis aplasia (if large, this is a major anomaly)
- Ear lobe crease
- Ear lobe notch
- Ear pit or tag
- Extra nipples (supernumerary nipples)
- Facial asymmetry
- Hydrocele
- Hypoplastic fingernails
- Hypoplastic toenails
- Iris coloboma
- Lop ear
- Micrognathia
- Natal teeth
- Overlapping digits
- Plagiocephaly
- Polydactyly type B tag, involves hand and foot
- Polydactyly type B, of fingers, postaxial
- Polydactyly type B, of toes, postaxial
- Preauricular appendage, tag or lobule
- Redundant neck folds
- Rocker-bottom feet
- Single crease, fifth finger
- Single transverse palmar crease
- Single umbilical artery
- Small penis (unless documented as micropenis)
- Syndactyly involving second and third toes
- Tongue-tie (ankyloglossia)
- Umbilical hernia
- Undescended testicle, bilateral
- Undescended testicle, unilateral
- Webbed neck (pterygium colli)

When establishing a new congenital anomalies surveillance programme, the initial anomalies that are included can be limited to structural anomalies that are readily identifiable and easily recognized on physical examination at birth or shortly after birth. The list may vary, depending on the capacity and resources of the health-care system and surveillance programme, but typically includes major external congenital anomalies. Examples include orofacial clefts, neural tube defects and limb deficiencies. In contrast, detecting the vast majority of internal structural anomalies (e.g. congenital heart defects, intestinal malrotation and unilateral kidney agenesis) requires imaging techniques or other specialized procedures that may not be available consistently. In some cases, internal anomalies have external manifestations that allow the observer to suspect a particular diagnosis, as is the case with the urethral obstruction sequence. Classification by developmental mechanism or clinical presentation can be important in surveillance, because the same congenital anomaly can have different etiologies. Also, the distinction may be important both clinically and in etiological studies. Please refer to Appendix C for more information about the causes of congenital anomalies and their classification according to developmental mechanism and clinical presentation.

2. Planning activities and tools

Many steps are required before conducting surveillance and collecting data. A logic model can be developed to help plan how a programme will be funded and staffed, identify activities and specify short- and- long-term outputs of the surveillance. The planning process would include identifying the existing rules and regulations pertaining to privacy and confidentiality issues surrounding data collection and reporting, and having a protocol in place that addresses handling of privacy and confidentiality.

Logic models

One approach that can be helpful when planning, implementing and evaluating a congenital anomalies surveillance programme is the use of logic models. A logic model is a graphic representation of how the surveillance programme will work. Logic models can identify what activities are needed, the order in which they would occur, and how the outcomes are going to be achieved. Most often, logic models will include the following components:

- *Resources*: what resources currently exist? What resources will be required to build or expand a surveillance programme?
- *Activities*: what activities are required for the surveillance programme to function? Keep in mind that there may be more than one intended audience.
- *Outputs*: what are the expected outputs that will result from the activities?
- *Expectations*: what are the short-term, intermediate, and long-term expectations (or outcomes) for each programme area?

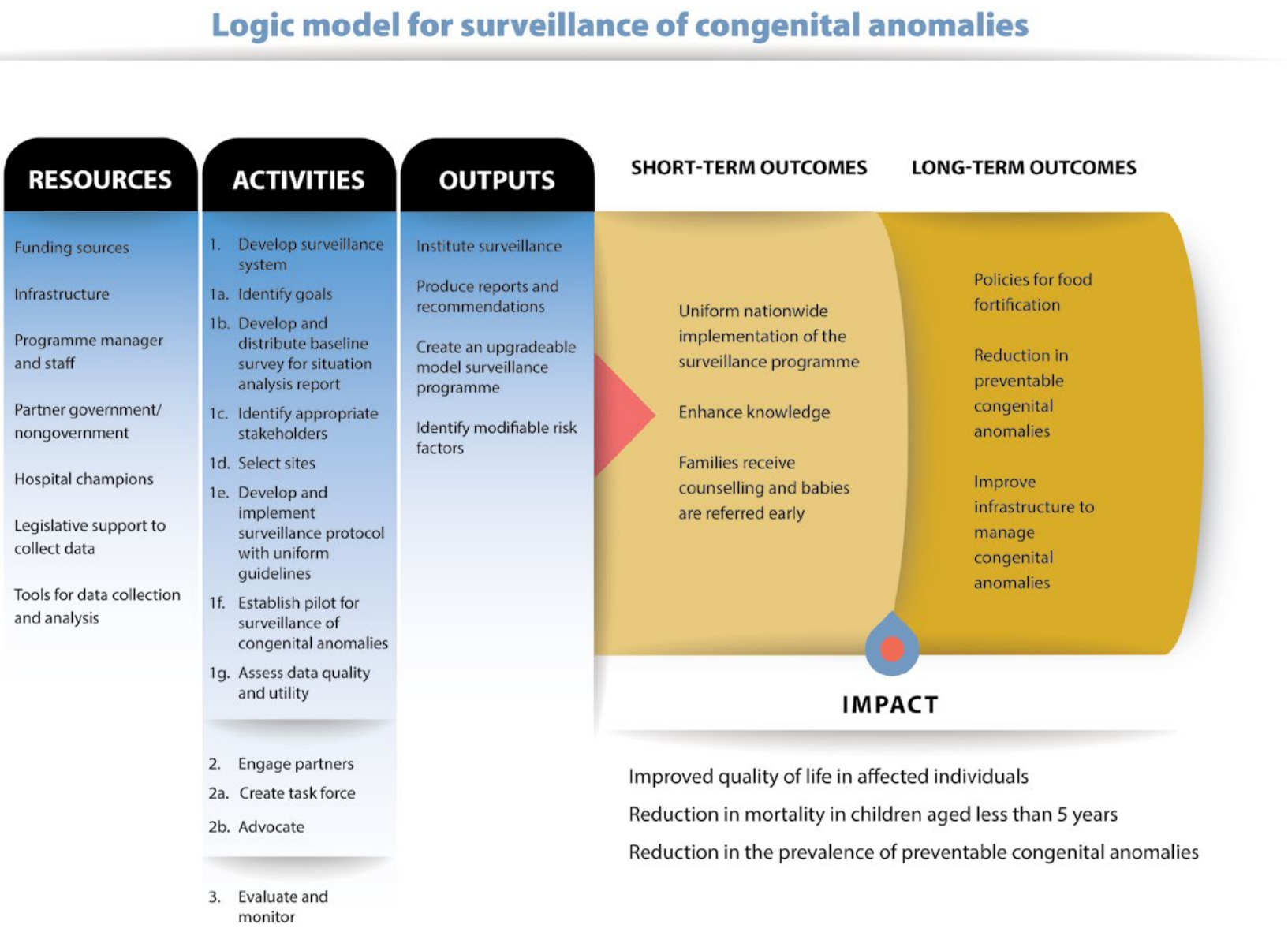
Logic models can have any shape (i.e. round, linear, columnar or a combination of these), and have any level of detail (i.e. simple, moderate or complex). It is probably best to begin by placing all relevant information into a table format (see Table 2.1) and then develop a logic model based on that information (see Fig. 2.1).

Table 2.1. Sample information to include in a logic model

Resources	Activities	Outputs	Short- and long-term outcomes	Impact
Funding sources	1. Develop surveillance system	Institute surveillance system	Uniform nationwide implementation of the surveillance programme	Improved quality of life for affected individuals
Infrastructure	1a. Identify goals	Produce reports and recommendations	Enhance knowledge	Reduction in mortality in children aged less than 5 years
Programme manager and staff	1b. Develop and distribute baseline survey for situation analysis report	Create an upgradeable model surveillance programme	Develop policies	Reduction in the prevalence of preventable congenital anomalies
Partner government/ nongovernment	1c. Identify appropriate stakeholders	Identify modifiable risk factors	Improve need-based infrastructure to manage congenital anomalies	
Hospital champions	1d. Select sites			
Legislative support to collect data	1e. Develop and implement surveillance protocol with uniform guidelines			
Tools for data collection and analysis	1f. Establish pilot for surveillance of congenital anomalies			
	1g. Assess data quality and utility			
	2. Engage partners			
	2a. Create task force			
	2b. Advocate			
	3. Evaluate and monitor			

Adapted from: India team, Regional Workshop on Birth Defects Surveillance; Colombo, Sri Lanka, April 2012.

Fig. 2.1. Logic model for surveillance of congenital anomalies



Adapted from: India team, Regional Workshop on Birth Defects Surveillance; Colombo, Sri Lanka, April 2012.

Creating a logic model has benefits. It can help define goals and objectives, as well as foster agreements among stakeholders about roles and responsibilities related to different activities. It can also help to identify gaps or barriers and build connections between activities and results. Please refer to Appendix D for another example of a logic model.

Partners and funding

The engagement of a wide variety of partners is essential for the development, implementation and maintenance of a surveillance programme. Key partners, funders and stakeholders can be identified and involved during the initial planning stages of the development of a surveillance programme. This can help to ensure that a surveillance programme is implemented and sustained for the long term. Determining what roles and responsibilities are needed can also help identify what kind of partners would be invited to participate in the initiative.

Examples of possible areas for partner engagement are the development of goals and objectives for the surveillance programme, the development of policy measures and shepherding of measures through appropriate channels, and the identification of funding support for training hospital personnel.

The following are examples of potential partners for consideration when developing or implementing a surveillance programme:

- ministry of health and other government agencies
- environment ministries, toxicology departments, poison control centers and children's environmental health units
- organizations and agencies that regulate hospitals, birthing centres and labour and delivery units
- medical and nursing professional associations
- health-care providers
- insurance companies
- universities
- community-based organizations that have an interest in congenital anomalies
- advocacy and community groups
- parent and family support groups for those with children affected by congenital anomalies
- privacy protection and legal ethics offices
- security, data-access and information-management offices
- researchers
- policy professionals
- media
- religious leaders.

Appendix E can help in the development of a list of partners and determination of how partners can best participate and collaborate with the surveillance programme.

Legislation

Mandatory reporting

How a country defines mandatory reporting, who creates the mandate, and whether or not it is enforced will vary by country. In the United States of America (USA), mandatory reporting for a surveillance programme means that hospital staff are required to keep a log and report all cases of congenital anomalies and fetal deaths to the surveillance programme within a determined time frame, and in a standardized format. Structured and reliable data provide a justification for countries to invest in sustainable programmes for prevention of congenital anomalies, and can help in the development of public policy for adequate distribution of resources for babies born with a congenital anomaly. Furthermore, regular reporting in a standardized format can greatly facilitate analyses of prevalence and trends for the congenital anomalies being monitored.

Voluntary reporting

As countries vary in how they define mandatory reporting, so too they can vary in how they define voluntary reporting. Generally, in voluntary reporting, hospital/facility staff members are encouraged by the ministry of health of the country to keep a log and report all cases of congenital anomalies and fetal deaths to the surveillance programme; however, hospitals can choose whether or not to comply. The ministry of health can request that hospitals report cases in a uniform manner, but each hospital can decide whether, how and when they will report the information.

Privacy and confidentiality issues

Each country has different laws, regulations and protocols for how to protect patient data. It is important to understand the laws or regulations related to the collection, use, dissemination and protection of personal information. Laws can be reviewed, and policies for collection, management and use of data can be implemented prior to initiating a congenital anomalies surveillance programme. Ideally, the authority to operate a surveillance programme will be made explicit by law and its regulations. It is important to have regulations in place to protect the public, as well as the providers and surveillance staff who report the information. During the preparation of the protocol for a congenital anomalies surveillance programme, it is important to specify the purpose of surveillance, the types of data that will be collected and why these are necessary, how they will be collected (paper based, electronically or both), who will have access to the data, how the data will be used, where the data will be stored and secured, and for how long the law requires the data to be archived. Also, it is important to educate hospital personnel on the purpose of the surveillance programme, and how patient privacy and confidentiality will be protected. Lastly, it is essential that surveillance programme personnel sign confidentiality agreements prior to beginning work in the programme.

Privacy

In the matter of personal health information, privacy is an individual's right to control the acquisition, use and disclosure of his or her identifiable health information. To avoid using personal information, and to protect a family's privacy, each fetus or neonate with a congenital anomaly is assigned a unique identifier.

Confidentiality

In terms of patient data, confidentiality refers to an individual's right to have his or her personal, identifiable medical information kept secure. Confidential information must be kept secure according to the regulations in each country, and out of sight of unauthorized people. It is important to note that confidential information can be made available only to specific health-care providers and to specific personnel overseeing the surveillance programme. When sharing the data with others in the country (e.g. hospital managers and policy-makers), all reports are aggregated and do not have any potential patient identifiers (e.g. name or address). If possible, confidentiality agreements are signed on a regular basis, to ensure that personnel are reminded of the importance of this practice.

Security

When dealing with patient information, security refers to the technological and administrative safeguards and practices designed to protect data systems against unwarranted disclosure, modification or destruction. All individuals have the right to have personal, identifiable medical information kept secure. Security, in this context, refers specifically to how personal information is stored, who has access to this information, and with whom this information can be shared.

Informed consent

The processes and requirements related to informed consent vary by country. Because of the public health importance of evaluating and tracking the occurrence of congenital anomalies, most countries do not require informed consent prior to reporting a congenital anomaly diagnosis to a surveillance programme. If the country has a law that requires a consent form, then information may be shared only once this form has been signed. If the law does not require a consent form, parents can be told orally that the non-identifiable information will be shared.

Data dissemination

One important aspect of the implementation process for a congenital anomalies surveillance programme, other than the collection and analysis of the surveillance data, is to plan in advance the way the information generated will be disseminated. Part of this advance planning involves identification of the processes by which documents (e.g. statistical reports and annual reports) are tailored to the different potential audiences who will be receiving information about the surveillance findings. Potential audiences can include partners, stakeholders, health-care providers and the public. Use of the surveillance data includes:

- identifying trends of congenital anomalies
- planning, implementing and evaluating evidence-informed interventions
- motivating action in the community
- informing policy-makers and government officials
- informing clinical and public health practitioners, nongovernmental organizations (NGOs) and the public
- identifying and referring children with special needs to applicable services.

With this information, strategies for improving health outcomes among an intended population can be developed, infrastructure barriers can be identified and remediated, and efforts can be made to gain the support of local and regional partners.

The primary users of surveillance information are usually public health professionals and health-care providers. The information directed primarily to those individuals includes the analysis and interpretation of surveillance results, along with recommendations that stem from the surveillance data. It is important that participating providers and institutions be informed of the situation in their participating facilities or hospitals, as well as in areas of the health system using the information to assess progress in this type of programme. If possible, a committee can be established, with the participation of technical experts and stakeholders, to facilitate discussions of issues related to security and confidentiality, statistical analyses, presentation and sharing of data, and evaluation of the feasibility and merit of collaborative projects. If data are analysed and presented effectively, decision-makers at all levels can visualize and understand better the implications of the information.

A protocol for communication and dissemination of information can be developed to address the needs of a variety of audiences. This protocol can address questions such as:

- What message is most relevant for a particular audience?
- Is there a timeline for when data will be disseminated?
- What will determine whether the information that was disseminated was useful and whether the objectives were reached?
- What format and avenue needs to be used to reach this audience?

There are different avenues for data dissemination: paper based or electronic, or a combination of the two. By using technology, news releases, letters, brochures, reports and scientific articles can be made available in a web-page format or can be disseminated using social media outlets. Some examples of ways that data are disseminated can be found in references *(6–11)*.

Communicating with parents

It is important to remember that abstractors, those individuals who will be extracting information from hospital logs or medical records for the identification and classification of congenital anomalies, do not give information to parents about a diagnosis or services. This is commonly done by a health-care provider. This section is included in this manual simply as a reminder to all programme staff that every identified "case" means that family members now have to cope with the death or disability of a child. Certified health-care providers – those doctors, nurses and midwives who have direct patient care responsibilities and are working as part of the surveillance programme – will benefit from training on how to communicate sensitive information appropriately. Grieving parents may not fully comprehend a complicated diagnosis; therefore, it could be helpful to provide parents with written information about the diagnosis, along with available organizations, support groups, bereavement services and genetic counselling services. Please refer to Appendix F for a listing of how health-care providers can communicate to parents information about a diagnosis of a congenital anomaly.

3. Approaches to surveillance

This chapter describes some of the different methodological approaches used in the surveillance of congenital anomalies.

Population coverage

Once the purposes of surveillance have been established, the next steps are to define the population under surveillance and identify the area of coverage. The programme may be population based or facility or hospital based (see Fig. 3.1). The coverage (geographical area) for a population-based surveillance programme can be a city, a region or an entire country. A population-based programme has a defined source population (typically defined by maternal residence), and all identified congenital anomalies occurring within that source population are ascertained and included, regardless of delivery site.

In contrast, the source population for a hospital-based programme typically cannot be defined accurately. The coverage for a hospital-based surveillance programme is usually at least a few hospitals or clinics in one geographic region. However, there generally are no distinct catchment areas for specific hospitals and, thus, no defined denominator of the entire source population from which all cases are ascertained. Hospital-based programmes work best when they capture most of the population of interest in a geographic region.

Owing to lack of resources or other restrictions, some countries may not find it feasible to start a surveillance programme as a population-based programme and, therefore, will choose to begin with the development of a facility-based or hospital-based programme. However, it is critical to understand the limitations of a hospital-based programme, and interpret any findings from such a system within those limitations.

Fig. 3.1. Population coverage in surveillance programmes

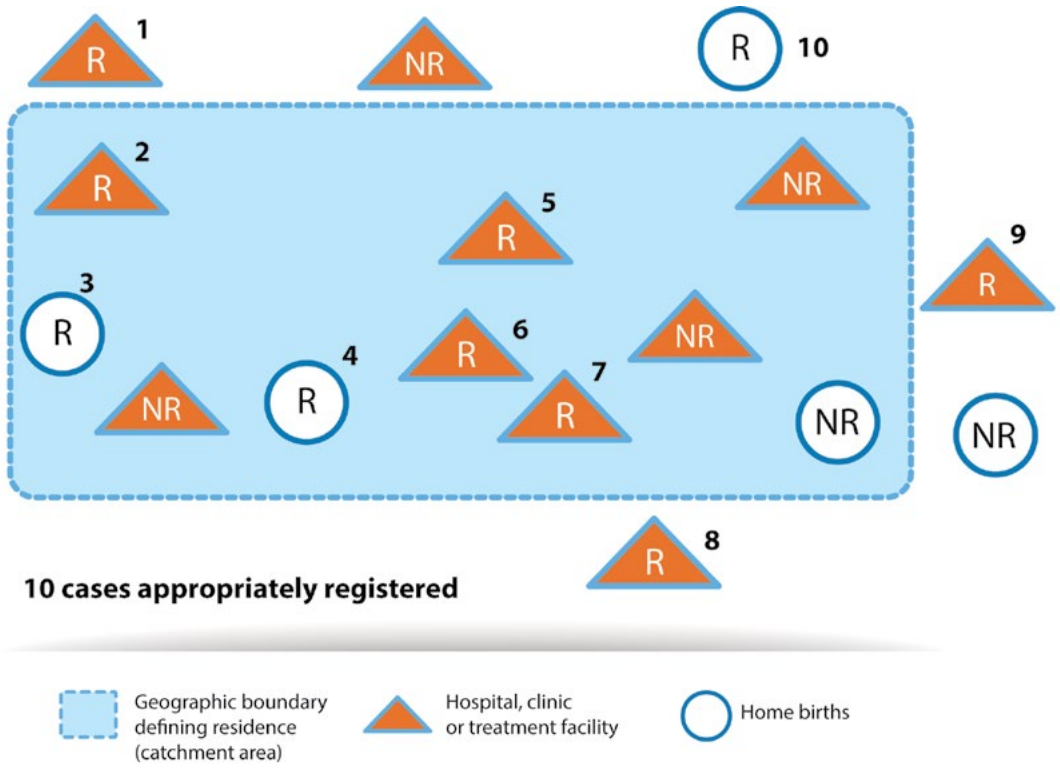
Population-based surveillance programmes

Population-based congenital anomalies surveillance programmes collect data from an entire source population (fetuses or neonates with a congenital anomaly and the total number of births) born to resident mothers living in a defined catchment area (geographical area), within a defined time period.

Thus, the denominator used to calculate prevalence in a population-based programme consists of births to resident mothers. The corresponding numerator consists of fetuses or neonates with congenital anomalies born to resident mothers. Because of this definition, all births are collected in a population-based programme, meaning not only births occurring in hospitals or maternity hospitals but also those occurring at home.

Most congenital anomalies surveillance programmes use the mother's primary residence at the time of delivery or pregnancy termination to define the source population among which the cases occur. For example, residence can be defined as the mother's primary address during the 3 months prior to pregnancy and the first trimester of pregnancy. However, the crucial issue is that the definition of resident status used for cases (the numerator) must be the same as that used for all births (the denominator). Fig. 3.2 illustrates an example of a population-based programme. All fetuses or neonates identified with a congenital anomaly born to mothers residing *within* the catchment area (dashed area) are included in the programme (2 to 7 in Fig. 3.2). Similarly, a fetus or neonate with a congenital anomaly who is born outside of the defined catchment area (including one who is born at home while the mother is visiting a family member living outside of the catchment area, for example) would still be included if the mother is herself a resident of the catchment area (1, 8, 9 and 10 in Fig. 3.2). Fetuses or neonates identified with congenital anomalies and born to *non-resident mothers* are not included. Data sources include all health facilities within the catchment area where births occur, vital records (e.g. birth and death registries), referral treatment centres for individuals with congenital anomalies (up to the defined age period for inclusion), administrative databases, and any health-care facility that identifies a fetus or neonate with a congenital anomaly.

Fig. 3.2. Catchment area for a population-based surveillance programme



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.

The steps for calculating prevalence, and information on how to define the denominator, are described later on in this chapter.

Hospital-based surveillance programmes

Hospital-based congenital anomalies surveillance programmes capture all pregnancy outcomes with congenital anomalies that occur in selected hospitals in a defined geographic area (e.g. a state, province or county).

The denominator used to estimate prevalence in a hospital-based programme consists of births occurring in the participating hospitals. The numerator (cases) typically consists of affected live births and stillbirths occurring in these hospitals. Fetuses or neonates with congenital anomalies who are delivered at home are not included, even if they are identified and captured in participating hospitals (because they are not part of the denominator).

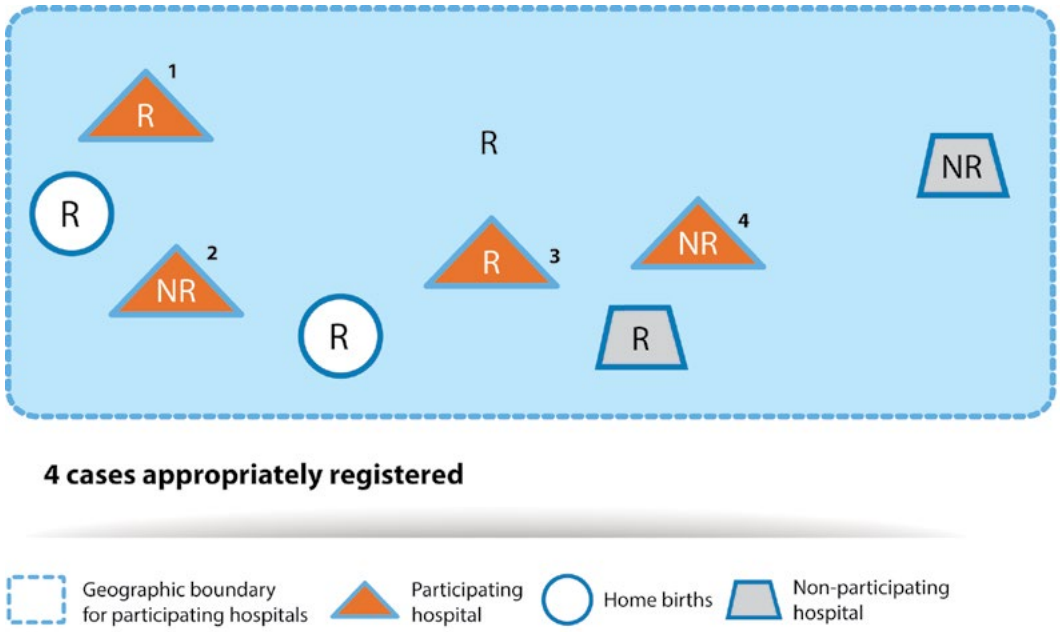
Because the inclusion in a hospital-based programme depends on where the birth occurred rather than on the residence at birth, the source population of cases is difficult to establish. This becomes an issue in the surveillance of congenital anomalies when referral patterns skew the likelihood that an affected fetus or neonate is delivered at a hospital in the system. Thus, a major concern in hospital-based programmes is referral bias of cases, that is, the selective delivery of affected pregnancies in hospitals participating in the hospital-based programme. This referral bias can also vary over time, either because referral patterns change or because hospitals are added or removed from the surveillance programme. This, in turn, adds to the problem of using these hospital-based data longitudinally for monitoring.

Such hospital-based programmes typically collect data on live births and stillbirths. Because neonates are discharged from maternity hospitals within days following birth, hospital-based programmes typically capture only those congenital anomalies that are evident during the hospital stay, unless those readmitted to the hospital for surgery or other procedures are captured. Note that fetuses or neonates diagnosed after delivery in a hospital participating in a hospital-based programme are not included for the purposes of surveillance, *unless* they were also delivered at a participating site in a hospital-based programme.

For illustration, Fig. 3.3 presents an example of participating and non-participating hospitals in a hospital-based surveillance programme. All fetuses or neonates with congenital anomalies born to mothers in participating hospitals, regardless of maternal residency, are included in the programme (1 to 4 in Fig. 3.3). Fetuses or neonates with congenital anomalies born to resident mothers *but born outside of a participating hospital or at home* are not included. Fetuses or neonates with congenital anomalies born to *non-resident mothers* are included if they are born in a participating hospital.

Ascertainment of fetuses or neonates identified with congenital anomalies in participating hospitals can vary. While some are primary hospitals, others may be specialized centres for certain conditions, or for prenatal diagnosis and care, and serve as referral hospitals for patients outside the catchment area. As discussed, such hospitals would disproportionately serve fetuses or neonates with congenital anomalies, thus introducing bias in the calculation of their birth prevalence.

Fig. 3.3. Catchment area for a hospital-based surveillance programme



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.

The magnitude of bias may change over time, with fluctuations in referral patterns and the proportion of births occurring outside the hospital setting. This could lead to changes in rates that have nothing to do with the underlying prevalence, but are the result of referral patterns. Also, the bias will depend on how many hospitals or facilities are included – all, half or only a small percentage.

Estimates of birth outcomes with congenital anomalies in hospital-based surveillance programmes represent only those births at reporting hospitals in which data are collected. The prevalence estimates could, therefore, be biased, particularly if the hospital births are a minority of all births, if they receive a high proportion of difficult or complicated pregnancies, and/or if they are not representative of the population of interest. Bias limits the representativeness and usefulness of the data for surveillance. However, if nearly all hospitals in a country participate in the surveillance programme and nearly all births occur in hospitals, the surveillance programme may approximate a population-based surveillance programme.

Finally, a subset of facility-based or hospital-based surveillance known as *sentinel surveillance* is generally set up in key sites, to obtain rapid estimates of the occurrence of a birth outcome. Because congenital anomalies are relatively rare events, sentinel surveillance programmes may not be very effective for capturing congenital anomalies.

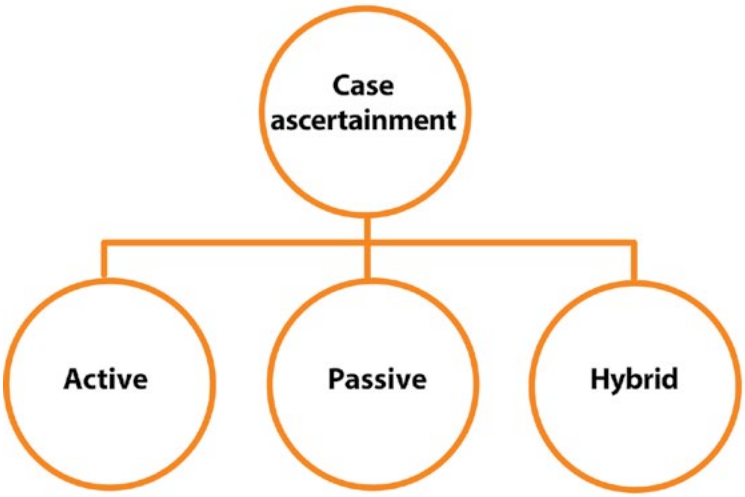
Although population-based and hospital-based surveillance programmes have clear differences, there are some characteristics that are common to both. These include:

- participating clinicians can be motivated "champions" – leaders who are committed to the programme;
- data collected are useful for documenting that a problem may exist;
- data collected are useful for alerting health and government officials to the need for investing further in possible causes and prevention strategies;
- affected children can be refered to services;
- high-quality case data, including data on potential risk factors, can be generated.

Case ascertainment

Once the type of population coverage has been decided, the next step is to determine how cases will be ascertained. This can be done using active, passive or hybrid approaches (see Fig. 3.4).

Fig. 3.4. Case ascertainment



Active case ascertainment

With active case ascertainment, the surveillance personnel typically are hired and trained to conduct data abstraction (abstractors). Abstractors regularly visit, or have electronic access to, participating institutions (e.g. hospitals and clinics), and actively review multiple data sources (e.g. logbooks and medical, discharge and deaths records), to identify cases. Therefore, visiting all areas of the hospital where a potential fetus or neonate with a congenital anomaly can be identified could be important (e.g. a labour and delivery unit or a neonatal intensive care unit). For those fetuses or neonates

identified in the logbooks as having a congenital anomaly, abstractors usually request maternal and infant medical records to abstract relevant information onto a reporting form (see Appendix G). It is noted that for this process to work well, medical records need to contain relevant information in a format that can be identified and abstracted easily by the abstractors, who usually have limited medical background.

Although this type of case ascertainment requires considerable resources and personnel, active case ascertainment tends to improve case detection and case reporting, and improves data quality because more extensive clinical details are collected.

To improve case ascertainment, the surveillance programme can link administrative databases (e.g. vital records, discharge data and insurance databases) with the surveillance programme database, to identify potential additional cases that were not ascertained from the more common sources. The identified cases will require verification by personnel going to the maternity hospitals and reviewing the medical records.

Passive case ascertainment

With passive case ascertainment, hospital personnel who identify a fetus or neonate with a congenital anomaly or anomalies report this information directly to the surveillance registry. Also, cases may be ascertained by linking administrative databases (e.g. vital records, discharge data and insurance databases) to the surveillance programme database. With passive case ascertainment, the information that is reported to the surveillance registry is typically not verified by direct abstraction of the medical record.

This type of case ascertainment is less expensive because fewer resources and personnel are required. However, the burden of reporting falls on hospitals or clinics, which may require time and effort of hospital staff who are already busy. This could result in a less than optimal reporting rate, less complete documentation or less timely reporting, or a combination thereof. It also usually yields less complete detail on each case and underestimates the number of congenital anomalies that occur. In addition, because reported information is not validated, it could also overestimate certain congenital anomalies.

Hybrid case ascertainment

Hybrid case ascertainment refers to a combination of passive case ascertainment of most types of congenital anomalies, with active case ascertainment of specific congenital anomalies, or for a percentage of all reported congenital anomalies as a quality control tool. For example, a surveillance programme can conduct active ascertainment of neural tube defects to gather more detailed case information in a more timely manner, but carry out passive ascertainment of all other congenital anomalies under surveillance. Similarly, a programme can use passive reporting with active follow-up verification of certain congenital anomalies.

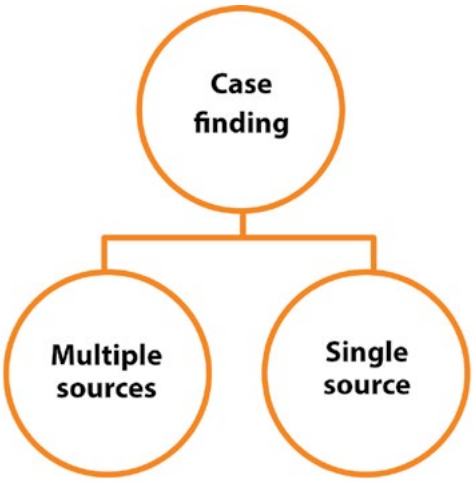
Regardless of the method selected for case ascertainment (active or passive), each participating hospital can identify a "champion" who is committed to the programme. This could help to ensure more complete participation of the different hospital units and services participating in the surveillance programme. Also, the role of this leader could

be to train other personnel (such as doctors, nurses and technicians) on how to identify cases, record the information and oversee the information flow, so as to maintain an ongoing and active quality control on the quantity and completeness of information. The role of a leader, or champion, can be important to a programme's success.

Case finding

Congenital anomalies surveillance programmes can decide the sources from which cases will be identified (see Fig. 3.5).

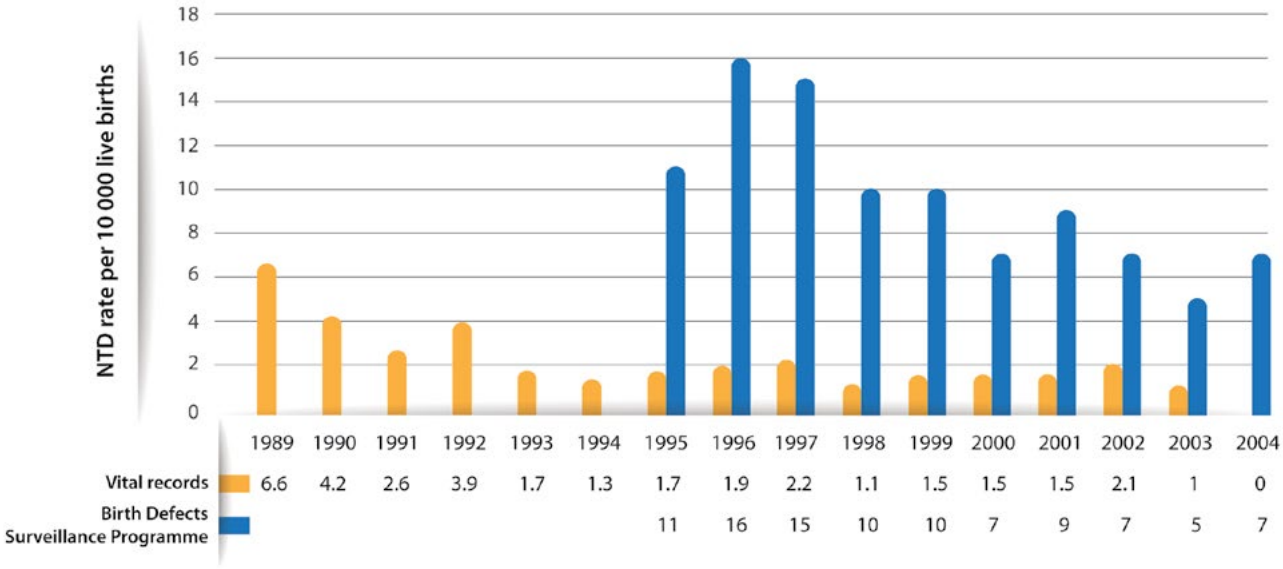
Fig. 3.5. Case finding



Data sources

Using multiple sources may improve the completeness of case ascertainment by identifying cases that are not available from only one individual source. Additionally, it may improve the quality of the data, as having multiple sources may increase the amount and level of information available for a given case. For example, a diagnosis may not be possible in the delivery unit but may be established by specialists in the paediatric unit and further confirmed by laboratory tests. While the use of multiple data sources is more time consuming and delays the process of gathering information, it can improve overall case ascertainment and data quality. Using a single source for case ascertainment does not allow for ascertainment of the majority of fetuses or neonates with a congenital anomaly in most settings. For example, Fig. 3.6 depicts a marked under-ascertainment of neural tube defects in Puerto Rico when cases were ascertained from vital records alone, compared with case ascertainment from multiple sources (e.g. logbooks in the delivery room, neonatal care units, paediatric units) used by the Birth Defects Surveillance Programme.

Fig. 3.6. Case ascertainment using vital records versus multiple sources; Birth Defects Surveillance Programme Puerto Rico



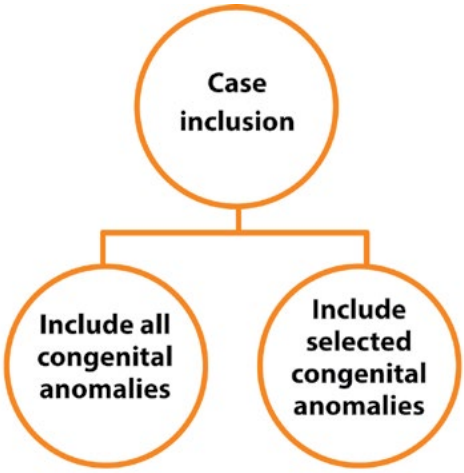
NTD: neural tube defects.

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

Case inclusion

Each surveillance programme decides which congenital anomalies to include. A programme may choose to include all major congenital anomalies, while another programme may decide to include selected congenital anomalies, according to the needs of the country (see Fig. 3.7). As discussed in Chapter 4, one consideration is to start with a small number of easily recognizable congenital anomalies and then expand to include additional anomalies, as a programme gains experience and resources.

Fig. 3.7. Case inclusion



Description formats for congenital anomalies

As Fig. 3.9 illustrates, on the abstraction form, information about congenital anomalies is ideally captured using either a verbatim description, or a checkbox, although sometimes both may be used. From a quality standpoint, checkboxes alone are typically insufficient to achieve high data quality, both in initial data abstraction and in case review. However, if a country has the resources to collect data electronically, a checkbox could be useful as a first step, if there is a drop-down menu with more options to categorize the congenital anomaly. Nevertheless, having a field for verbatim descriptions allows details to be collected and the additional information may help clarify the diagnosis and classification, be used to plan the child's health-care management, and help assess the accuracy and completeness of the information and coding during case review and ongoing quality assessment.

Fig. 3.8. Description formats

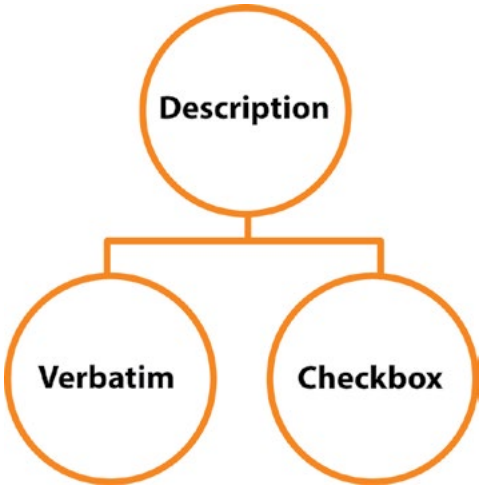


Fig. 3.9. Examples of verbatim description and checkbox formats for documenting congenital anomalies

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

When coding the example case in Fig. 3.9, if the information available is limited to one of the checked boxes, the *International statistical classification of diseases and related health problems*, 10th revision (ICD-10) *(12)* code will be Q36.99 (cleft lip). However, if the verbatim description is available, the code will be more specific: Q36.9 (cleft lip, unilateral).

Age of inclusion

Countries that have congenital anomalies surveillance programmes have different criteria for age of inclusion. Some include information about fetuses or neonates with a congenital anomaly ascertained only during the early neonatal period (see Table 3.1), while others include those ascertained up to 1 year of age and beyond (see Fig. 3.10).

Fig. 3.10. Age of inclusion in a congenital anomalies surveillance programme

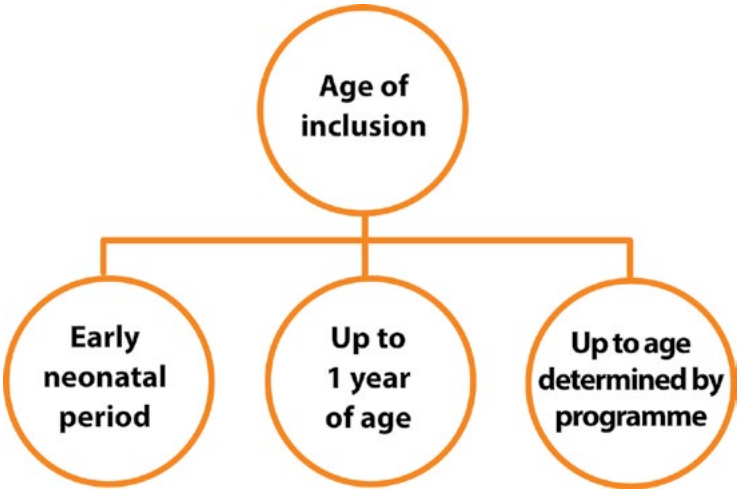
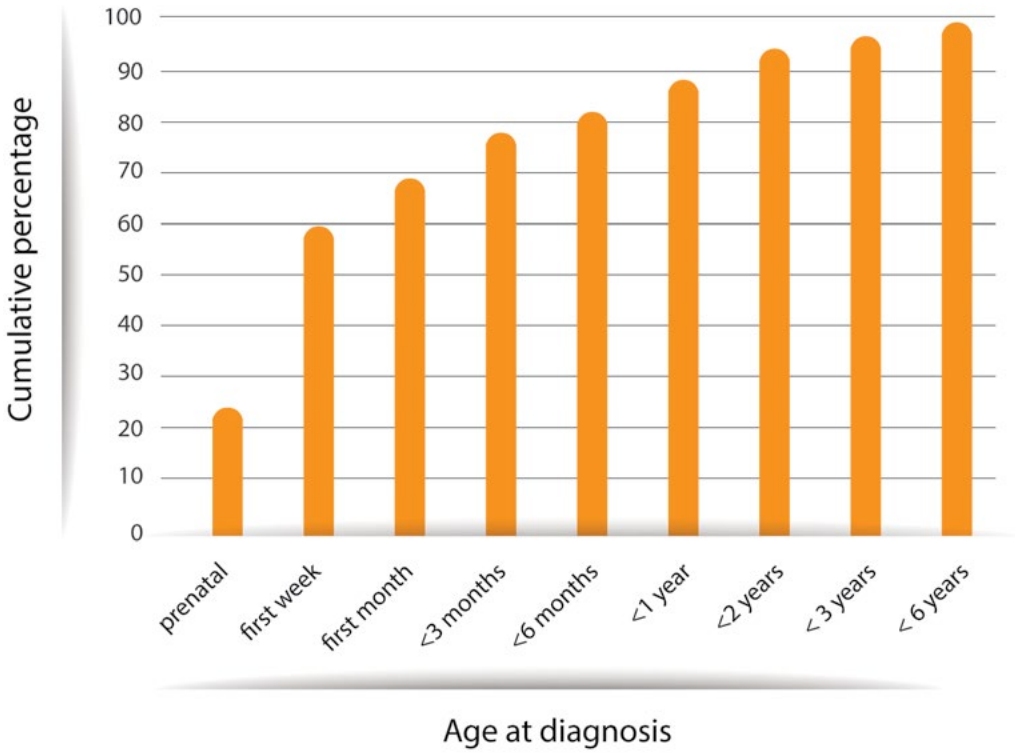


Table 3.1. Periods of infancy

Time period	Days
Neonatal	0–27
Early neonatal	0–6
Late neonatal	7–27
Post neonatal	28–364

The Western Australian Birth Defects Registry includes cases diagnosed up to 6 years of age. Fig. 3.11, derived from reference *(13)*, indicates the cumulative percentage of cases with major external and internal congenital anomalies by age at first diagnosis. The authors examined the age at diagnosis for all congenital anomalies notified to the Birth Defects Registry from 2000 to 2001. Nearly 60% of all major congenital anomalies were diagnosed during the first week of life, nearly 70% by the first month, nearly 90% by the first year, and nearly 100% by the sixth year.

Fig. 3.11. 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)*.

Age at diagnosis is a critical component of the case definition. Typically, the higher the cut-off age, the greater the reported frequency of conditions, especially for conditions involving internal organs that may not be evident at birth. For example, whereas external anomalies such as neural tube defects and gastroschisis are evident at birth, some internal anomalies such as heart defects may not be identified until days, or even weeks or months after delivery. In addition, certain anomalies may require postnatal confirmation.

Inclusion of pregnancy outcomes

Surveillance programmes aim to ascertain congenital anomalies among all pregnancy outcomes – live births, fetal deaths and terminations of pregnancy – if possible (see Fig. 3.12). Some countries have the ability and resources to ascertain all or most of these outcomes when they occur relatively late in pregnancy, but it is extremely difficult to systematically ascertain those occurring prior to 28 weeks' gestation and, in particular, those in which the pregnancy is terminated. For these reasons, if prenatal ascertainment of congenital anomalies is not an available option in a given catchment area, it would be more feasible initially to limit the ascertainment to live births and to fetal deaths occurring at 28 weeks' gestation or older, or, alternatively, with a birth weight of at least 1000 g (if gestational age is

not available). However, in many countries and settings, ascertainment among 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' gestation (e.g. anencephaly). If a country has the capacity to ascertain cases prior to 28 weeks' gestation, doing so can help provide a more accurate estimate of the prevalence of a condition such as anencephaly. Programmes interested in more detailed information on inclusion of prenatal diagnosis in congenital anomaly surveillance can find some useful and practical suggestions and tips in the guidelines developed by the National Birth Defects Prevention Network (NBDPN) in the USA *(14)*.

Fig. 3.12. Inclusion of pregnancy outcomes

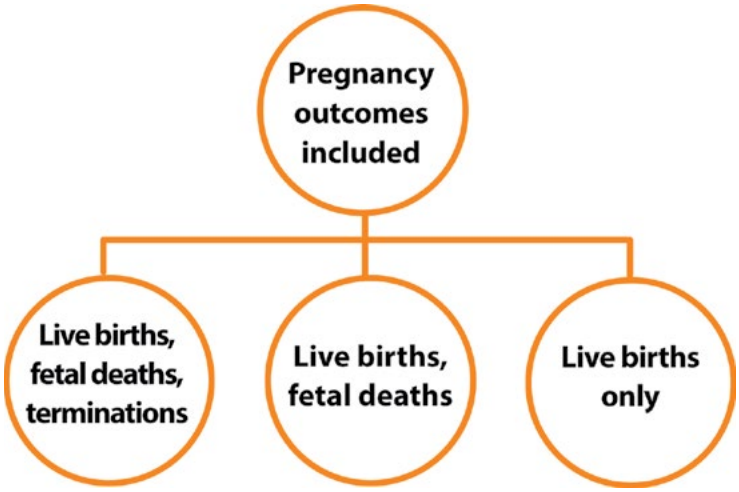
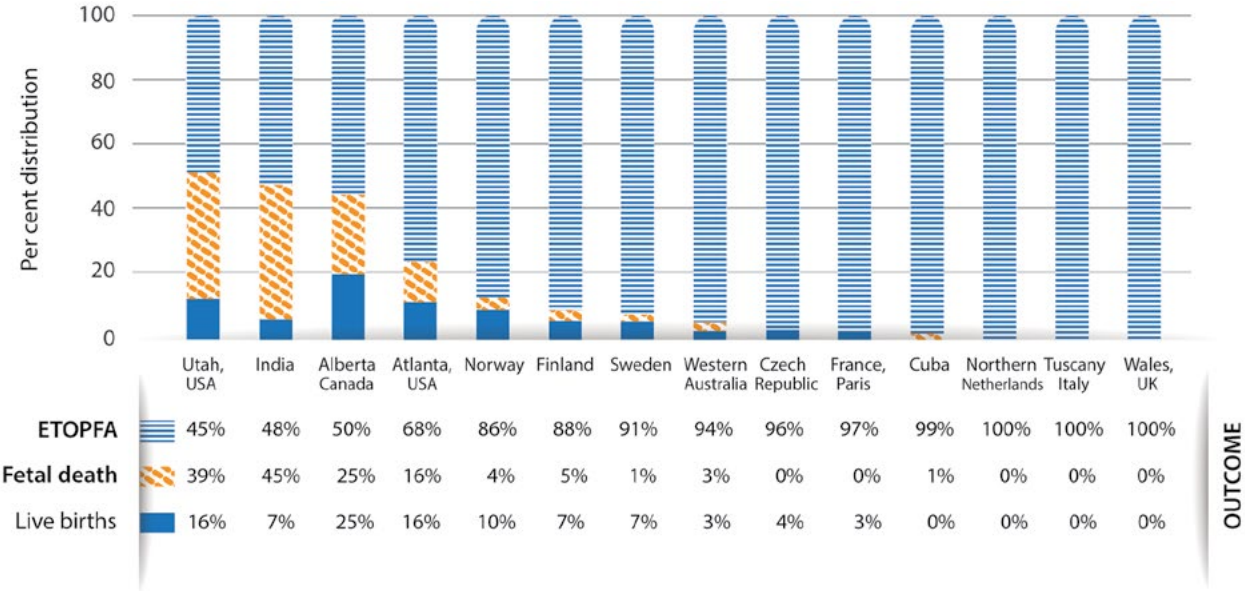


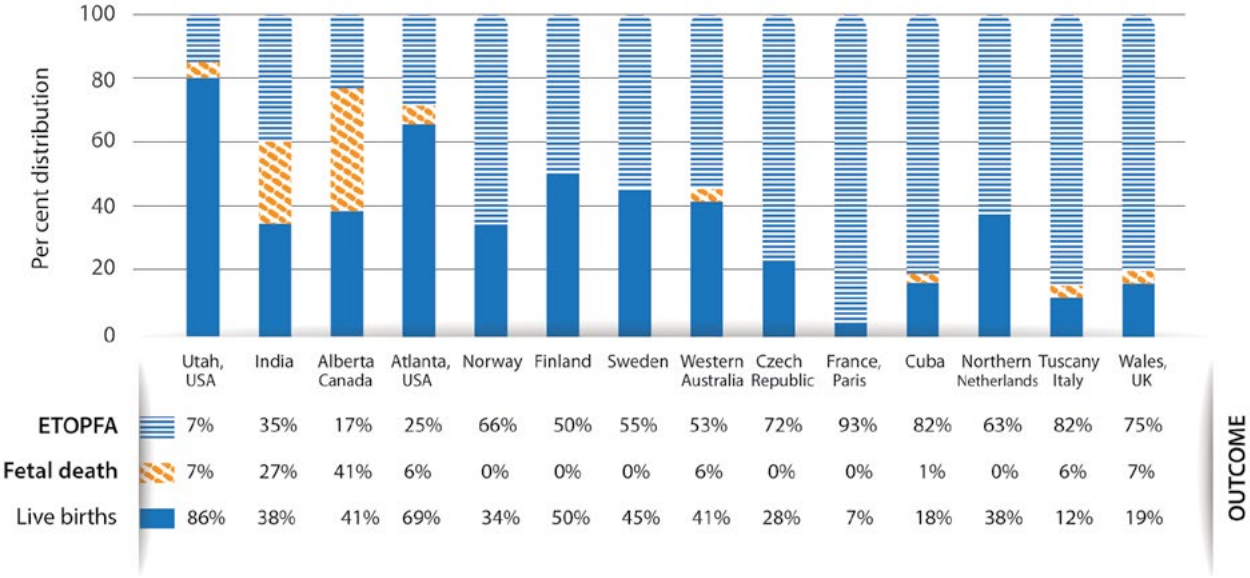
Fig. 3.13 and 3.14 show how inclusion of the different types of pregnancy outcomes has improved case ascertainment for anencephaly and spina bifida in 14 countries. It is important to note that programmes that include terminations of pregnancy find the terminations based on monitoring prenatal diagnosis. For example, the majority of fetuses with anencephaly are ascertained through fetal deaths or terminations. Fig. 3.13 indicates that in Wales, Tuscany (in Italy) and the Northern Netherlands, for example, 100% of fetuses with anencephaly are ascertained through pregnancy terminations, while in Utah (in the USA), 50% are ascertained through terminations, 40% through fetal deaths and only 10% as live births. Similarly, as Fig. 3.14 indicates, a much greater proportion of fetuses with spina bifida in Wales, Tuscany and Northern Netherlands are ascertained through pregnancy terminations, as compared to the other countries, regions or states represented.

Fig. 3.13. Distribution of pregnancy outcomes among ascertained anencephaly cases, 2007–2009



Note: all surveillance programmes are population based except for that in India.
ETOPFA = elective termination of pregnancy for fetal anomaly.
Source: International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR).

Fig. 3.14. Distribution of pregnancy outcomes among ascertained spina bifida cases, 2007–2009



Note: all surveillance programmes are population based except for that in India.
ETOPFA = elective termination of pregnancy for fetal anomaly.
Source: International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR).

Coding system

Most countries use the ICD-10 coding system *(12)* to classify congenital anomalies (please refer to Chapter 5 for more information on coding). While countries can develop their own local coding system, WHO recommends the use of ICD-10 for international reporting and comparisons.

The following sections describe case inclusion and exclusion criteria, procedures for data collection and components of a protocol for data collection.

Potential inclusion/exclusion criteria

To standardize the inclusion criteria for a case (fetus or neonate with a congenital anomaly) in a congenital anomalies surveillance programme, it is essential to characterize the criteria related to the diagnoses. Some examples of these criteria include the age at which the anomaly is diagnosed (discussed previously), the type of pregnancy outcome (discussed previously), the gestational age at delivery and birth weight, and maternal residence. More information about the latter two criteria follows.

Gestational age at delivery and birth weight

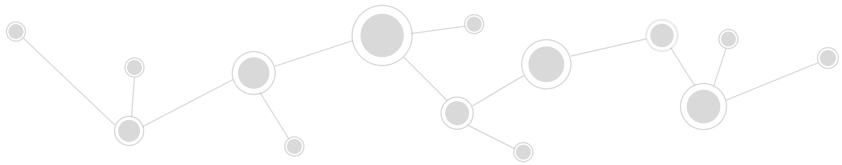
Gestational age at delivery and birth weight are important components of the case definition, because the frequency of some congenital anomalies varies depending on these factors. For example, preterm and low-birth-weight babies have a higher frequency of patent ductus arteriosus and undescended testes than term infants, and these conditions are considered physiologically normal among preterm infants if they resolve within a short time frame without intervention. Please refer to the Glossary of terms (p.76) for definitions of birth weight and gestational age.

Maternal residence

The mother's primary residence at the time of delivery or pregnancy termination is used by most congenital anomalies surveillance programmes to define the source population in which the cases occur. For example, residence can be defined as the mother's primary address during the 3 months prior to pregnancy and the first trimester of pregnancy. This is important because residence and place at delivery may be different, particularly in areas with strong referral patterns. It is essential to focus on residence rather than place at delivery, in order to correctly identify the appropriate denominator (the population of births from which the cases derive) and numerator. Correct denominators and numerators are prerequisites for accurate monitoring of the prevalence of a congenital anomaly and monitoring of changes over time.

Examples of inclusion criteria for population-based surveillance

- Live births and fetal deaths (stillbirths):
  - delivered with at least one of the selected major congenital anomalies (see Appendix A);
  - delivered to a mother who resides within a catchment area;
  - delivered at an age of 28 weeks' gestation or more, or, alternatively, a birth weight of at least 1000 g when gestational age is not available,



or with a gestational age defined by the programme. WHO recommends using an age of 28 weeks' gestation or more or birth weight of at least 1000 g when gestational age is not available. However, each country can use its own standards, which will allow it to link with vital statistics data.

- The congenital anomaly may be diagnosed prenatally (and confirmed at birth), at birth, during the neonatal hospitalization period, or up to an age limit predetermined for case ascertainment.
- If follow-up of the infants is available in the country, then the surveillance programme could consider capturing infants within a defined time period, to include the follow-up period (e.g. up to 1 year after birth).
- If the site has the capacity to capture terminations of pregnancy, the programme can include those fetuses with at least one of the selected major congenital anomalies at any gestational age, for the subset of congenital anomalies for which a prenatal diagnosis is considered definitive (e.g. anencephaly). Each country will have different provisions to capture termination of pregnancies, but in many settings this is done by including prenatal diagnostic centres as potential case-finding sources.
- Programmes that are interested in more detailed information on inclusion of prenatal diagnosis in congenital anomalies surveillance can find some useful and practical suggestions and tips in the guidelines developed by the NBDPN in the USA *(14)*.

Examples of inclusion criteria for hospital-based surveillance

- Live births and fetal deaths (stillbirths):
  - delivered with at least one of the selected major congenital anomaly (see Appendix A);
  - delivered at a participating hospital;
  - delivered with an age of 28 weeks' gestation or more, or, alternatively, a birth weight of at least 1000 g if gestational age is not available. The gestational age can be determined by each country, depending on its capacity to identify congenital anomalies occurring earlier than 28 weeks' gestation.
- The congenital anomaly may be diagnosed prenatally (and confirmed at birth), at birth or during the neonatal hospitalization period. The usual hospitalization period after delivery varies among countries, but could be defined as up to 7 days after birth.
- If the programme has the capacity to capture terminations of pregnancies, it can consider including those fetuses with at least one of the selected major congenital anomalies at any gestational age for the subset of congenital anomalies for which a prenatal diagnosis is considered definitive (e.g. anencephaly). Each country will have different provisions to capture termination of pregnancies, but in many settings this is done by including prenatal diagnostic centres as potential case-finding sources.

- Programmes that are interested in more detailed information on inclusion of prenatal diagnosis in congenital anomalies surveillance can find some useful and practical suggestions and tips in the guidelines developed by the NBDPN in the USA *(14)*.

Examples of exclusion criteria for both population- and hospital-based surveillance

- All neonates who do not have one of the selected major congenital anomalies listed in the initial inclusion list (see Appendix A).
- All neonates – with or without congenital anomalies – of less than 28 weeks' gestational age or with a birth weight of less than 1000 g, if gestational age is not available (or who are less than the gestational age or weight defined by the programme).
- All live births and fetal deaths with congenital anomalies identified outside of the participating hospital (hospital based) or outside of the ascertainment area (population based).
- Maternal residence status (the 3 months prior to pregnancy and the first trimester of pregnancy) outside of the catchment area.

Table 3.2 gives examples of criteria used by different countries to define congenital anomalies.

Table 3.2. Examples of criteria used by select countries to define congenital anomalies (ICBDSR, 2010)

Programme	Coverage	Age at diagnosis	Fetal death criteria
Australia: VBDR	Population based, statewide	≤18 years	20 weeks or 400 g
Costa Rica: CREC	Population based, national	≤3 days	22 weeks or 500 g
Finland	Population based, national	≤1 year	22 weeks or 500 g
Japan	Hospital based, national	≤7 days	22 weeks
Spain	Hospital based, national	≤3 days	24 weeks or 500 g
USA – California	Population based, regional	1 year	20 weeks
USA – Utah	Population based, regional	2 years	20 weeks

VBDR = Victorian Birth Defects Register
CREC= Costa Rican Birth Defects Register Center.

After a programme has defined the inclusion and exclusion criteria, a standardized data-collection process can be developed. This would include identifying the core ascertainment variables to be included in the surveillance programme and the development of a protocol for standardized data-collection procedures.

Core ascertainment variables

The first step in determining what core variables will be included in a surveillance programme is to define the goals and objectives of the congenital anomalies surveillance programme. Countries that already have a surveillance programme in place that includes the identification of populations at risk may consider including demographic variables such as maternal age, race and ethnicity, consanguinity, and other factors relevant to the local setting.

To facilitate data collection, countries can evaluate and summarize the availability of existing data sources (e.g. vital registries, and logbooks in hospital units), to determine what information regarding congenital anomalies is already being collected. Also, establishing the percentage of all births that are documented in vital registries, and determining whether the documentation includes only live births or both live births and fetal deaths, can be very useful.

After the list of core variables has been determined, these variables can then be incorporated into the methodology for case ascertainment. Table 3.3 lists core variables to be considered for inclusion in a congenital anomalies surveillance programme. Please refer to Appendix H for a list of core variables for consideration, and their definitions.

Table 3.3. Potential core ascertainment variables

Report	Father	Mother	Infant
• Case identification code • Date of report • Reporting hospital • City, province, state or territory • Name of person completing report	**Identification information** • Name: given name, family name • Date of birth or age • Race and ethnicity (if applicable)	**Identification information** • Name: given name, family name (including maiden name if appropriate) • Date of birth or age • Race and ethnicity (if applicable) • Address during the 3 months prior to pregnancy and the first trimester of pregnancy • Current address • Telephone number **Obstetric history** Total number of: • live births • stillbirths (fetal deaths) • spontaneous abortions • terminations of pregnancy	**Identification information** • Name: given name, family name • Date of birth • Sex • Date of diagnosis • Birth outcome **Birth measurements** • Gestational age (weeks) • Weight (g) • Length (cm) • Head circumference (cm) **Birth information** • Pregnancy outcome • Birth order, if multiple birth • Date of diagnosis • Date of death • Parental consanguinity[a] **Congenital anomaly/ anomalies present:** • Type • Description: o detailed description of congenital anomaly o drawings or illustrations of congenital anomaly • Code • Diagnostic technique(s) (e.g. radiographs) • Photographs *Note:* if a baby has more than one anomaly, any major congenital anomaly/anomalies are recorded first, followed by any other anomalies. **Autopsy results** • Description

[a]Consanguinity has long been recognized as a significant factor in the occurrence of autosomal recessive diseases. However, its effect in the determination of single major congenital anomalies remains controversial. Even though some studies have shown variable degrees of association between consanguinity and non-syndromic neural tube defects, hydrocephalus and oral clefts, the majority are based on small numbers of individuals. In addition, differences in methodological approaches hinder comparisons between the different studies. The situation appears to be different for congenital heart defects, for which significant increases among the offspring of consanguineous couples have been identified in several multinational studies *(15–21)*.

The following optional variables can be included if the surveillance programme in the country has the information available. See Appendix I for a listing of optional variables, and definitions for each.

- Report
  - source of information
- Father
  - occupation or work
  - family health history, environmental health history/ form or occupational health form
- Mother
  - demographic information
    - civil or marital status
    - occupation or work at time of conception
    - years and months residing in country
    - country identification number
    - weight (before pregnancy)
    - education (years or highest level)
    - religion (if applicable)
    - socioeconomic status
  - obstetric history
    - chronic diseases
    - date of last menstrual period
    - prenatal care, when it started, measure of adequacy
    - prenatal tests
    - family health history
    - medications or vaccines taken during pregnancy
    - any traditional medications commonly used
- Infant
  - Environmental health history/ form or occupational health form
  - Type of delivery

Data-collection methods and tools

Data gathering involves the use of appropriate recording forms. The data will provide the opportunity to measure the programme objectives, collect numbers of cases and help to determine trends. Once a decision is made regarding the data variables to be collected, an abstraction form (see Appendix G) can be created.

Paper-based data collection

For many years, data for congenital anomalies surveillance have been collected and processed using either a predetermined list (checkbox) format or the recording of verbatim descriptions on paper. These data-collection methods are still used widely for vital registration and various surveillance and research purposes. Paper-based data collection can be cost effective, but the process can require more time from data collection to analysis. It is also more prone to errors than electronic data collection because the data are first collected in a paper form and then transcribed into an electronic format for analysis *(22–24)*. Nevertheless, well-structured, paper-based forms are often still used in low-resource settings for collecting data on congenital anomalies.

Electronic data collection

An alternative to paper-based data collection is electronic data collection. Gradually, data-collection methods have evolved from manual, paper-based formats to electronic formats. Improving electronic surveillance programmes can be a long and costly process that requires regular update of a system's hardware and software to maintain a high level of security and data quality. The availability of electronic data collection will depend on the resources of each country.

The ideal collection tool allows data to be collected, transmitted securely to a data-management centre for storage and analysis, and retrieved, processed or analysed when necessary. In the last few decades, the evolution of technology has significantly improved the options for potential electronic data-collection tools.

Internet advances have allowed web-based reporting to progress gradually into real-time reporting *(25)*. The more recently introduced use of laptops, tablets and smart phones provides additional options for data collection. Because of the variability in access to, use of, and resources for electronic systems, each country will need to determine which method best fits its needs.

Data collection using smart phones or tablets

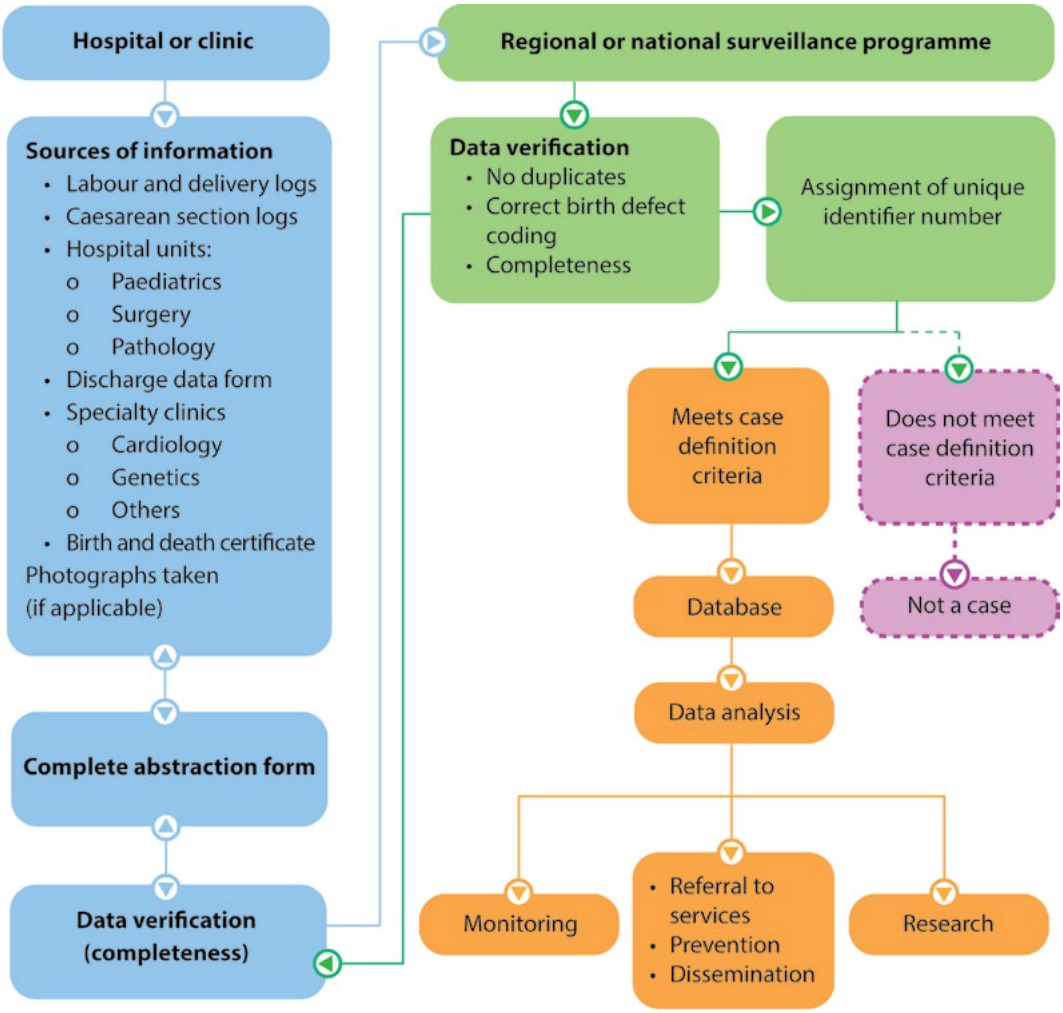
With the growing availability of smart phones and tablets in countries whose populations are predominantly middle and low income, their use as part of a congenital anomaly surveillance programme may improve the accuracy of data collection, and reduce the time required for, and cost of, data transmission and retrieval.

Users of smart phones and tablets can capture and transmit pictures, and may have access to databases of clinical information, including photographs to assist with differential diagnosis. Furthermore, the use of these mobile devices can be a novel, simple, efficient and instructive approach to the collection of data. The larger-sized tablets can also facilitate data entry. The use of these technologies could offer great potential for encouraging motivated personnel to contribute data to central databases using their mobile devices; however, such devices can easily be lost or stolen, so it is essential that they are programmed to encrypt all data, to ensure the privacy and security of information collected by the system.

Data management and protocols

Data management is essential to ensuring the integrity and confidentiality of surveillance data. Data management will not be possible unless all participating personnel are trained in the protocol for data collection. This ensures the proper use of all tools and a standardized method for data collection. This can be achieved by creating and maintaining an organized system for smooth data flow that ensures the regular availability of data but that also has high levels of security to preserve confidentiality. Fig. 3.15 is an example of how data collected at the hospital or clinic level progress through a surveillance programme.

Figure 3.15. Example flow chart for data management



The protocol for data collection and management includes procedures for:

- identification and registration of congenital anomalies by health-care professionals in each participating hospital unit;
- training of personnel responsible for coding congenital anomalies according to the ICD-10 *(12)* coding system;
- taking photographs of fetuses or neonates with congenital anomalies, if appropriate for the setting (see Appendix J);
- verifying the information at the participating hospital site;
- sending information to the regional or national-level surveillance programme.

Cases of congenital anomalies are usually identified as they occur in the participating delivery settings in a catchment area. Hospital personnel who identify a fetus or neonate with a congenital anomaly/anomalies usually record this information in a logbook, based on established standardized procedures.

Identification of cases is based on specific criteria, and diagnosis at birth is made by an experienced health-care provider. If an experienced health-care provider is not available at the site, photographs of the fetus or neonate with a congenital anomaly can be taken and kept in the medical record, for a later verification of the diagnosis by an experienced health-care provider or specialist, or a panel of experts, working as part of the congenital anomalies surveillance programme *(26)*. A member of the hospital staff, or a specially trained individual, usually takes at least three pictures – one frontal photograph of the fetus or neonate, one showing the back, and one or more pictures of the affected part(s) of the body. It is important, if possible, to place a tape measure next to the affected area or areas when taking the photograph, to document the size of the affected area, and ensure that some form of identification number is included in the photograph in order that it can be correctly linked to a particular case. For more suggestions on taking photographs of the fetus or neonate with a congenital anomaly, please refer to Appendix J.

The frequency of reporting data to the regional or national registry (e.g. weekly or monthly reporting) can be defined in the surveillance protocol and will depend on the availability of surveillance personnel and the individual circumstances of the participating unit.

A protocol is developed for the regional or national level, for personnel working in the surveillance programme. The protocol includes procedures for:

- data verification
- criteria to include cases in the database
- analysing data
- reporting and sharing data
- protecting the patient's and family's private information
- maintaining confidentiality (please refer to Chapter 2 for more information on privacy and confidentiality)
- case referral and management – clinical and surgical, if applicable.

Data-management personnel are responsible for reviewing information sent from the participating hospitals, and assessing the completeness of data forms, whether each item has been completed, and whether the verbatim and coded diagnoses have been included. In situations in which information appears incorrect or incomplete, personnel overseeing the verification of data can return the form to the site and ask for it to be re-reviewed or completed, or both. Cases submitted to the surveillance programme are then to be reviewed by a clinician, to verify the congenital anomaly and its coding prior to the case being entered into the database.

Data collection and management

Accurate data collection and management, including storage and analysis, are key components of any programme conducting congenital anomalies surveillance, and different instruments and methods of data gathering can be used for this purpose. Well-designed data systems improve data management, permit statistical analyses and data sharing among different surveillance programmes, and support linking of congenital anomalies data with other available information for surveillance, research and prevention purposes.

Data collection

It is important that the collection and analysis of data for the surveillance of congenital anomalies is done in a systematic way by trained surveillance personnel. It is also important that data are accurate and of high quality before analysis is performed. If done well, data analysis will provide accurate, timely and complete information on the occurrence of congenital anomalies.

Data quality

There are three main attributes to data quality: completeness, accuracy and timeliness.

Completeness refers to the extent to which data are all-inclusive and comprehensive. For example, all cases at a given source in a specific time frame have been identified, and all required data have been abstracted. Hospital audits and linkage of cases to vital records or to 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 (e.g. identification of missed diagnoses or coding issues), clinical reviews (e.g. verification of codes, tests and procedures), and verification of data entry (e.g. customized programmes for range checks, automated fields, rejection of data that are known to be inaccurate, routinely running data queries to identify duplicate entries, and identifying problems with variables).

Timeliness refers to the extent to which data are collected and analysed in a timely manner. It is measured by time that elapses between the date of diagnosis and date of abstraction; the date of abstraction and the date information is sent to the office; and the date of arrival in the office to the date entered in the system.

Data-collection procedures are carried out properly and systematically. Protocols usually include reviews of the information in the data sources, to verify that data are

being recorded in a standardized way. Also, if feasible, having a process whereby a sample of the medical records can be reviewed will ensure that information in the abstraction forms reflects the information on the medical record.

Poor-quality data can lead to erroneous conclusions about the occurrence of a congenital anomaly among a population and could have a substantial effect on the decision-making process of public health authorities.

The following are examples of factors that could affect data quality:

- missing values (e.g. empty data fields in the abstraction form);
- duplicate entry of cases;
- errors in the diagnosis, description or coding of congenital anomalies;
- bias related to lack or excess of representation:
  - if data include only very severe cases;
  - if data include only cases from urban settings;
  - if data include only private sector data sources;
  - if data include cases from outside of the catchment area.

Programmes interested in more detailed information on data management can find suggestions in the guidelines developed by the NBDPN in the USA *(14)*.

Data analysis and interpretation

Prevalence

In surveillance of congenital anomalies, the word "incidence" is not commonly used to describe the occurrence of congenital anomalies. This term refers to *all* new cases of congenital anomalies. Because spontaneous abortions cannot be counted accurately, the suggested measure of occurrence of congenital anomalies is "live birth prevalence", "birth prevalence" or "total prevalence".

In a population-based surveillance programme, the prevalence of congenital anomalies is calculated by aggregating the number of unduplicated existing cases (i.e. live births and fetal deaths or terminations) as the numerator, and the total number of live births among the source population as the denominator, for a specific catchment area and time period. For hospital-based surveillance, the prevalence of congenital anomalies is calculated by aggregating the number of unduplicated hospital cases as the numerator, and the total number of hospital live births as the denominator for a specific hospital. Hospital-based prevalence can include one or more hospitals.

Note: it is important to remember that hospital-based prevalence estimates can be biased, in that they give the prevalence of a condition only for the participating hospital. Prevalence estimates based on hospital data are not true estimates of the prevalence of a condition among a population.

When measuring the prevalence of congenital anomalies, it is important to note what is being counted in the numerator and in the denominator.

Usually, the prevalence of congenital anomalies is calculated and presented as prevalence per 10 000 live births. This prevalence can be calculated for all congenital

anomalies, for a specific individual anomaly, or for groups of anomalies. 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 $= \dfrac{\text{live birth cases}}{\text{total live births}} \times 10\,000$

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

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

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), or all. 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, its 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.

Case counts and crude prevalence are common measures of burden that are often presented with respect to time, geographic area, demographic characteristics, or various combinations (e.g. age-by-race-by-sex). When variations in prevalence are identified, they are described and analysed. Many factors could affect the prevalence of a health event: population changes due to migration, improved diagnostic procedures, enhanced reporting techniques, and changes in the surveillance system or methods. It is important to consider these factors when interpreting the results.

Description of changes over time is an important way of detecting trends. A comparison of the number of case reports collected during a particular time period may help identify differences in the number of cases for a current time period compared with time periods in previous years. These differences can help to determine seasonal patterns. The number of cases can vary by geographic location, and analysis by place can help identify where an increase in cases is occurring. In the case of rare congenital anomalies, the size of the geographic unit to be considered is important in order to provide stable

estimates. The analysis of demographic characteristics provides information on the characteristics of those individuals with particular congenital anomalies. The most frequently used demographic variables for analysis are age, sex, and race and ethnicity.

Appendix K lists prevalence estimates for the selected congenital anomalies presented in this manual for the years 2004–2008 and captured by surveillance programmes reporting to the ICBDSR in the USA.

Interpretation

Table 3.4 presents an example of calculating the prevalence of congenital anomalies that highlights the importance of knowing the denominator. Knowing only the number of cases (numerator data), without having information about the denominator, can result in a misinterpretation of the true burden of a congenital anomaly.

Table 3.4. Example of calculating prevalence and the importance of the denominator

	Numerator: total number of cases of congenital anomalies per year	**Denominator**	**Prevalence**	**Cases per 10 000 live births**
Country (example A)	100	100 000 (total live births per year in region or total catchment area)	0.001	10 per 10 000
Country (example B)	100	10 000 (total live births per year in eight hospitals of the total catchment area)	0.01	100 per 10 000
Country (example C)	100	1000 (total live births per year in one referral hospital of the total catchment area)	0.1	1000 per 10 000

Example A

A country decides to start a congenital anomalies surveillance programme in one region where the total number of live births per year is estimated to be 100 000. The surveillance programme will be population based and will include all fetuses or neonates identified with congenital anomalies in the region. After one year, the programme identifies 100 fetuses or neonates with congenital anomalies. The prevalence of congenital anomalies for that region will be 0.001 (10 cases per 10 000 live births).

Example B

A country decides to start a congenital anomalies surveillance programme in all maternity hospitals in one region, and eight hospitals will participate. Only fetuses or neonates with congenital anomalies born in one of the eight participating hospitals will be counted. The total number of births per year in the eight hospitals is estimated to be 10 000. After one year, the programme identifies 100 fetuses or neonates with congenital anomalies. The prevalence of congenital anomalies for those hospitals will be 0.01 (100 cases per 10 000 live births).

Example C

A country decides to start a congenital anomalies surveillance programme in a referral hospital in one region. This hospital is where prenatally identified fetuses with congenital anomalies are usually referred for delivery. The hospital typically has 1000 births per year. After one year, the hospital identifies 100 fetuses or neonates with congenital anomalies. The prevalence of congenital anomalies for that particular hospital is 0.1 (1000 cases per 10 000 live births).

Without knowing the denominator for each example, the prevalence estimate could be misinterpreted. The prevalence estimate for example C might indicate that this country has a high prevalence of congenital anomalies, when in reality the estimate resulted from a small denominator and the site is a referral hospital. The prevalence estimates for examples B and C represent the prevalence for eight hospitals and one referral hospital, respectively. These would not be considered true prevalence estimates. The prevalence estimate for example A is based on the total number of live births for a population and, thus, it yields the most accurate prevalence estimate.

4. Diagnosing and coding congenital anomalies

Initial list of congenital anomalies to consider for monitoring

Surveillance programmes can be developed to capture a variety of conditions. Although some countries may have more developed programmes than others, for the purpose of this manual, the following are suggested as an initial list of congenital anomalies to consider for monitoring. They were chosen because they are relatively easy to identify at birth, have significant public health impact, and, for some of them, the potential for primary prevention. Further, programmes interested in more detailed information on the inclusion of prenatal diagnosis in congenital anomalies surveillance programmes can find some useful and practical suggestions and tips in the guidelines developed by the NBDPN in the USA *(14)*.

- Congenital malformations of the nervous system
  - Anencephaly
  - Craniorachischisis
  - Iniencephaly
  - Encephalocele
  - Spina bifida

- Cleft lip and cleft palate
  - Cleft palate alone
  - Cleft lip alone
  - Cleft palate with cleft lip
- Congenital malformations of genital organs
  - Hypospadias
- Congenital malformations and deformations of the musculoskeletal system
  - Talipes equinovarus
  - Reduction defects of upper and lower limbs (longitudinal, transverse, and intercalary)
  - Exomphalos/omphalocele
  - Gastroschisis

As participating facilities or hospitals and surveillance programme personnel gain experience during the development process, additional congenital anomalies can be added in a stepwise fashion, starting with those that are of special interest or concern to the country or region, and eventually could include all of the major congenital anomalies listed in Chapter XVII: "Congenital malformations, deformations and chromosomal anomalies (Q00–Q99)" of the ICD-10 *(12)*. However, high-quality data on a smaller number of congenital anomalies will be more useful for public health than poor-quality data on all congenital anomalies. It is important that decisions on which defects to include are evaluated based on available resources. If the fetus or neonate has at least one eligible congenital anomaly, this and any other observable major and minor congenital anomalies are described in detail and included on the abstraction form (see Appendix G). When coding the congenital anomalies, it is important to be as specific as possible and avoid using codes that are nonspecific or too general. Please refer to Chapter 5 for more information about coding.

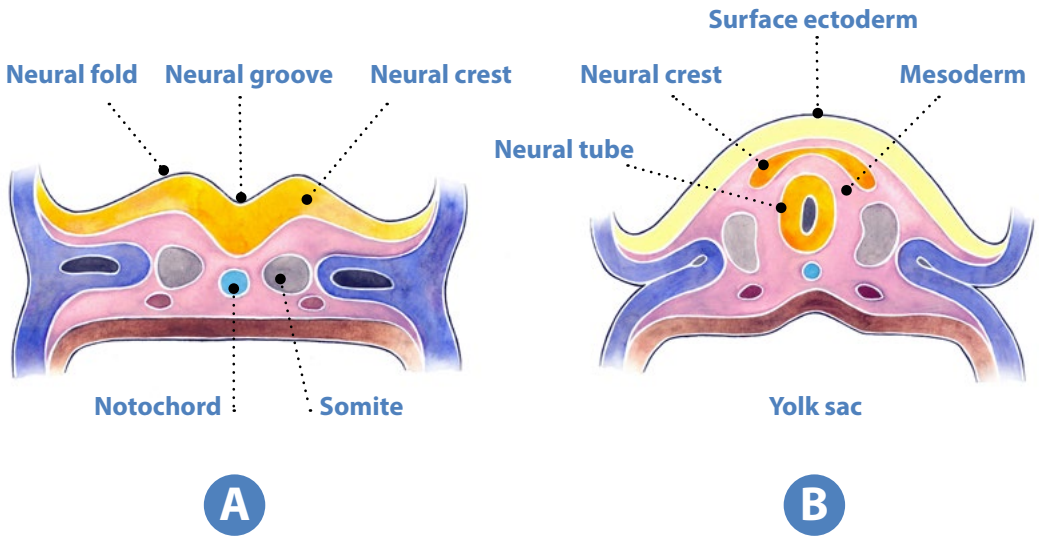
Congenital anomalies of the nervous system

Neural tube defects affect the brain and spinal cord, and are among the most common of the congenital anomalies (see Fig. 4.1). Panel A shows a cross section of the rostral end of the embryo at approximately three weeks after conception, showing the neural groove in the process of closing, overlying the notochord. The neural folds are the rising margins of the neural tube, topped by the neural crest, and demarcate the neural groove centrally. Panel B shows a cross section of the middle portion of the embryo after the neural tube has closed. The neural tube, which will ultimately develop into the spinal cord, is now covered by surface ectoderm (later, the skin). The intervening mesoderm will form the bony spine. The notochord is regressing. Panel C shows the developmental and clinical features of the main types of neural tube defects. The diagram in the centre is a dorsal view of a developing embryo, showing a neural tube that is closed in the centre but still open at the cranial and caudal ends. The dotted lines marked A and B refer to the cross sections shown in Panels A and B. Shaded bars point to the region of the neural tube relevant to each defect.

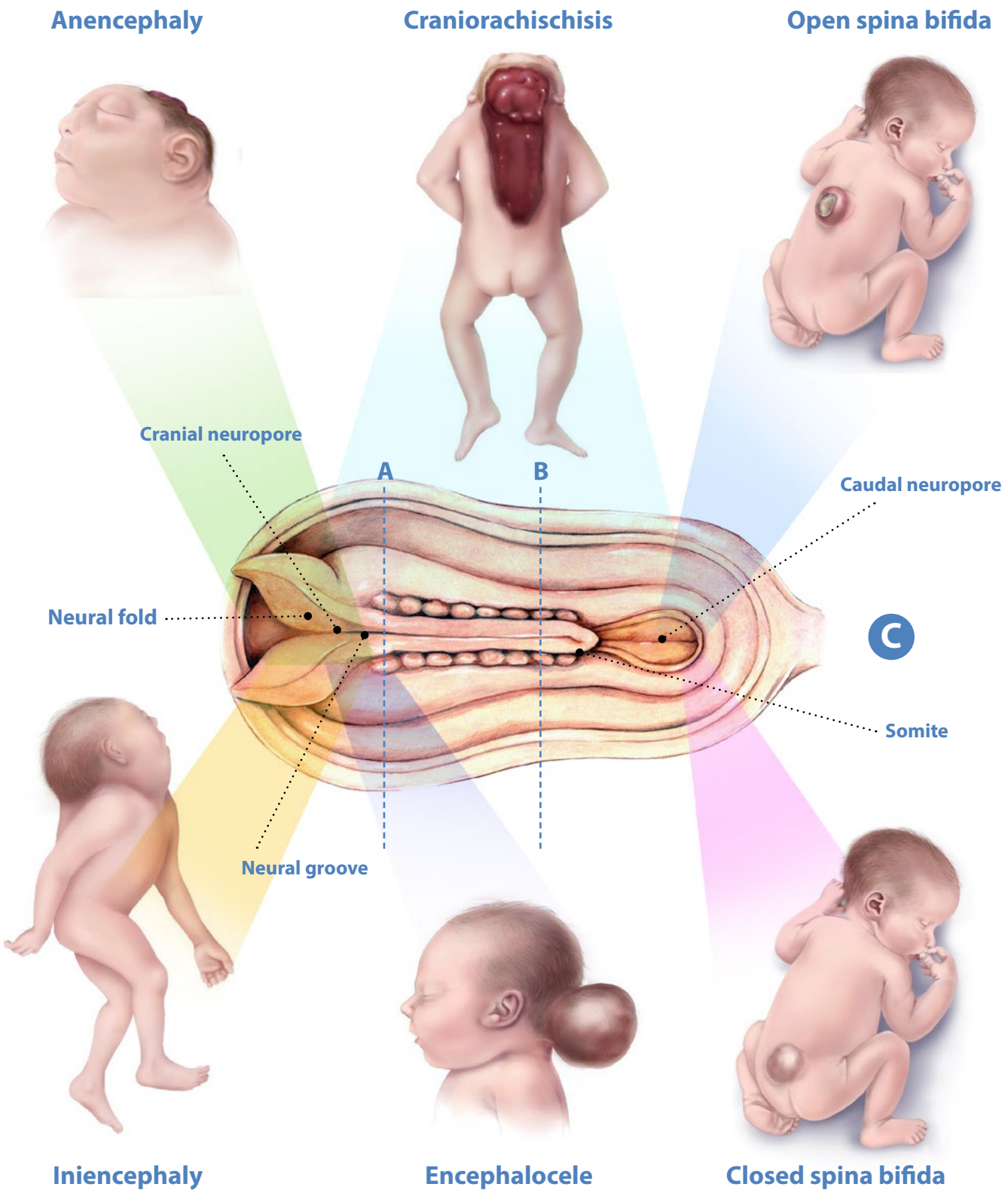
In anencephaly, the absence of the brain and calvaria can be total or partial. Craniorachischisis is characterized by anencephaly accompanied by a contiguous bony defect of the spine and exposure of neural tissue. In open spina bifida, a bony defect of the posterior vertebral arches (in this case, the lower thoracic vertebrae) is accompanied by herniation of neural tissue and meninges and is not covered by skin. In iniencephaly, dysraphia in the occipital region is accompanied by severe retroflexion of the neck and trunk. In encephalocele, the brain and meninges herniate through a defect in the calvaria. In closed spina bifida, unlike open spina bifida, the bony defect of the posterior vertebral arches (in this case, the lumbar vertebrae), the herniated meninges, and neural tissue are covered by skin.

The most prevalent types of neural tube defects are anencephaly, encephalocele and spina bifida.

Fig. 4.1. Neural tube defects



Source: adapted, with permsiion from the publisher, from Botto et al. N. Engl. J. Med. 1999;341:1509–19.


Anencephaly
Craniorachischisis
Open spina bifida
Cranial neuropore
A
B
Caudal neuropore
Neural fold
C
Somite
Neural groove
Iniencephaly
Encephalocele
Closed spina bifida

Anencephaly (Q00.0)

Anencephaly (see Fig. 4.2) 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. In addition to the term *anencephaly,* two other terms are used, although rarely, to describe this anomaly. One is *holoanencephaly*, in which the bone defect extends through the foramen magnum, affecting the entire skull; in the other, *meroanencephaly*, the bone defect is limited to the anterior part of the skull.

Two additional terms that are occasionally used as synonyms of anencephaly may be sources of confusion, because they also are used to describe other conditions. One is *acrania*, often used to refer to *acalvaria*, or absence of the neurocranium (calvarial bones, dura mater, and associated muscles) and believed to be unrelated to neural tube defects. The other is *acephaly,* which literally means "absence of the head" and is part of a pattern of anomalies observed in acardiac twins. These two terms – acrania and acephaly – are not coded as anencephaly; a diagnosis of acrania can be scrutinized to determine whether the diagnosis of anencephaly is more appropriate.

Fig. 4.2. Anencephaly (Q00.0)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Craniorachischisis (Q00.1)

Craniorachischisis (see Fig. 4.3) 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. Neonates with craniorachischisis may also have spinal retroflexion resembling the body habitus of neonates with iniencephaly.

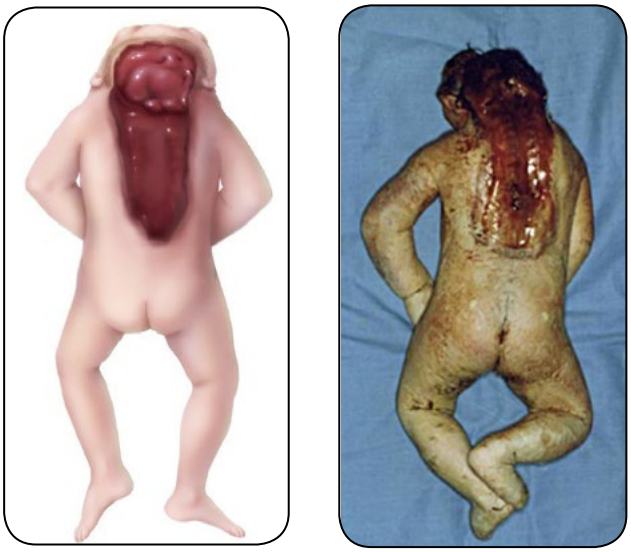


Fig. 4.3. Craniorachischisis (Q00.1)

Photograph source: courtesy CDC-Beijing Medical University collaborative project.

Iniencephaly (Q00.2)

Iniencephaly (see Fig. 4.4) is a rare and complex neural tube defect involving the occiput and inion, resulting in extreme retroflexion of the head, variably combined with occipital encephalocele or rachischisis of the cervical and thoracic spine. In iniencephaly, the cranium is always closed. This fact helps to differentiate iniencephaly from cases of anencephaly with spinal retroflexion.

Fig. 4.4. Iniencephaly (Q00.2)

Photograph source: courtesy CDC-Beijing Medical University collaborative project.

Relevant ICD-10 codes and Royal College of Paediatrics and Child Health (RCPCH) uses

Q00 Anencephaly and similar malformations (avoid using this general code if more specific information is available)

Q00.0 Anencephaly; Holoanencephaly; Meroanencephaly

Q00.00 Acrania

Q00.00 Amyelencephaly

Q00.01 Hemianencephaly

Q00.01 Hemicephaly

Q00.1 Craniorachischisis

Q00.2 Iniencephaly

Note: in cases in which anencephaly and spina bifida are present, but are not continuous, both are to be coded.

Exclusions

Q00.00 Acephaly

Q75.8 Acalvaria (specified absence of skull bone)

Q79.80 Other congenital malformations of musculoskeletal system. Amniotic band/ constriction band presence[a]

[a]The ICBDSR uses Q79.80 to identify the presence of an amniotic band. The anomaly is coded as the code for the specific congenital anomaly, as well as the Q79.80 amniotic band code.

Encephalocele
(Q01, Q01.0, Q01.1, Q01.2, Q01.8, Q01.9, Q61.90)

An encephalocele (see Fig. 4.5) is a pedunculated or sessile cystic lesion protruding through a defect in the skull. Encephaloceles can contain herniated meninges and brain tissue (encephalocele or meningoencephalocele) or only meninges (cranial meningocele). Most frequently, they are located in the occipital area, but in South-East Asia, the anterior location (frontal or nasofrontal) is most common (see Fig. 2.5c, d) *(28)*. Encephaloceles also are observed in the amniotic band sequence with entrapment of the head.

Fig. 4.5 Encephalocele

a. Frontal encephalocele (Q01.0)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

b. Nasofrontal encephalocele (Q01.1)

Photograph source: courtesy of Jaime Frías MD, USA.

c. Occipital encephalocele (Q01.2)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Relevant ICD-10 codes and RCPCH uses

Q01 Encephalocele (avoid using this general code if more specific information is available); Encephalomyelocele; Hydroencephalocele; Hydromeningocele, cranial; Meningocele, cerebral; Meningoencephalocele

Q01.0 Frontal encephalocele

Q01.1 Nasofrontal encephalocele

Q01.2 Occipital encephalocele

Q01.8 Encephalocele of other sites; if more specific codes are available, use the specific code below

Q01.80 Parietal encephalocele

Q01.81 Orbital encephalocele

Q01.82 Nasal encephalocele

Q01.83 Nasopharyngeal encephalocele

Q01.9 Encephalocele, unspecified

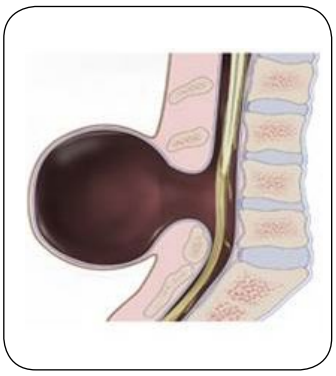
Spina bifida

(Q05, Q05.0, Q05.1, Q05.2, Q05.3, Q05.4, Q05.5, Q05.6, Q05.7, Q05.8, Q05.9)

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 (see Fig. 4.6). Hydrocephalus is a common complication, especially among children with open (membrane-covered) meningomyeloceles. Specific types of spina bifida include:

- Meningocele: this type of spina bifida is characterized by herniation of the meninges through a spine defect, forming a cyst filled with cerebrospinal fluid. It does not contain spinal cord, but might have some nerve elements.
- Meningomyelocele: this type of spina bifida consists of a protrusion of the meninges and the spinal cord through an opening in the vertebral column. This is the most common type of spina bifida, constituting about 90% of all cases.
- Myelocele: in this type of spina bifida, the open spinal cord, covered by a thin membrane, protrudes through the defect in the vertebral column.

Fig. 4.6. Spina bifida



a. Meningocele (Q05)

b. Myelomeningocele (Q05)

c. Myelocele or myeloschisis (Q05)

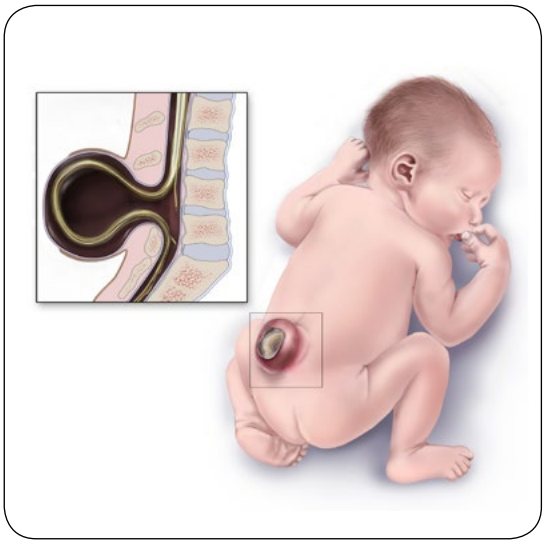
Photograph source (c): courtesy of CDC-Beijing Medical University collaborative project.

Fig. 4.6. Spina bifida

d. Lumbar spina bifida

e. Cervical spina bifida without hydrocephalus (Q05.5)

f. Lumbosacral spina bifida without hydrocephalus (Q05.7)



g. Lumbar spina bifida without hydrocephalus (Q05.7)

Photographs source (d): courtesy of Idalina Montes, MD and Rafael Longo, MD, FACS; Puerto Rico.; (e, f): courtesy of CDC-Beijing Medical University collaborative project.

Relevant ICD-10 codes and RCPCH uses

Q05 Spina bifida
(avoid using this general code if more specific information is available)
Hydromeningocele (spinal); Meningocele (spinal); Meningomyelocele
Myelocele; Myelomeningocele; Spinal rachischisis; Spina bifida (aperta)
(cystica); Syringomyelocele

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 (site unspecified)

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

Q05.8 Sacral spina bifida without hydrocephalus

Q05.9 Spina bifida, unspecified

Note: in cases in which anencephaly and spina bifida are present, but are not continuous, both are to be coded; however, when the malformations are counted, only anencephaly is counted.

Exclusions

Q07.0 Arnold–Chiari syndrome

Q76.0 Spina bifida occulta

Cleft lip and cleft palate

Cleft lip with or without cleft palate, and cleft palate alone, are referred to collectively as orofacial clefts. Descriptions for each of these conditions follow. To aid understanding of the individual conditions, the structure of a normal palate is shown in Fig. 4.7.

Fig. 4.7. Normal palate

Cleft palate
(Q35, Q35.1, Q35.3, Q35.5, Q35.59, Q35.9, Q87.0)

Cleft palate is characterized by an incomplete fusion of the secondary palate and can affect the soft and the hard palate (see Fig. 4.8), or only the soft palate. Laterality of cleft palate is difficult to ascertain and some believe it does not exist.

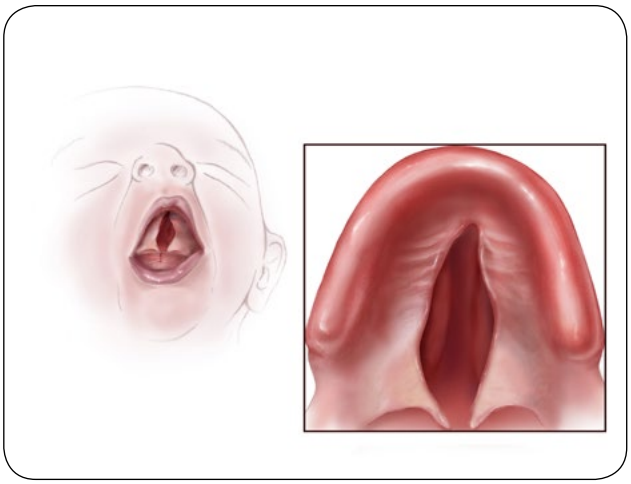


Fig. 4.8. Cleft hard palate with cleft soft palate (Q35.59)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Relevant ICD-10 codes and RCPCH uses

Q35	Cleft palate (avoid using this general code if more specific information is available) Fissure of palate; Palatoschisis
Q35.1	Cleft hard palate
Q35.3	Cleft soft palate
Q35.5	Cleft hard palate with cleft soft palate
Q35.59	Complete cleft palate Cleft hard palate with cleft soft palate
Q35.9	Cleft palate, unspecified
Q87.0	Robin (sequence or defect; includes micrognathia, posterior displacement of the tongue, cleft palate)[a]

Note: most specialists do not agree on the existence of cleft palate laterality, as this defect is the result of a failure of the palatal shelves to fuse in the midline. For this reason, the preferred codes are: Q35.1 Cleft hard palate; Q35.3 Cleft soft palate; Q35.59 Cleft hard palate with cleft soft palate; Q35.9 Cleft palate and Q87.0 Robin (sequence or defect; includes micrognathia, posterior displacement of the tongue, cleft palate).

Exclusions

Q35.7	Cleft uvula Bifid uvula
Q35.9	Submucous cleft
Q37–Q37.9	Cleft palate with cleft lip[b]
Q38.5	Absence of uvula

[a] ICD-10 lists Robin sequence as an exclusion from the Q35 series. However, because it is such a common condition, the 87.0 code is suggested.

[b] Cleft palate with cleft lip is included in a separate group (see following).

Cleft lip (Q36, Q36.0, Q36.9, Q36.90, Q36.99)

Cleft lip (see Fig. 4.9) is characterized by a partial or complete fissure of the upper lip. It can be unilateral or bilateral, and can be associated with a cleft of the gum. Cases of cleft lip with a cleft of the primary palate (anterior to the incisive foramen) is coded as cleft lip alone, because clefts of the primary palate involve only the alveolus, and are embryologically related to cleft lip and different from clefts of the secondary palate.

Fig. 4.9. Cleft lip. a, b. unilateral (Q36.9) c. bilateral (Q36.0)

Photographs sources (b)*:* courtesy of Jaime Frías MD, USA. (c): courtesy of Pedro Santiago DMD and Miguel Yanez MD, FACS, USA.

Relevant ICD-10 codes and RCPCH uses

Q36	Cleft lip (avoid using this general code if more specific information is available) Cheiloschisis; Congenital fissure of lip; Harelip; Labium leporinum
Q36.0	Cleft lip, bilateral
Q36.9 or Q36.90	Cleft lip, specified as unilateral
Q36.99	Cleft lip, unspecified
Exclusions	
Q36.1	Cleft lip, median
Q37–Q37.9	Cleft palate with cleft lip[a] Pseudocleft or microform cleft lip (no associated ICD-10 codes) Oblique facial clefts (Tessier type facial clefts) (no associated ICD-10 codes)

[a]Cleft palate with cleft lip is included in a separate group (see following).

Cleft palate with cleft lip
(Q37, Q37.0, Q37.1, Q37.2, Q37.3, Q37.4, Q37.5, Q37.8, Q37.9, Q37.99)

Cleft palate with cleft lip is characterized as a cleft of the upper lip extending through the primary and secondary palates, with or without extension through the soft palate (see Fig. 4.10).

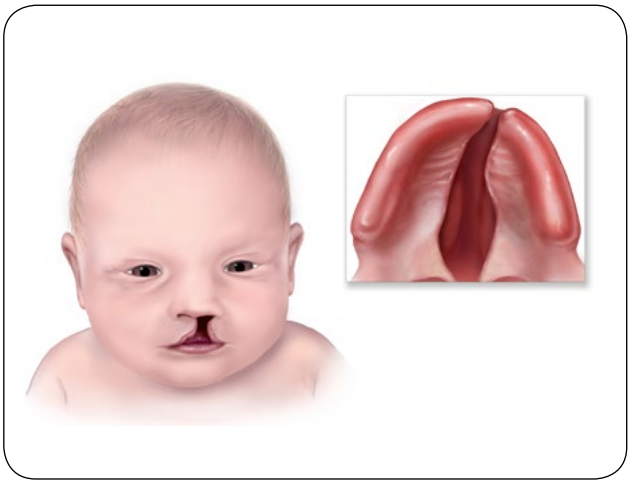


Fig. 4.10. Cleft palate with unilateral cleft lip (Q37.10)

Photograph source: courtesy of Pedro Santiago DMD and Miguel Yáñez MD, FACS, USA.

Relevant ICD-10 codes and RCPCH uses

Q37 Cleft palate with cleft lip
(avoid using this general code if more specific information is available)

Q37.0 Cleft hard palate with bilateral cleft lip

Q37.10 Cleft hard palate with cleft lip, specified as unilateral

Q37.19 Cleft hard palate with cleft lip, unspecified

Q37.2 Cleft soft palate with bilateral cleft lip

Q37.3 Cleft soft palate with unilateral cleft lip

Q37.4 Cleft hard palate and soft palate with bilateral cleft lip

Q37.5 Cleft hard palate and soft palate with unilateral cleft lip

Q37.59 Cleft hard palate and soft palate with cleft lip, unspecified

Q37.8 Unspecified cleft palate with bilateral cleft lip

Q37.9 Unspecified cleft palate with unilateral cleft lip

Q37.99 Cleft palate with cleft lip, unspecified

Exclusions

Oblique facial clefts (Tessier type facial clefts)

Q04.2 Cases with known or probable holoprosencephaly

Q79.80 Cases with known or probable amniotic band/constriction band presence[a]

[a]ICBDSR recommends using Q79.80 to identify the presence of an amniotic band. The anomaly is coded as the code for the specific anomaly, as well as the Q79.80 amniotic band code.

Congenital anomalies of genital organs

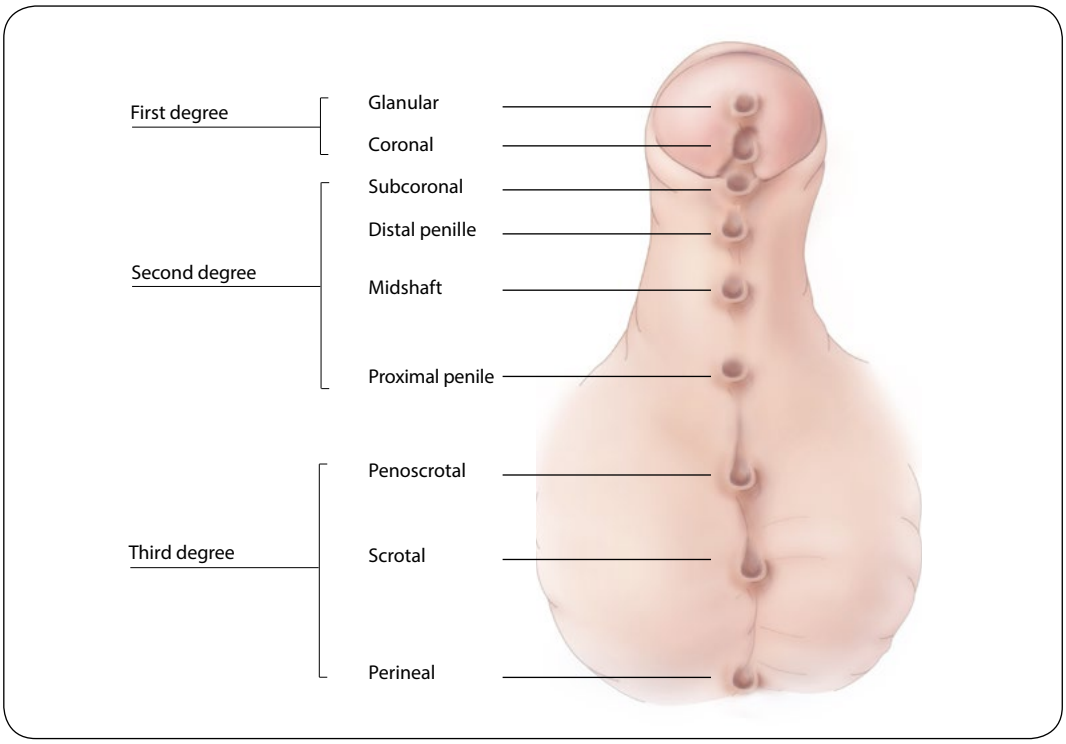
Hypospadias (Q54, Q54.0, Q54.1, Q54.2, Q54.3, Q54.4, Q54.8, Q54.9)

Displacement of the urethral meatus ventrally and proximally from the tip of the penis. It is classified according to the position of the meatus on the penis:

- First degree (Q54.0): the urethral meatus is located on either the glans (glanular hypospadias) or the corona (coronal hypospadias).
- Second degree (Q54.1): the urethral meatus is located in the balanopenile furrow or coronal sulcus (subcoronal hypospadias) or in the shaft of the penis (distal penile, midshaft, and proximal penile hypospadias).
- Third degree (Q54.2, Q54.3): the urethral meatus is located in the junction of the penis and the scrotum (penoscrotal or scrotal hypospadias) or the perineum (perineoscrotal, perineal, or pseudovaginal hypospadias).

The shortening of the ventral side of the penis found in hypospadias can result in a penile curvature, known as chordee. This is present more commonly in severe cases, but also can occur independently of hypospadias.

Fig. 4.11. Hypospadias (Q54, Q54.0, Q54.1, Q54.2, Q54.3, Q54.4, Q54.8, Q54.9)



Note: illustration indicates all possible locations for the malformation, but one case will not have all.

Subtypes:

Q54 Hypospadias (avoid using this general code if more specific information is available)

Q54.0 Hypospadias, balanic coronalglanular

Q54.1 Hypospadias, penile (subcoronal hypospadias)

Q54.2 Hypospadias, penoscrotal

Q54.3 Hypospadias, perineal

Q54.8 Other hypospadias, excludes: female hypospadias **(Q52.81)**

Q54.9 Hypospadias, unspecified

Congenital malformations and deformations of the musculoskeletal system

Talipes equinovarus (Q66.0, Q66.1, Q66.4, Q66.8)

The term "clubfoot" is sometimes used to describe several kinds of ankle or foot defects present at birth. However, orthopaedic specialists use it as a synonym for talipes equinovarus (see Fig. 4.12). The condition, which has a wide spectrum of severity, is characterized by adduction of the forefoot and midfoot, adduction of the heel or hind foot, and a fixed plantar flexion (equinus position) of the ankle *(29)*. In other words, the foot points downward and inward and is rotated outward axially. Other defects of the foot and ankle include talipes calcaneovalgus (in which the ankle joint is dorsiflexed and the forefoot deviated outwards) and talipes calcaneovarus (in which the ankle joint is dorsiflexed and the forefoot deviated inwards).

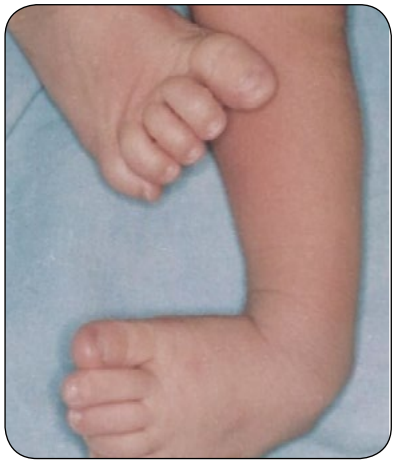
Relevant ICD-10 codes and RCPCH uses

Q66	Congenital deformities of feet (avoid using this general code if more specific information is available)
Q66.0	Talipes equinovarus
Q66.1	Talipes calcaneovarus
Q66.4	Talipes calcaneovalgus
Q66.8	Other congenital deformities of feet Clubfoot NOS (not otherwise specified)

Exclusions

Clubfoot, positional (no associated ICD-10 codes)

Clubfoot associated with neuromuscular diagnoses or syndromes, such as arthrogryposis multiplex congenital, congenital myotonic dystrophy, and diastrophic dysplasia (no associated ICD-10 codes)



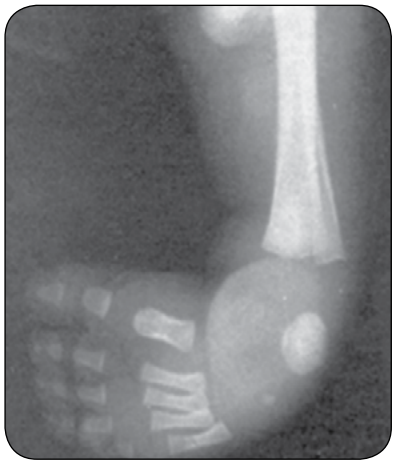


Fig. 4.12. Talipes equinovarus (Q66.0)

Photograph and X-ray source: courtesy of Idalina Montes MD and Rafael Longo MD, FACS, Puerto Rico.

Reduction defects of upper and lower limbs (longitudinal, transverse and intercalary)

Limb deficiencies make up a large number of defects in limb development, characterized by the total or partial absence or different degrees of hypoplasia of the skeletal structures of the limbs. They are classified into three large groups: longitudinal, transverse and intercalary limb deficiencies. Some cases will have multiple limb defects, and therefore will be classified in more than one of these groups.

Longitudinal limb deficiencies (Q71.4, Q71.5, Q71.6, Q72.4, Q72.5, Q72.6, Q72.7)

Longitudinal limb deficiencies (see Fig. 4.13–4.18) refer to the partial absence of a limb extending parallel to the long axis of the limb. They typically involve specific components of the limbs: preaxial (first ray: thumb or radius in the arm(s), or both, or first toe or tibia in the leg(s), or both); postaxial (fifth ray: fifth finger or ulna in the arm(s), or both, fifth toe or fibula in the leg(s), or both); or central components (typically, third or fourth rays in the hand(s) (also called split hand or lobster-claw hand) or foot (also called split foot), or both.

Fig. 4.13. Radial aplasia (Q71.4)

X-ray source: courtesy of John Wiley and Sons ©2011. Am. J. Med. Genet. A. 155:3071–3074.

Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

Fig. 4.14. Ulnar aplasia/ hypoplasia (Q71.5)

X-ray source: courtesy of John Wiley and Sons ©2011. AM. J. Med. Genet. A. 70:107-113.

Fig. 4.15. Femoral aplasia on the right (Q72.4) and femoral (Q72.4) and fibular aplasia (Q72.6) (on the left)

X-ray source: courtesy of Jaime Frías MD, USA.

Fig. 4.16. Tibial aplasia (Q72.5)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Fig. 4.17. Split hand (Q71.6)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Fig. 4.18. Split foot (Q72.7)

Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

Transverse limb deficiencies (Q71.0, Q71.2, Q71.3, Q72.0, Q72.2, Q72.3)

Transverse limb deficiencies (see Fig. 4.19–4.29) refer to the complete or partial absence of distal structures of a limb in a transverse plane at the point where the deficiency begins, with proximal structures being essentially intact. Transverse limb deficiencies also are known as congenital amputations.

Fig. 4.19. Congenital absence of forearm and hand (Q71.2)

Fig. 4.20. Congenital absence of both forearm and hand (Q71.2)

Fig. 4.21. Congenital absence of lower leg and foot (Q72.2)

Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

Fig. 4.22. Aphalangia of the hand. Partial absence of the phalanges (Q71.30)

Fig. 4.23. Aphalangia of the feet. Partial absence of the phalanges (Q72.30)

Photographs source: courtesy of John Wiley and Sons ©2009. Am. J. Med. Genet. A. 149A:93–127.

Fig. 4.24. Adactyly of the hand (Q71.30)

Photographs and X-ray source: courtesy Dr E Gene Deune MD, Associate Professor, Johns Hopkins Dept of Orthopedic Surgery, Division of Hand Surgery, Baltimore, MD.

Fig. 4.25. Adactyly of the feet (Q72.30)

Photographs source: courtesy of John Wiley and Sons ©2009. Am. J. Med. Genet. A. 149A:93–127.

Fig. 4.26. Oligodactyly of the hand (Q71.30)

Fig. 4.27. Oligodactyly of the foot (absent hallux) (Q72.31)

Photographs source: courtesy of John Wiley and Sons ©2009. Am. J. Med. Genet. A. 149A:93–127.

Fig. 4.28. Amelia upper limb (Q71.0)

Fig. 4.29. Amelia of the lower limb (Q72.0)

Photographs source: courtesy of CDC-Beijing Medical University collaborative project.

Intercalary limb deficiencies (Q71.1 and Q72.1)

Intercalary limb deficiencies refer to the complete or partial absence of the proximal or middle segment(s) of a limb, with all or part of the distal segment present (see Fig. 4.30-4. 31).

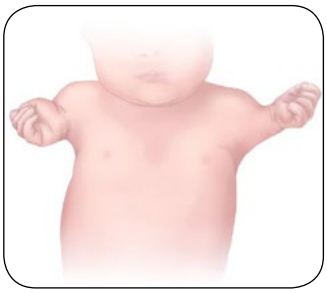


Fig. 4.30. Reduction defects of upper arm and forearm with hand present (Q71.1)

Photograph source: courtesy of Jaime Frías MD, USA.

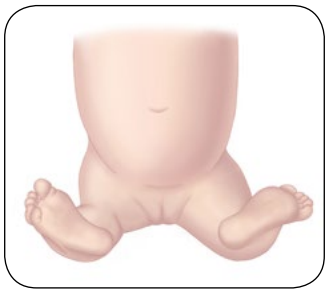


Fig. 4.31. Reduction defects of thigh and lower leg with foot present (Q72.1)

Relevant ICD-10 codes and RCPCH uses

Reduction defects of upper limbs

Q71 Reduction defects of upper limb (avoid using this general code if more specific information is available)

Q71.0 Congenital complete absence of upper limb(s)

Amelia of upper limb

Q71.1 Congenital absence of upper arm and forearm with hand present

Phocomelia of upper limb

Q71.2 Congenital absence of both forearm and hand

Q71.3 Congenital absence of hand and finger(s)

Q71.30 Congenital absence of finger(s)

(Remainder of hand intact)

Aphalangia: absent phalanx (an individual bone in a finger) or phalanges

Adactyly: absence of fingers (generally refers to all fingers on a hand, although soft tissue nubbins without bones can be present)

Oligodactyly: fewer than 10 complete fingers

Q71.31 Absence or hypoplasia of thumb

(Other digits intact)

Q71.4 Longitudinal reduction defect of radius

Radial aplasia/hypoplasia

Clubhand (congenital)

Radial clubhand

Q71.5 Longitudinal reduction defect of ulna

Ulnar aplasia/hypoplasia

Q71.6 Lobster-claw hand

Split hand

Congenital cleft hand

Q71.8 Other reduction defects of upper limb(s)

Congenital shortening of upper limb(s)

Q71.9 Reduction defect of upper limb, unspecified

Reduction defects of lower limbs

Q72 Reduction defects of lower limb (avoid using this general code if more specific information is available)

Q72.0 Congenital complete absence of lower limb(s)

Amelia of lower limb

Q72.1 Congenital absence of thigh and lower leg with foot present

Phocomelia of lower limb

Q72.2 Congenital absence of both lower leg and foot

Q72.3 Congenital absence of foot and toe(s)

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

Aphalangia: absent phalanx (an individual bone in a toe) or phalanges

Adactyly: absence of toes (generally refers to all toes on a foot, although soft tissue nubbins without bones can be present)

Oligodactyly: fewer than 10 complete toes

Q72.31 Absence or hypoplasia of first toe with other digits present

Q72.4 Longitudinal reduction defect of femur

(commonly referred to as femoral aplasia/hypoplasia)

Proximal femoral focal deficiency

Q72.5 Longitudinal reduction defect of tibia

Tibial aplasia/hypoplasia

Q72.6 Longitudinal reduction defect of fibula

Fibular aplasia/hypoplasia

Q72.7 Split foot

Q72.8 Other reduction defects of lower limb(s)

Congenital shortening of lower limb(s)

Q72.9 Reduction defect of lower limb, unspecified

Reduction defect of unspecified limb

Q73 Reduction defects of unspecified limb (avoid using this general code if more specific information is available)

Q73.0 Congenital absence of unspecified limb(s)

Amelia NOS

Q73.1 Phocomelia, unspecified limb(s)

Phocomelia NOS

Q73.8 Other reduction defects of unspecified limb(s)

Longitudinal reduction deformity of unspecified limb (s)

Ectromelia NOS

Hemimelia NOS

Reduction defect NOS

Q73.80 Absent digits, unspecified

Exclusions

Q77.0–Q77.9, Q78.0–Q78.9 Generalized limb shortening including skeletal dysplasias (osteochondrodysplasias)

Q79.80 Cases with known or probable amniotic band/constriction band presence[a]

Q84.6 Nail hypoplasia

Q89.80 Lower extremity deficiencies with caudal dysgenesis

Q87.2 Sirenomelia

All types of brachydactyly (no associated ICD-10 codes)

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[a] ICBDSR recommends using Q79.80 to identify the presence of an amniotic band. The anomaly is coded with the code for the specific anomaly as well as the Q79.80 amniotic band code.

Exomphalos/omphalocele (Q79.2)

Omphalocele (see Fig. 4.32) is a birth defect of the anterior abdominal wall, in which the herniated intestines and abdominal organs are usually covered by a membrane consisting of the peritoneum and amnion. In contrast to gastroschisis, in which the abdominal defect is lateral to the umbilicus, 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.

Fig. 4.32. a, b. Omphalocele (Q79.2) and c. with ruptured membrane

Photographs Source: courtesy of CDC-Beijing Medical University collaborative project.

Relevant ICD-10 codes and RCPCH uses

Q79 Congenital malformations of the musculoskeletal system, not elsewhere classified (avoid using this general code if more specific information is available)

Q79.2 Exomphalos/omphalocele

Exclusions

Q79.3 Gastroschisis

Q79.8 Umbilical hernia

Gastroschisis (Q79.3)

Gastroschisis is also a birth defect of the anterior abdominal wall, accompanied by herniation of the small intestine and part of the large intestine, and occasionally other abdominal organs, into the amniotic cavity (see Fig. 4.33). Importantly, the herniated organs lack a protective membrane. The extruded abdominal contents can be matted and covered by a thick fibrous material, but this membrane does not resemble skin. Gastroschisis occurs lateral to the umbilicus (generally to the right).

Gastroschisis and omphalocele can be confused with one another when the membrane covering the omphalocele has ruptured. However, careful examination demonstrating the position of the abdominal opening lateral to the umbilical cord insertion helps confirm the diagnosis of gastroschisis.

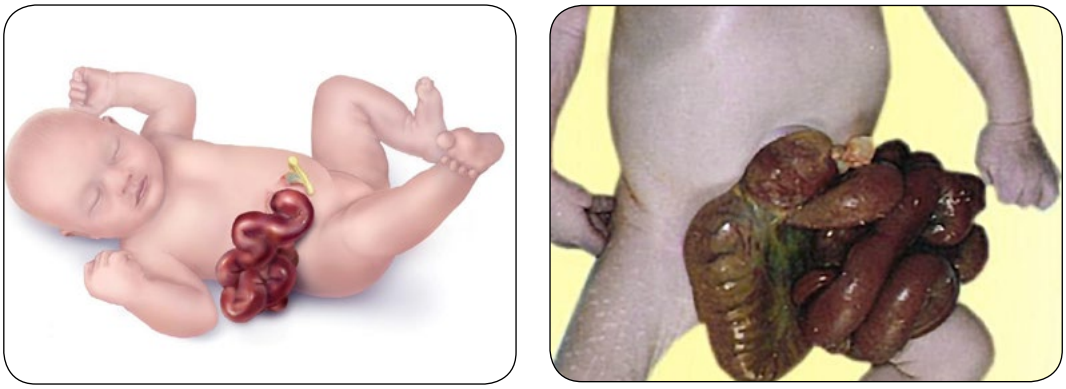

Fig. 4.33. Gastroschisis (Q79.3)

Photograph source: courtesy of CDC-Beijing Medical University collaborative project.

Relevant ICD-10 codes and RCPCH uses

Q79 Congenital malformations of the musculoskeletal system, not elsewhere classified (avoid using this general code if more specific information is available)

Q79.3 Gastroschisis

Exclusions

Q79.2 Exomphalos/omphalocele

Q89.81 Limb–body wall complex

Coding of congenital anomalies

One of the essential aspects of a congenital anomalies surveillance programme is its ability to efficiently generate information. Central to this process is the proper and accurate coding of the recorded diagnostic information. Coding of diagnostic information using a disease classification system allows a surveillance programme to capture and classify cases with congenital anomalies in a standardized way. Entering coded information into an electronic system makes it easier to retrieve and analyse the data. It is important to understand and follow a standardized coding system, in order to accurately and consistently classify and code the various types of congenital anomalies.

The more precise the clinical description of congenital anomalies present in a fetus or neonate is, the more accurate the classification and coding that can be achieved. For example, not knowing the lesion level of spina bifida (such as cervical, thoracic or lumbar) or whether hydrocephalus is present, or both, would result in coding the congenital anomaly as "spina bifida, unspecified". It is important to obtain the best possible clinical description, carefully review and classify the congenital anomaly, and assign the right code(s) based on the description. To the extent possible, the database can preserve both the codes and the detailed clinical description.

Photographs of the external congenital anomalies present can supplement the clinical description and help to ensure that the proper code is assigned. Although it is relatively easy to take photographs, it requires some training to obtain the best photographs (e.g. timing, views). Please refer to Appendix J for suggestions for taking photographs of fetuses or neonates with congenital anomalies. Privacy issues also need to be considered and appropriate measures to ensure confidentiality should be in place. Because some photographs may identify the neonate, it is critical to maintain these securely as confidential surveillance documents. More information on privacy and confidentiality is included in Chapter 2.

International Classification of Diseases

The ICD-10 is considered the international standard diagnostic classification system for all general epidemiological purposes, health data-management purposes, and clinical use, and is widely used in many countries as a classification system for diseases. Use of this standardized coding system will facilitate partnerships and collaborations with other programmes using the same coding system.

The ICD-10 is developed and maintained by WHO. The most recent version is available on the WHO website *(12)*. It is available in the six official languages of WHO (Arabic, Chinese, English, French, Russian and Spanish), as well as in 36 other languages. A list of contact points for the 42 language versions of ICD-10 can be found in reference *(32)*.

On the WHO website, an ICD-10 interactive self-learning tool is available for training purposes *(33)*.

The ICD-10 has been used to classify diseases in health records and vital records, as a basis for the compilation of national mortality and morbidity statistics by WHO Member States.

The 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)" *(12)*. This chapter contains the following blocks of codes:

- **Q00–Q07** Congenital malformations of the nervous system
- **Q10–Q18** Congenital malformations of eye, ear, face and neck
- **Q20–Q28** Congenital malformations of the circulatory system
- **Q30–Q34** Congenital malformations of the respiratory system
- **Q35–Q37** Cleft lip and cleft palate
- **Q38–Q45** Other congenital malformations of the digestive system
- **Q50–Q56** Congenital malformations of genital organs
- **Q60–Q64** Congenital malformations of the urinary system
- **Q65–Q79** Congenital malformations and deformations of the musculoskeletal system
- **Q80–Q89** Other congenital malformations
- **Q90–Q99** Chromosomal abnormalities, not elsewhere classified.

ICD-10 modifications

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 contains additional codes for some specific congenital anomalies not found in the ICD-10, or add an extra digit, or both, to allow for more detailed coding of some defects and certainty of diagnosis.

The following is an example of how the RCPCH (formerly known as the British Paediatric Association) developed an adaptation of the ICD-10 by adding an extra digit to the ICD-10 codes to expand and allow for more detailed coding *(34)*. For example, in this adaptation, specific codes are added to differentiate parietal, orbital, nasal and nasopharyngeal encephaloceles, as follows:

- **Q01.8** Encephalocele of other sites (ICD-10 code)
- **Q01.80** Parietal encephalocele
- **Q01.81** Orbital encephalocele
- **Q01.82** Nasal encephalocele
- **Q01.83** Nasopharyngeal encephalocele

Personnel responsible for diagnosing and coding

Depending on how a congenital anomalies surveillance programme is set up, the coding of congenital anomalies may take place in a hospital or clinic, or at the central registry, based on the clinical information provided. It is important to train the hospital or clinic staff responsible for diagnosing and coding congenital anomalies. If coding is done at the hospital or clinic, it also is important and recommended that someone who is knowledgeable about congenital anomalies (e.g. a neonatologist, paediatrician, clinical geneticist or dysmorphologist) reviews and confirms the diagnosis and assigns the proper codes. Codes for, or specific descriptions of, congenital anomalies are then submitted to the central registry, where final review and verification of all codes reported by participating sites occurs.

Not every site will have personnel who are knowledgeable about congenital anomalies. If no knowledgeable staff member is available, it is suggested that coding be done at the central registry level. Having a description of a congenital anomaly that is as complete and thorough as possible, and that includes photographs with the description, will increase the likelihood that the reviewer at the registry will be able to assign an accurate code. It is important to remember, however, that a description that includes abbreviated words can easily be misunderstood or misinterpreted by the reviewer.

The reliability of coding can also be affected by the expertise of the personnel recording the information and the expertise of the surveillance staff reviewing the information.

Effect of the certainty of diagnosis on coding

Prenatal and postnatal diagnosis

The certainty of a diagnosis can vary for live births and fetal deaths (stillbirths), as well as when the diagnosis is prenatal only or postnatal. With pregnancy terminations, a prenatal diagnosis may not be verified for many reasons, including the method of termination, the condition of the specimen, or a lack of post-termination examination or autopsy. Programmes that are interested in more detailed information on inclusion of prenatal diagnosis in congenital anomaly surveillance can find some useful and practical suggestions and tips in the guidelines developed by the NBDPN in the USA *(14)*. Among liveborn neonates who die shortly after birth, the diagnosis could also cause difficulties if certain examinations (e.g. X-rays and karyotyping) or an autopsy are not done.

Coding possible and confirmed diagnoses

When the diagnosis is uncertain (e.g. hydrocephalus suggested by prenatal ultrasound, but for which no postnatal confirmation is done), it is beneficial to distinguish possible diagnoses from confirmed diagnoses. This can be done by using a separate field on the congenital anomalies abstraction form to include this information (see Appendix G), or adding an extra digit to the ICD-10 codes, which has been done by some surveillance programmes *(34)*.

Coding multiple congenital anomalies

Approximately 75% of fetuses and neonates with a major congenital anomaly present as isolated anomalies, and the remaining 25% have more than one major anomaly *(35, 36)*.

More details about the types of congenital anomalies according to clinical presentation are presented in Appendix C.

When more than one congenital anomaly is present, a detailed description of each major anomaly is recorded. Congenital anomalies surveillance programmes vary regarding the number of codes they record for a fetus or neonate, but allowing coding for at least 10 anomalies should be sufficient. Major anomalies are given priority over any minor anomalies for being captured within the available 10 (or more) diagnoses recorded.

Certain syndromes can also be coded according to the ICD-10 classification *(12)*. When the ICD-10 code is not specific enough (e.g. codes listed in the group Q87 – "Other specified congenital malformation syndromes affecting multiple systems"), then using the classification developed by the RCPCH could be beneficial *(34)*. Regardless of which classification(s) is used, a thorough description of any observed anomaly is very important for accurate coding of congenital anomalies.

Use of codes for surveillance, data analysis and presentation

The following information is intended primarily for the staff of the central registry. To record ICD-10 codes, the most specific code format (i.e. Q##.#) is used. For example, frontal encephalocele is coded as Q01.0. The three-character format or group code (i.e. Q##) is commonly used only for data analysis and presentation purposes, to group and report all types of any condition. For example, when analysing and reporting all types of (total) encephalocele, the three-character format (Q01) can be used. Diagnoses coded as possible would still be excluded.

Along with the ICD-10 codes, a list with exclusions of several anomalies is provided in the ICD-10 classification system. The term "exclusion" does not necessarily mean that the case is excluded from the registry. Rather, it means that the particular anomaly is not coded with the same code or codes. For example, because "spina bifida occulta" is considered a different anomaly from the other types of spina bifida and has a specific code (Q76.0), the Q05.# ICD-10 codes are not used. Another example is cleft palate with cleft lip. If a fetus or neonate has both a cleft palate and cleft lip, the anomaly is not coded with a cleft palate code (Q35.1–Q35.9), but instead with a code listed under cleft palate with cleft lip (Q37.0–Q37.9).

It is important to keep in mind that while, for surveillance purposes, all major anomalies affecting a fetus or neonate can be coded, for data analysis and presentation, the criteria to include or exclude certain anomalies can determine which codes are used. For example, although a case may have both anencephaly (Q00.0) and lumbar spina bifida without hydrocephalus (Q05.7), for reporting purposes, the case may be analysed only with other cases of anencephaly. In addition, if an anomaly is secondary to another anomaly, such as clubfoot with spina bifida, the case would be included in analyses of spina bifida (Q05.#) but not in analyses of clubfoot (Q66.0 or Q66.8). However, when both anencephaly and spina bifida are present and are contiguous, this is the condition called craniorachischisis and there is a unique ICD-10 code for that (Q00.1); therefore, anencephaly and spina bifida are not coded separately.

Examples of the assignment of codes based on clinical description are presented next.

Example 1

The following diagnosis and clinical description are provided for a neonate:

"spina bifida with LS meningocele and massive hydrocephalus"

In this case, "LS" is used to abbreviate "lumbosacral". Although the description may 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. The suggested ICD-10 code to assign to this case is Q05.2 (lumbosacral spina bifida with hydrocephalus). This case would not be included in analyses of hydrocephalus as a primary anomaly.

Example 2

The following diagnosis and clinical description are provided for a neonate:

"cleft lip and palate"

Because 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 suggested ICD-10 code is Q37.9 (unspecified cleft palate with unilateral cleft lip).

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.

Example 3

The following diagnosis and clinical description are provided on a medical record:

"cleft lip NOS; spina bifida NOS; ear tags"

The abbreviation "NOS" means "not otherwise specified". The suggested ICD-10 code for cleft lip NOS is Q36.9 (cleft lip NOS) and for spina bifida NOS is Q05.9 (spina bifida, unspecified). Ear tags are considered minor anomalies; therefore, coding them is optional. If coded, the suggested ICD-10 code for ear tags is Q17.0 (preauricular appendage or tag). Although "NOS" is a valid code in the ICD-10, it is used only when there is no possibility of obtaining a better description for a specific congenital anomaly.

Example 4

The following diagnosis and clinical description are provided for a neonate:

"amelia upper and lower limbs"

There are two ICD-10 codes to be assigned. One is for amelia of upper limbs: Q71.0 (congenital complete absence of the upper limb(s)); the other is for amelia of lower limbs: Q72.0 (congenital complete absence of lower limb(s)). However, for reporting and analytical purposes, it is suggested to count them only once, using the ICD-10 code Q71.0.

Example 5

The following diagnosis and clinical description are provided as part of an autopsy report:

"anencephaly infant with gross abnormalities; bilateral cleft lip; cleft palate"

The suggested ICD-10 code for anencephaly is Q00.0 (anencephaly). The "gross abnormalities" description is vague, and coding is optional. If coded, the suggested ICD-10 code is Q89.9 (congenital malformation, unspecified). 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 suggested ICD-10 code is Q37.8 (unspecified cleft palate with bilateral cleft lip).

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.

Example 6

The following diagnosis and clinical description are provided for a neonate:

"myelomeningocele, T3–T4 open"

Since it is not mentioned or specified whether hydrocephalus is present or not, one can assume that the defect is "spina bifida without hydrocephalus" and code as Q05.6 (thoracic spina bifida without hydrocephalus). However, it is also 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 birth defect surveillance programme include information in its protocol on how to code spina bifida when hydrocephalus is not mentioned or described in medical records.

The ICD-10 *(12)* and references *(37–40)* provide more information on coding and classification of congenital anomalies.

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Glossary of terms

Abnormality or anomaly: a deviation or departure from what is typical.

Abstraction: the act or process of extracting necessary information from hospital logs or medical records for the identification and classification of congenital anomalies in a case.

Abstraction form or recording form: a tool or instrument used in data collection.

Acalvaria: absence of bones of the calvarium with normal skull base, normal facial bones and intact scalp.

Acephaly: a term that is inappropriately used occasionally to refer to anencephaly; its meaning – absence of the head – is more correctly applied to the description of acardiac twins.

Amelia: congenital complete absence of an upper or lower limb.

Amnion: the inner of the two fetal membranes that form the amniotic sac, which surrounds the embryo or fetus.

Amniotic band: strands of the amniotic sac tissue that entangle limbs or other parts of the fetus, causing disruption of the affected areas.

Amniocentesis: a medical procedure used to remove a small amount of fluid from the sac that surrounds the fetus in the uterus; it is used most often to: (i) diagnose chromosomal or other genetic disorders early in the second trimester of pregnancy; and (ii) determine fetal lung maturity before birth.

Amniotic cavity or sac: the fluid-filled cavity that surrounds the developing embryo or fetus.

Amyelencephaly: a rarely used synonym of complete anencephaly or holoanencephaly.

Anencephaly: a neural tube defect characterized by partial or complete absence of the brain and skull *(14)*.

Anomaly: a deviation from the norm.

Arthrogryposis: a multiple, nonprogressive congenital joint contracture in two or more body areas *(41)*.

Arnold–Chiari malformation: a malformation of the brain consisting of downward displacement of the cerebellar tonsils through the foramen magnum.

Ascertaining: in birth defects surveillance, the process of identifying embryos, fetuses, neonates, infants and children who have a congenital anomaly, using existing sources and case definitions.

Association: in birth defects surveillance, a pattern of multiple anomalies that occur with a higher than random frequency, and that is not a sequence or a syndrome.

Autopsy: a postmortem examination to determine the cause of death.

Birth defect: see **Congenital anomaly**.

Birth outcome: a group of indicators that help measure the health and well-being of a neonate.

Birth weight: the first weight of the fetus or neonate obtained after birth; for live births, birth weight can be measured within the first hour of life before postnatal weight loss has occurred; actual weight is recorded to the degree of accuracy by which it is measured *(12)*.

- **Low birth weight**: less than 2500 g, up to and including 2499 g.
- **Very low birth weight**: less than 1500 g, up to and including 1499 g.
- **Extremely low birth weight**: less than 1000 g, up to and including 999 g.

Brachydactyly: a shortening of the fingers and/or toes; at least 13 clinically and genetically distinct groups have been identified.

British Paediatric Association (BPA): see Royal College of Paediatrics and Child Health.

Burden of disease: a time-based measure that combines years of life lost due to premature mortality and years of life lost due to time lived in states of less than full health.

Capture: when used in the context of surveillance, indicates that a case has been identified, abstracted and coded.

Case: in epidemiological terms, an individual who meets the criteria for inclusion in a surveillance programme. *Note*: although this term is not commonly used in clinical settings when referring to a patient, it is a term that is widely used in epidemiology.

Case definition: the criteria used for inclusion of a case in a surveillance programme.

Catchment area: a defined population from which cases for surveillance are collected.

Caudal dysgenesis: a developmental anomaly characterized by abnormalities of the lumbar and sacral vertebrae, hypoplasia of the pelvis and lower extremities, and anal abnormalities.

Central nervous system: the part of the nervous system consisting of the brain and the spinal cord.

Chorion: the outer of the two fetal membranes that form the amniotic sac, which surrounds the embryo or fetus.

Chorionic villus sampling (CVS): a medical procedure done late in the first trimester of pregnancy that removes a small piece of placental tissue (chorionic villi) to detect chromosomal abnormalities and other genetic disorders in the fetus.

Chromosomal abnormality: the excess or absence (whether total or partial) of a chromosome, or structural changes in the chromosome that most commonly produce a set of intellectual and physical problems (congenital anomalies).

Cleft lip: a partial or complete fissure of the upper lip; it can be either unilateral or bilateral, and can be associated with a cleft of the gum.

Cleft palate: Fissure of the palate, which can affect the soft and hard palate, or only the soft palate.

Cleft palate with cleft lip: an association of a unilateral or bilateral cleft of the upper lip with a fissure of the secondary palate (the hard and soft palate posterior to the incisive foramen).

Clubfoot, positional: a normal foot that has been held in an abnormal position in utero and on examination of the neonate is found to be flexible and amenable to moving into a normal position.

Clubfoot secondary to neuromuscular conditions: rigid clubfoot associated with spina bifida, arthrogryposis, myotonic dystrophy and other conditions.

Clusters: an unusual combination, whether real or apparent, of health events that are grouped in time or space, or both.

Confidentiality: an individual's right to have his/her personal, identifiable medical information kept secure.

Congenital: a condition that occurs during intrauterine life and that might be evident at birth or later in life; it may or may not be genetic.

Congenital anomaly: a structural or functional anomaly of organs, systems or parts of the body that occurs during intrauterine life and is caused by genetic or environmental factors (e.g. exposure to toxic substances, micronutrient deficiencies or maternal diseases), or both.

Consanguinity: the relationship among people who descend from a common ancestor.

Craniorachischisis: anencephaly with a contiguous spine defect without skin and meninges covering the neural tissue (rachischisis); it can be limited to the cervical region or affect the entire spine.

Deformation: the abnormal form, shape or position of a part of the body caused by mechanical forces; these forces affect structures after their initial development.

Developmental toxicant: a chemical that causes adverse effects on the developing organism, including death, structural abnormality, altered growth and functional deficiency, or any combination thereof.

Disability: a restriction or lack of ability (resulting from an impairment) to perform an activity in the manner or within the range considered normal for a human being *(12)*.

Disruption: a structural defect of an organ, part of an organ or a larger region of the body, resulting from the extrinsic breakdown of, or an interference with, an originally normal developmental process.

Dysplasia: an abnormal organization of cells into tissue(s) and its morphologic results, which most often affect skin, brain, cartilage or bone.

Embryo: the term given to the product of conception from implantation through the first 8 weeks after conception (equivalent to 10 weeks of gestation computed from the day of the last menstrual period).

Embryology: the branch of biology and medicine concerned with the study of prenatal development.

Encephalocele: a pedunculated or sessile cystic lesion protruding through a defect in the skull; it can contain herniated meninges and brain tissue (encephalocele or meningoencephalocele) or only meninges (cranial meningocele); the vast majority of these defects are covered by skin.

Epidemiology: the study of the frequency and distribution of health events and their determinants among human populations, and the application of such research to the prevention and control of health problems.

Epispadias: a congenital defect in which the urethra opens on the upper surface (dorsum) of the penis.

Exclusion criteria: the specific factors or characteristics that define an individual and that are not considered as a case.

External congenital anomaly: a type of anomaly that can be identified by inspection during physical examination.

Fetal death: a fetus that is deceased at delivery; fetal death refers to death prior to the complete expulsion or extraction of a product of conception from its mother, irrespective of the duration of pregnancy; the death is indicated by the fact that, after such separation, the fetus does not breathe or show any other evidence of life, such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles *(12)*.

Folic acid: the synthetic form of vitamin B_9 used in fortified foods and supplements; folic acid is more bioavailable than the natural form – folate – found in foods.

Gastroschisis: a congenital fissure of the anterior abdominal wall lateral to the umbilicus, accompanied by herniation of the small intestine and part of the large intestine, and occasionally other abdominal organs.

Gestational age: the time elapsed, measured in weeks, since conception. Because the exact date of conception is not always known, gestational age may also be defined as the time elapsed from the first day of the woman's last normal menstrual cycle. The duration of a normal pregnancy can range from 38 to 42 weeks. Gestational age is frequently a source of confusion when calculations are based on menstrual dates; for the purposes of calculation of gestational age from the date of the first day of the last normal menstrual period and the date of delivery, it is borne in mind that the first day is day zero and not day one; days 0–6 therefore correspond to "completed week zero"; days 7–13 to "completed week one"; and the 40th week of actual gestation is synonymous with "completed week 39"; where the date of the last normal menstrual period is not available, gestational age is based on the best clinical estimate; in order to avoid misunderstandings, tabulations are indicated both weeks and days *(12)*.

- **Pre-term or premature**: less than 37 completed weeks (less than 259 days) of gestation.
- **Term**: from 37 completed weeks to less than 42 completed weeks (259 to 293 days) of gestation.
- **Post-term**: 42 completed weeks or more (294 days or more) of gestation.

Gum: the mucosal tissue surrounding the maxilla and mandible.

Health risk: the likelihood of suffering ill-health, disease or an adverse effect.

Hemianencephaly: see **Hemicephaly**.

Hemicephaly: a rarely used synonym of incomplete **anencephaly** or **meroanencephaly**.

Histogenesis: the differentiation of cells into the specialized tissues forming the various organs and parts of the body.

Holoanencephaly: a rarely used term to describe a type of anencephaly characterized by the bone defect extending through the foramen magnum, affecting the entire skull.

Holoprosencephaly: a malformation of the forebrain commonly associated with severe central cleft lip and premaxillary agenesis.

Hospital-based surveillance programme: a programme aimed at capturing all birth outcomes with congenital anomalies that occur in selected birthing hospitals. This approach can be useful in locations in which most births occur in hospital settings and a population-based surveillance programme is not feasible.

Hypoplasia: the underdevelopment or incomplete development of a tissue or organ.

Hypospadias: a common congenital defect of the male external genitalia in which the urethral meatus opens in the ventral side (underside) of the penis.

Incidence: the number of new cases of a disease among a given population and over a given time frame; not used when reporting congenital anomalies (see Prevalence).

Inclusion criteria: the specific factors or characteristics that define a case.

Infant mortality: a demographic indicator that shows the number of deaths among children in their first year of life out of every 1000 live births registered.

Infancy period: the time from birth to one year of age.

Informed consent: an agreement to participate in a study or procedure after receiving and understanding full disclosure of the risks and benefits of participation.

Iniencephaly: a rare and complex neural tube defect involving the occiput and inion, resulting in extreme retroflexion of the head, variably combined with occipital encephalocele or rachischisis of the cervical and thoracic spine; in iniencephaly, the cranium is always closed, which helps to differentiate iniencephaly from cases of anencephaly with spinal retroflexion.

Inion: the most prominent projecting point of the occipital bone at the base of the skull.

Intercalary limb deficiency: the complete or partial absence of proximal or middle segment(s) of a limb, with all or part of the distal segment present.

Internal congenital anomaly: an anomaly that requires imaging techniques, surgery, autopsy or other specialized procedures to detect.

International Clearinghouse on Birth Defects Surveillance and Research (ICBDSR): an international non-profit organization affiliated with WHO, whose mission is to bring together birth defects programmes from around the world, with the aim of conducting worldwide surveillance and research to prevent birth defects and to ameliorate their consequences.

***International statistical classification of diseases and related health problems,* 10th revision (ICD-10)**: the standard diagnostic classification tool for epidemiology, health management and clinical purposes. It includes an analysis of the general health situation of population groups and monitoring of the incidence and prevalence of diseases and other health problems in relation to other variables, such as the characteristics and circumstances of the individuals affected, reimbursement, resource allocation, quality and guidelines *(12)*.

Isolated anomaly: a single anomaly; most (about 75% in the aggregate) congenital anomalies present as an isolated anomaly. Occasionally, an isolated major anomaly is associated with one or more minor anomalies.

Limb deficiency: an anomaly in limb development, characterized by the total or partial absence or different degrees of hypoplasia and abnormal shape of the skeletal structures of the limbs.

Limb–body wall complex: a complex anomaly involving lateral body wall defects, limb reduction defects, and occasionally neural tube defects, heart defects and other anomalies.

Live birth: the complete expulsion or extraction of a product of conception from its mother, irrespective of the duration of the pregnancy, which, after such separation, breathes or shows any other evidence of life such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles, whether or not the umbilical cord has been cut or the placenta is attached. Each product of such a birth is considered live born *(12)*.

Logic model: a visual element depicting how a programme operates, including the theories and assumptions underlying the programme; a logic model links outputs (both short and long term) with programme activities and the theoretical assumptions of the programme.

Longitudinal limb deficiency: the partial absence of a limb bone or segment extending parallel with the long axis of the limb and involving the pre-axial, post-axial or central components.

Malformation: a structural defect of an organ, part of an organ, or a larger region of the body that arises during organogenesis (initial formation of a structure). For most organs, organogenesis takes place during the first 8 weeks after fertilization; the resulting structure can be abnormally formed or incompletely formed, or may fail to form altogether. The term is occasionally used, incorrectly, as a synonym for birth defect.

Major congenital anomaly: a structural change that has significant medical, social or cosmetic consequences for the affected individual; this type of anomaly typically requires medical intervention.

Meninges: the membranes covering the brain and spinal cord.

Meningomyelocele: the most common type of spina bifida, constituting about 90% of all cases. It consists of a protrusion of the meninges and the spinal cord through an opening in the vertebral column, and most frequently is located in the lumbosacral area. It also is referred to as myelomeningocele.

Meningocele: a type of spina bifida characterized by herniation of the meninges through a spine defect, forming a cyst filled with cerebrospinal fluid. It does not contain the spinal cord, but can have some nerve elements.

Meroanencephaly: a rarely used term to describe a type of anencephaly characterized by the bone defect being limited to the anterior part of the skull.

Microcephaly: a disorder in which the head circumference is two standard deviations or more smaller than the average for sex and age, associated with microencephaly and, in some cases, with altered structure of the brain and neurodevelopmental problems; the presence of a head circumference less than two standard deviations below the mean for sex and age without evidence of structural abnormalities of the brain is not considered a major anomaly.

Midline cleft of the upper and/or lower lip: vertical cleft in the centre of, more commonly, the upper lip; the prevalence is low and it is usually part of a syndrome.

Minor congenital anomaly: a structural change that poses no significant health problem and tends to have limited social or cosmetic consequences for the affected individual.

Miscarriage: a spontaneous loss for a clinical pregnancy before 20 completed weeks of gestational age (18 weeks after fertilization) or, if gestational age is unknown, the loss of an embryo or fetus of <400 g *(42)*.

Monitor: in birth defects surveillance, to watch, observe or check for the presence of congenital anomalies or diseases over a period of time.

Morbidity: the incidence or prevalence of a disease, or of all diseases in a population, in a given space and over time *(43)*. Morbidity is an important statistic in understanding the evolution, progress or regression of a disease, as well as the reasons for its appearance and potential solutions.

Mortality rate: a demographic indicator that shows the number of deaths within a population per each 1000 inhabitants during a given time frame (generally one year).

Multifactorial: arising through the action of many factors; in genetics, arising as the result of the interaction of several genes, and usually non-genetic (environmental) factors.

Mutation: a permanent change in the DNA sequence of a gene.

Myelocele or myeloschisis: a type of spina bifida in which the open spinal cord, covered by a thin membrane, protrudes through a defect in the vertebral column.

Neonatal period: the period that commences at birth and ends 28 completed days after birth.

Neonatal death: deaths among live-born infants during the first 28 completed days of life; neonatal deaths can be subdivided into early neonatal deaths, occurring within the first 7 days of life, and late neonatal deaths, occurring after 7 but before 28 completed days of life; the age at death during the first day of life (day zero) is recorded in units of completed minutes or hours of life; for the second (day one), third (day two) and through 27 completed days of life, the age at death is recorded in days *(12)*.

Neonate: any live-born infant.

Neural tube: the part of the embryo from which the brain and spinal cord develop.

Neural tube closure: process by which the neural folds fuse to form the neural tube; it occurs within the first 28 days after conception.

Neural tube defect: a failure of the neural tube to close correctly.

Oblique facial clefts: the term given to orofacial clefts, which fall into four groups based on their position: midline clefts, paramedian clefts, orbital clefts and lateral clefts.

Oligohydramnios: a diminished amount of amniotic fluid.

Omphalocele: a congenital defect of the anterior abdominal wall in which the herniated intestines and abdominal organs are usually covered by a membrane consisting of peritoneum and amnion. 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.

Organogenesis: the process through which the ectoderm, endoderm and mesoderm organize to develop the organs and systems of the body.

Orofacial cleft: the term used to refer to a cleft palate, a cleft lip or both.

Pathogenesis: the mechanisms or cellular events in the development of a pathologic condition or disease.

Perinatal period: the period that commences at 22 completed weeks (154 days) of gestation (the time when birth weight normally is 500 g), and ends seven completed days after birth *(12)*.

Phocomelia: an intercalary limb defect that refers to the congenital absence of an arm and forearm with the hand present, or the absence of a thigh and lower leg with the foot present.

Policy-maker: a person who determines or influences policies and practices.

Polymorphism: variations in the DNA sequence of a gene or in the structure of a chromosome that have no adverse effects on the individual and are not due to new mutations. They occur with a frequency of at least 1% among the general population.

Population-based surveillance programme: a collection of data about a population residing in a defined geographical area.

Preconception care: health care received before a woman becomes pregnant, with the purpose of helping reduce her risk for adverse pregnancy outcomes.

Prenatal screening: a systematic search for a specific condition among a large, asymptomatic subpopulation of pregnant women selected by personal or family history, or by demographic characteristics such as age and ethnicity; typically, it identifies at-risk groups for further diagnostic testing.

Pregnancy outcome: the result of conception and ensuing pregnancy, including live birth, stillbirth, spontaneous abortion and induced abortion.

Prevalence: a measure of the total number of existing cases of a condition, known as prevalent cases, for a given point in time or period, and among a given population, regardless of whether or not they are new cases; also an indicator of the magnitude of the occurrence of a disease or other health event in the population.

In birth defects epidemiology, the terms live birth prevalence, birth prevalence and total prevalence are used:

- **Live birth prevalence of congenital anomalies:** measures the number of cases with congenital anomalies among live births and is defined as number of cases of live births with any congenital anomaly (numerator) among a defined cohort of live births (denominator). For example, the live birth prevalence of congenital anomalies in 2014 is computed as live births born with any congenital anomaly in 2014 divided by all live births born in 2014.

- **Birth prevalence of congenital anomalies:** measures the number of cases with congenital anomalies among live births and fetal deaths (stillbirths), and is defined as number of cases of live births and fetal deaths (stillbirths) with any congenital anomaly (numerator) among a defined cohort of live births plus fetal deaths (stillbirths) (denominator). For example, the birth prevalence of congenital anomalies in 2014 is computed as live births plus fetal deaths (stillbirths) with any congenital anomaly in 2014 divided by all live births plus fetal deaths (stillbirths) in 2014.

- **Total prevalence of congenital anomalies:** measures the number of cases with congenital anomalies in live births, fetal deaths (stillbirths), plus elective terminations of pregnancy for fetal anomaly, and is defined as number of cases of live births, fetal deaths (stillbirths), elective terminations of pregnancy for fetal anomaly (numerator) among a defined cohort of live births, fetal deaths (stillbirths), and elective terminations (denominator). For example, the total birth prevalence of congenital anomalies in 2014 is computed as live births and fetal deaths (stillbirths) with any congenital anomaly plus elective terminations of pregnancy for fetal anomaly in 2014 (numerator) divided by all live births and fetal deaths (stillbirths) in 2014 plus all elective terminations of pregnancy for fetal anomaly occurring in 2014.

Primary palate: the front part, anterior to the incisor foramen, of the shelf separating the oral and nasal cavities, which is formed during early embryonic development.

Privacy: an individual's right to control the acquisition, use and disclosure of his or her identifiable health information.

Pseudocleft: a rare congenital anomaly that has the appearance of a cleft lip corrected in utero; it is also known as congenitally healed cleft lip.

Public health: the discipline responsible for protecting the health of a population; its purpose is to improve population health and to control and eradicate diseases.

Public health surveillance: the systematic, continuous, timely and reliable collection of relevant and necessary data regarding certain health conditions among a population; analysis and interpretation of the data must provide grounds for decision-making and be disseminated.

Reproductive age: the age at which a woman is biologically capable of becoming pregnant. WHO characterizes this as being 15 to 49 years of age.

Risk factor: a characteristic, attribute, circumstance or exposure that is detectable among individuals or groups and is associated with an increased likelihood of a disease, congenital anomaly or other health problem.

Royal College of Paediatrics and Child Health: formerly known as the British Paediatric Association (BPA); developed an adaptation of the ICD-10 by adding an extra digit to the ICD-10 codes, to expand, and allow for more detailed, coding.

Secondary palate: the roof of the mouth posterior to the incisor foramen; the front, bony part is known as the hard palate, and the back part, consisting of muscular tissue and mucous membrane, as the soft palate.

Security: the technological and administrative safeguards and practices designed to protect data systems against unwarranted disclosure, modification or destruction.

Sentinel surveillance programme: a collection of data generally set up at one or a few sites, to obtain rapid estimates of the occurrence of a birth outcome.

Sequence: a pattern of multiple anomalies derived from a single known or presumed primary anomaly or mechanical factor. It represents a cascade of events that are consequences of a single primary malformation, disruption or deformation, and is considered an isolated anomaly, except when it is part of a syndrome.

Single-gene defect: a change (**mutation**) in the structure of a specific gene.

Sirenomelia: a lethal pattern of congenital anomaly, consisting of underdevelopment of the caudal pole of the body, characterized by fusion of the legs, absence of the sacrum, kidney agenesis, abnormal genitalia and imperforate anus.

Spina bifida: a general term used to describe a congenital defect of the spine caused by a failure of the posterior elements of the vertebrae to close, resulting in exposure of the meninges, with or without associated spinal cord herniation. It is most often located in the lumbar or sacral portion of the spine, and usually affects two or three vertebrae, although sometimes more vertebrae may be affected.

Spina bifida occulta: a relatively common anomaly that affects the spinous process and lamina of the posterior process, usually at the level of the fifth lumbar or the first sacral vertebra, and is covered by the skin. It is a relatively common anomaly that affects the spinous process and lamina of the posterior process, usually at the level of the fifth lumbar or first sacral vertebra, and is covered by skin. Spina bifida occulta is not considered a major congenital anomaly.

Stakeholder: an individual who is involved in or affected by a course of action.

Stillbirth: WHO defines stillbirth as third trimester fetal death (1000 grams or more; 28 weeks or more) for international comparison purposes. However, in broader terms, a stillbirth is a fetal death after the gestational age of viability. The definition of viability is based on gestational age and/or weight, and is variable among countries.

Submucous cleft: a midline notch, covered by mucosa, in the bony segment of the secondary palate.

Surveillance programme: a public health programme that collects, monitors, analyses, interprets and disseminates data systematically in a timely manner, and that allows for planning, implementation and evaluation of health strategies.

Syndrome: a pattern of multiple anomalies thought to be pathogenetically related and not representing a sequence; it is due to a single cause – genetic or environmental – or to gene–environment interactions.

Talipes equinovarus: a deformity involving one or both feet, consisting of malalignment of the calcaneotalar–navicular complex.

Transplacentally: passing through, or occurring across, the placenta.

Transverse limb deficiency: the complete or partial absence of distal structures of a limb in a transverse plane at the point where the deficiency begins, with proximal structures being essentially intact.

Trend: the general tendency in a set of data.

Teratogen: an agent capable of interrupting or altering the normal development of an embryo or fetus, often resulting in a congenital anomaly or embryonic or fetal death.

United States Centers for Disease Control and Prevention (CDC): a leading health protection agency, based in the USA, that collaborates with partners throughout the nation and the world to create expertise, information and tools that people and communities need to protect their health through health promotion, prevention of disease, injury and disability, and preparedness for new health threats.

Urethral meatus: the external opening of the urethra.

Uvula, absence: congenital absence of the uvula is a minor anomaly occasionally seen as an isolated defect and, more frequently, in association with submucous cleft palate.

Uvula, cleft: a common minor anomaly in which the uvula is totally or partially bifurcated.

Validation: in surveillance, a process to evaluate surveillance data, using a quality control protocol that covers the integrity, consistency, uniformity and reliability of the data.

Vital records: records of life events kept under governmental authority, including fetal death certificates, birth certificates, adoption records, legitimation, marriages, divorces and death certificates.

World Health Organization (WHO): the directing and coordinating authority for health within the United Nations system, responsible for providing leadership on global health matters, shaping the health research agenda, setting norms and standards, articulating evidence-based policy options, providing technical support to countries, and monitoring and assessing health trends.

Appendix A

Suggested initial list of congenital anomalies to consider for monitoring and relevant ICD-10 codes

Congenital anomaly	ICD-10 code *(12)*
Congenital malformations of the nervous system	
Anencephaly	Q00.0
Craniorachischisis	Q00.1
Iniencephaly	Q00.2
Encephalocele	Q01.0–Q01.2, Q01.8, Q01.9
Spina bifida	Q05.0–Q05.9
Cleft lip and cleft palate	
Cleft palate	Q35, Q35.1, Q35.3, Q 35.5, Q35.59, Q35.9, Q87.0
Cleft lip	Q36, Q36.0, Q36.9, Q36.90, Q36.99
Cleft palate with cleft lip	Q37, Q37.0, Q37.1, Q37.2, Q37.3, Q37.4, Q37.5, Q37.8, Q37.9, Q37.99
Congenital malformations of genital organs	
Hypospadias	Q54.0–Q54.4, Q54.8, Q54.9
Congenital malformations and deformations of the musculoskeletal system	
Talipes equinovarus	Q66.0, Q66.1, Q66.4, Q66.8
Reduction defects of upper limb	Q71.0–Q71.6, Q71.8, Q71.9
Reduction defects of lower limb	Q72.0–Q72.9
Reduction defects of unspecified limb	Q73.0, Q73.1, Q73.8
Exomphalos/omphalocele	Q79.2
Gastroschisis	Q79.3

Appendix B

External minor congenital anomalies

Congenital anomaly	ICD-10 *(12)* or RCPCH code *(34)*
Eye	
Congenital ectropion	Q10.1
Congenital entropion	Q10.2
Absence of eyelashes	Q10.3
Dystopia canthorum	Q10.3
Epicanthal folds	Q10.3
Epicanthus inversus	Q10.3
Fused eyelids	Q10.3
Long palpebral fissure(s)	Q10.3
Upward or downward slanting palpebral fissures	Q10.3
Short palpebral fissures	Q10.3
Long eyelashes	Q10.3
Weakness of eyelids	Q10.3
Stenosis or stricture of lacrimal duct	Q10.5
Coloboma of iris	Q13.0
Brushfield spots	Q13.2
Iris freckles	Q13.2
Blue sclera	Q13.5
Exophthalmos	Q15.8
Strabismus	Q15.8
Ear	
Accessory tragus	Q17.0
Auricular tag or pit	Q17.0
Double ear lobule	Q17.0
Ear pit or tag	Q17.0
Preauricular appendage, tag or lobule	Q17.0
Large ears	Q17.1
Macrotia	Q17.1

Absent tragus	Q17.3
Asymmetric sized ears	Q17.3
Crumpled ears	Q17.3
Cup ear	Q17.3
Ear lobe crease	Q17.3
Ear lobe notch	Q17.3
Lack of helical fold	Q17.3
Lop ear	Q17.3
Misshapen ears	Q17.3
Pointed ear	Q17.3
Primitive shape of ear	Q17.3
Protruding ears	Q17.3
Simple ear	Q17.3
Small ears (excludes true microtia)	Q17.3
Thickened or overfolded helix	Q17.3
Low-set ears	Q17.4
Misplaced ear	Q17.4
Posteriorly rotated ears	Q17.4
Bat ear	Q17.5
Prominent ears	Q17.5
Darwinian tubercle	Q17.8
Narrow external auditory meatus	Q17.8
Face and neck	
Branchial vestige	Q18.0
Branchial tag or pit	Q18.0
Fistula of auricle, congenital and fistula cervicoaural	Q18.1
Pretragal (commonly referred to as preauricular) sinus and cyst	Q18.1
Redundant neck folds	Q18.3
Webbed neck (pterygium colli)	Q18.3
Macrostomia	Q18.4
Microstomia	Q18.5
Hypertrophy of lip, congenital or macrocheilia or large wide lips	Q18.6
Short or long columella	Q18.8

Angular lip pits	Q18.8
Small lips	Q18.8
Short neck	Q18.8
Thin vermilion border	Q18.8
Synophrys, confluent or medial flare eyebrows	Q18.80
Peripheral vascular system	
Single umbilical artery	Q27.0
Nose	
Notched or hypoplastic alae nasi	Q30.2
Anteverted nares	Q30.8
Flat or wide nasal bridge	Q30.8
Small nares	Q30.8
Smooth philtrum	Q30.8
Mouth	
Cleft uvula	Q35.7
Tongue tie	Q38.1
Macroglossia	Q38.2
Adhesion of tongue	Q38.3
Bifid tongue	Q38.3
Fissure of tongue	Q38.3
Hypoglossia	Q38.3
Hypoplasia of tongue	Q38.3
Microglossia (hypoplasia of tongue)	Q38.3
Ranula	Q38.4
Absent uvula	Q38.5
High arched palate	Q38.50
Aberrant frenula	Q38.6
Broad alveolar ridge	Q38.6
Cleft gum (in the absence of cleft lip)	Q38.6
Natal teeth	Q38.6
Anus and genitalia	
Anterior anus (ectopic anus)	Q43.5
Imperforate hymen	Q52.3

Embryonal cyst of vagina	Q52.4
Fusion of labia	Q52.5
Fusion of vulva	Q52.5
Prominent clitoris	Q52.6 or Q52.8
Hypoplastic labia majora	Q52.8
Hypoplastic labia minora	Q52.8
Undescended testicle, unilateral	Q53.1
Undescended testicle, bilateral	Q53.2
Undescended testicle, unspecified	Q53.9
Chordee (without hypospadias)	Q54.4
Hypoplasia of testis and scrotum	Q55.1
Shawl scrotum	Q55.2
Retractile testis	Q55.20
Bifid scrotum	Q55.21
Absent or hooded foreskin of penis	Q55.6
Curvature of penis lateral	Q55.6
Phimosis	Q55.6
Redundant foreskin	Q55.6
Small penis (unless documented as micropenis)	Q55.6
Hydrocele of testis	Q55.8
Scrotalization of phallus	Q55.9
Foot	
Metatarsus varus or metatarsus adductus	Q66.2
Hallux varus	Q66.3
Congenital pes planus	Q66.5
Hallux valgus	Q66.6
Metatarsus valgus	Q66.6
Pes cavus	Q66.7
Hammer toe, congenital	Q66.8
Long toes	Q66.8
Prominent calcaneus	Q66.8
Prominent heel	Q66.8
Short great toe	Q66.8
Vertical talus	Q66.8

Widely spaced first and second toes	Q66.8
Recessed fourth and fifth toes	Q66.8
Short fourth metatarsus	Q66.8
Short or broad great toe	Q66.8
Rocker-bottom feet	Q66.8
Overlapping toe	Q66.8
Head, face, spine and chest	
Facial asymmetry	Q67.0
Compression facies	Q67.1
Dolichocephaly	Q67.2
Flat occiput	Q67.3
Head asymmetry	Q67.3
Plagiocephaly	Q67.3
Squashed or bent nose, congenital	Q67.4
Deviation of nasal septum	Q67.41
Funnel chest	Q67.6
Pectus excavatum	Q67.6
Congenital pigeon chest	Q67.7
Pectus carinatum	Q67.7
Barrel chest	Q67.8
Deformed chest	Q67.8
Prominent sternum	Q67.8
Shield-like chest	Q67.8
Other musculoskeletal (including limbs)	
Congenital deformity of sternocleidomastoid muscle	Q68.0
Contracture of sternocleidomastoid (muscle)	Q68.0
Congenital torticollis	Q68.0
Camptodactyly	Q68.1
Congenital clubfinger	Q68.1
Long fingers	Q68.1
Overlapping digits, not otherwise specified	Q68.1
Short fourth metacarpal	Q68.1
Single crease fifth finger	Q68.1

Tapered fingers	Q68.1
Short fingers	Q68.1
Clinodactyly	Q68.10
Genu recurvatum	Q68.21
Cubitus valgus	Q68.8
Hyperextended joints, not otherwise specified	Q68.8
Hyperextended knee	Q68.8
Polydactyly type B of fingers (type B is, by definition, post axial and rudimentary (postminimi); type A is postaxial, fully developed is a major anomaly – Q69.02A, and is not a minor anomaly)	Q69.02B
Polydactyly type B, not otherwise specified	Q69.02B
Polydactyly type B of toes	Q69.22B
Syndactyly (involving second and third toes)	Q70.3
Genu valgum	Q74.1
Other anomalies of skull, face and spine	
Scaphocephaly	Q75.0
Trigonocephaly, other head deformations without synostosis	Q75.0
Hypertelorism	Q75.2
Macrocephaly (includes familial benign macrocephaly)	Q75.3
Hypotelorism	Q75.8
Maxillary hypoplasia or prominence	Q75.8
Micrognathia	Q75.8
Prognathia	Q75.8
Frontal bossing	Q75.8 or Q75.80
Large or small fontanelles	Q75.8 or Q75.80
Metopic suture open to bregma	Q75.8 or Q75.80
Narrow bifrontal diameter	Q75.8 or Q75.80
Prominent occiput	Q75.8 or Q75.80
Prominent or hypoplastic supraorbital ridges	Q75.8 or Q75.80
Third fontanelle	Q75.8 or Q75.80
Spina bifida occulta	Q76.0
Congenital lordosis, postural	Q76.43

Abdomen	
Diastasis recti	Q79.5
Inguinal hernia	Q79.8
Umbilical hernia	Q79.8
Skin, breast and other integument	
Skin cyst	Q82.4 or Q84.4
Angioma	Q82.5
Benign skin neoplasm, pigmented naevus (ear and auditory canal)	Q82.5
Benign skin neoplasm, pigmented naevus (eyelid)	Q82.5
Benign skin neoplasm, pigmented naevus (face)	Q82.5
Benign skin neoplasm, pigmented naevus (lip)	Q82.5
Benign skin neoplasm, pigmented naevus (lower limb, hip)	Q82.5
Benign skin neoplasm, pigmented naevus (other specified site)	Q82.5
Benign skin neoplasm, pigmented naevus (scalp, neck)	Q82.5
Benign skin neoplasm, pigmented naevus (trunk)	Q82.5
Benign skin neoplasm, pigmented naevus (unspecified site)	Q82.5
Benign skin neoplasm, pigmented naevus (upper limb, shoulder)	Q82.5
Café-au-lait spot	Q82.5
Hemangioma (other than face and neck)	Q82.5
Lymphangioma	Q82.5
Noncavernous, single, small hemangioma (4 cm diameter)	Q82.5
Pigmented naevus, congenital non-neoplastic naevus	Q82.5
Birthmark	Q82.50
Naevus flammeus	Q82.51
Port-wine stain	Q82.51
Strawberry naevus	Q82.51
Mongolian spot	Q82.52
Cutis marmorata	Q82.8 or Q84.8
Dimple, hand	Q82.8 or Q84.8
Dimple, shoulder	Q82.8 or Q84.8
Extra or absent hand or interphalangeal creases	Q82.8 or Q84.8
Pilonidal or sacral dimple	Q82.8 or Q84.8
Plantar furrow	Q82.8 or Q84.8
Rectal fissure	Q82.8 or Q84.8

Sole crease	Q82.8 or Q84.8
Vaginal or hymenal tags	Q82.8 or Q84.8
Single transverse palmar crease	Q82.80
Anal tag	Q82.81
Skin tag	Q82.81
Unusual dermatoglyphics	Q82.84
Absent nipple	Q83.2
Extra nipples (supernumerary nipples)	Q83.3
Supernumerary nipple	Q83.3
Inverted nipples	Q83.8
Small nipple (hypoplastic)	Q83.8
Widely spaced nipples	Q83.8
Monilethrix	Q84.1
Pili annulati	Q84.1
Pili torti	Q84.1
Aberrant scalp hair patterning	Q84.1 or Q84.2
Depigmentary hair changes	Q84.1 or Q84.2
Hair upsweep	Q84.1 or Q84.2
Low posterior hairline	Q84.1 or Q84.2
Persistent lanugo	Q84.2
Congenital hypertrichosis	Q84.20
Absent nails (major if third phalanx is missing)	Q84.3
Enlarged or hypertrophic nails	Q84.5
Pachyonychia	Q84.5
Congenital clubnail, koilonychia, malformation of nail, not otherwise specified	Q84.6
Duplication of thumbnail	Q84.6
Hyperconvex fingernails	Q84.6
Hyperconvex toenails	Q84.6
Hypoplastic fingernails	Q84.6
Hypoplastic toenails	Q84.6
Thickened toenails	Q84.6
Aplasia cutis (major if large)	Q84.8

Appendix C

Causes of congenital anomalies and classification according to developmental mechanism and clinical presentation

Causes of congenital anomalies

It has been estimated that about one quarter of all congenital anomalies may have a genetic cause *(44)*. However, more recent estimates suggest the proportion could be higher, as advances in cytogenetic and molecular techniques in the last two decades are allowing the identification of previously undetected chromosomal abnormalities, gene mutations and genetic polymorphisms. The two most common genetic causes of congenital anomalies are single-gene defects and chromosomal abnormalities.

Single-gene defects are caused by changes (mutations) in the structure of genes. These are responsible for slightly over 17% of congenital anomalies (*44*). Single-gene defects may be inherited from either one or both parents, or be caused by a sporadic (new) mutation. Single-gene mutations seem to be associated more often with multiple congenital anomalies that are syndromic, rather than with isolated malformations, though new research is increasingly uncovering single-gene defects that cause isolated anomalies such as cleft lip with or without cleft palate and some types of congenital heart defects.

Abnormalities caused by chromosomal changes are identified in about 10% of children with congenital anomalies *(44)*, and may involve the autosomes or the sex chromosomes. Changes include numerical abnormalities such as having an extra chromosome – e.g. trisomies (Down syndrome or trisomy 21, trisomy 13 and trisomy 18); missing a chromosome – e.g. monosomies (monosomy X or Turner syndrome); and chromosomal structural abnormalities – deletions (e.g. deletion of the proximal region in the long arm of chromosome 22 associated with the DiGeorge and velocardiofacial syndromes) and duplications (e.g. duplication of the short arm of chromosome 9). Chromosomal abnormalities are almost always associated with patterns of multiple congenital anomalies.

Identified environmental and maternal causes are responsible for an estimated 4% to 10% of congenital anomalies *(45)*. Examples include:

- maternal nutritional status
- exposure to chemicals, and possibly illicit drugs
- maternal infections (e.g. rubella)
- physical factors, such as ionizing radiation and hyperthermia *(45)*
- chronic maternal diseases (e.g. diabetes)
- exposure to known teratogenic prescription medicines (e.g. retinoic acid, valproic acid) – for more information on medications see reference *(46)*.

For approximately 66% of congenital anomalies, the cause remains unknown *(45)*. This group includes those congenital anomalies that are believed to have environmental

causes or to be multifactorial. Multifactorial means that multiple undefined gene variants interact with environmental factors to cause a specific anomaly.

Many potential gene–environment interactions have been tested in relation to different congenital anomalies. For example, mutations and polymorphisms of numerous genes, including *TGFA, TGFB3, CYP1A1, NAT1, NAT2* and *GSTT1*, have been studied to determine their level of association with an increased risk for oral clefts in the offspring of women who smoke cigarettes *(47)*. Another example of a gene–environment interaction involves prenatal exposure to phenytoin, a widely used anticonvulsant drug. Phenytoin is associated with structural congenital anomalies in 3% to 10% of infants exposed to this medication in utero. It has been shown that the presence of congenital anomalies in these infants correlates with reduced activity of epoxide hydrolase, a microsomal enzyme that normally detoxifies phenytoin metabolites *(48)*. When the enzyme epoxide hydrolase is not working properly, some intermediate teratogenic metabolites do not get eliminated. This can result in a congenital anomaly in the developing fetus.

Congenital anomalies according to developmental mechanisms

Malformation

Malformation is a structural defect of an organ, part of an organ, or larger region of the body that arises during organogenesis, that is, during the initial formation of a structure, as a result of an intrinsically abnormal developmental process. For most organs, organogenesis takes place during the first 8 weeks after fertilization. The resulting structure may be abnormally formed or incompletely formed, or may fail to form altogether. Although the term malformation is occasionally used to refer to congenital anomalies, it is important to realize that congenital anomalies include more than malformations.

Disruption

Disruption is a structural defect of an organ, part of an organ, or larger region of the body, resulting from the extrinsic breakdown of, or an interference with, an originally normal developmental process. Examples of disruption defects include the amniotic band complex, some transverse limb deficiencies, and Moebius sequence (cranial nerve paralyses and limb and other abnormalities).

Dysplasia

Dysplasias refer to abnormalities of histogenesis or formation of tissues and most commonly affect skin, brain, cartilage or bone. Dysplasias may be localized (e.g. naevus) or generalized (e.g. achondroplasia and other chondrodysplasias, neurofibromatosis).

Deformation

Deformation is an abnormal form, shape or position of a part of the body, caused by mechanical forces. These forces affect structures after their initial development. Examples include intrauterine crowding as a result of twin pregnancies or uterine abnormalities, and oligohydramnios (diminished amniotic fluid) in bilateral renal agenesis leading to Potter sequence (i.e. distinctive facial findings, lung hypoplasia and some cases of clubfoot).

Congenital anomalies according to clinical presentation in a child

Isolated

Most major congenital anomalies (about 75%) occur in isolation, meaning that there are no other unrelated major congenital anomalies present. Frequently, isolated major anomalies are associated with one or more minor anomalies.

Sequence

A sequence is a pattern of related anomalies that are known, or presumed, to derive from a single primary anomaly or mechanical factor. A sequence represents a cascade of events (anomalies) that are consequences of a single primary malformation, disruption or deformation. Examples include the Robin sequence (in which, because of micrognathia, there is posterior displacement of the tongue, which interferes with closure of the palatal shelves, leading to cleft palate) and clubfoot associated with spina bifida. A sequence is considered as an *isolated* anomaly, except when it is part of a syndrome.

Multiple congenital anomaly

Multiple congenital anomaly is the occurrence of two or more major anomalies that are unrelated. This means that the major anomalies are presumed to be a random association, and do not constitute a sequence or a previously recognized syndrome. Most cases of multiple congenital anomalies fall into this category.

Association

Association is a pattern of multiple anomalies that occur with a higher than random frequency and that is not a sequence or a syndrome. Examples include the VACTERL association (**V**ertebral, **A**nal, **C**ardiac, **T**racheo–o**E**sophageal fistula, **R**enal, and **L**imb defects) and the MURCS association (**MU**llerian duct aplasia–**R**enal aplasia–**C**ervicothoracic **S**omite dysplasia). As knowledge and techniques advance, some of these entities may be recognized as syndromes. This was the case with the CHARGE association (**C**oloboma of the eye, **H**eart defects, **A**tresia of the choanae, **R**etardation of growth and/or development, **G**enital and/or urinary abnormalities, and **E**ar abnormalities and deafness) that was found in recent years to be caused by a mutation of the *CHD7* gene and is now considered to be a genetically determined syndrome *(49)*.

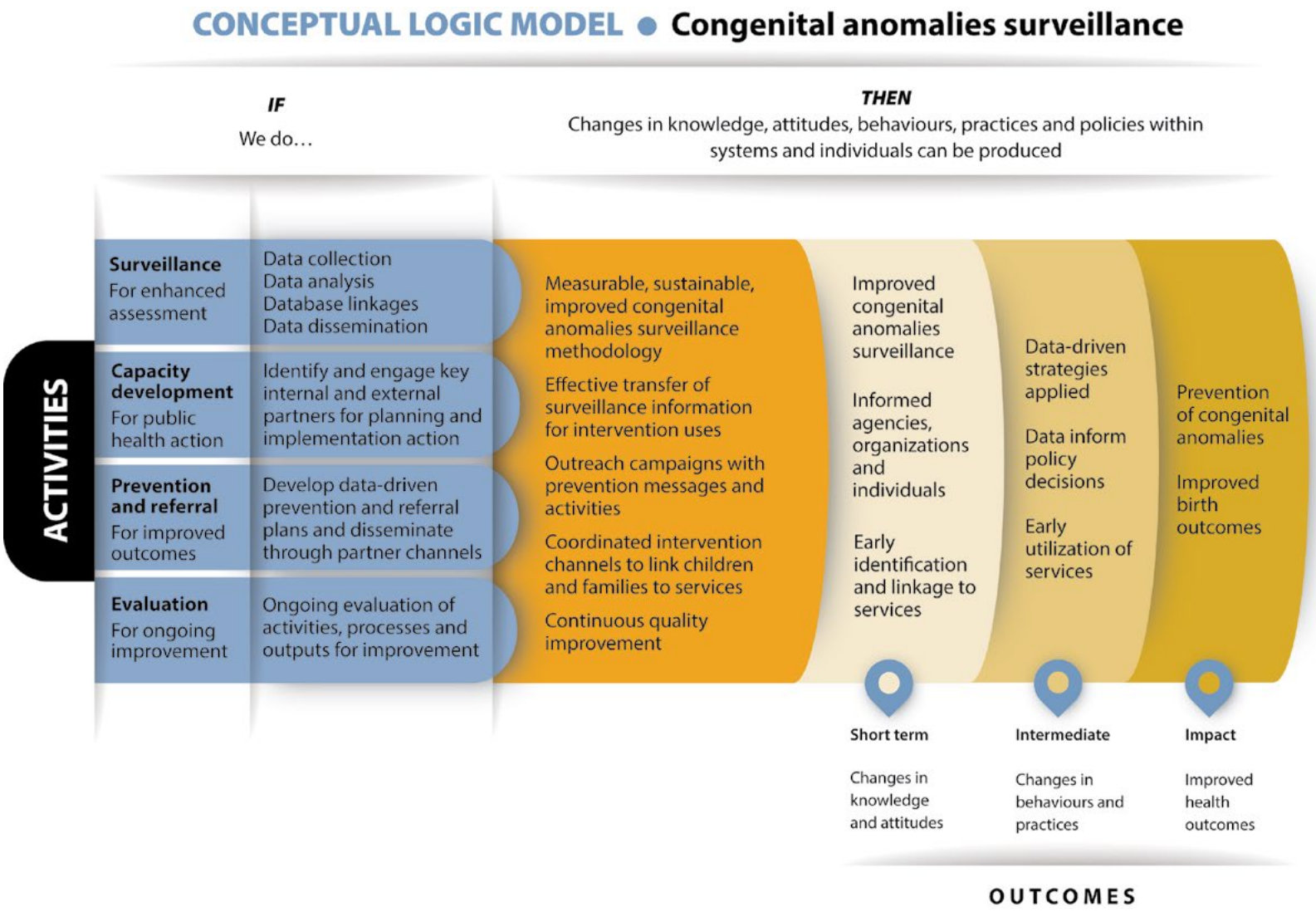
Syndrome

A syndrome is a pattern of multiple anomalies thought to be pathogenetically related, but not representing a sequence. They are due to a single cause – genetic or environmental – or to gene–environment interactions. Examples include Down syndrome (trisomy 21, a chromosomal abnormality), deletion of the proximal region in the long arm of chromosome 22 (a genomic disorder due to microdeletion), achondroplasia (single gene disorder), and congenital rubella syndrome (infectious cause). Despite advances in genetics, there are still clinically recognized syndromes for which the cause has not been identified.

For further information on case classification, please refer to reference *(50)*.

Appendix D

Sample logic model



Source: PRINCIPLES & PRACTICE OF PUBLIC HEALTH SURVEILLANCE edited by Lisa M. Lee et al. (2010) Ch. 12 "Public Health Surveillance for Chronic Diseases, Injuries, and Birth Defects" by Ali H. Mokdad et al., pp. 255–274, Figure 12.2 from p.270 (adapted), by permission of Oxford University Press Inc. *(51)*.

WORKING LOGIC MODEL ● Congenital anomalies logic model and process indicators

ACTIVITIES

Surveillance
Establish/enhance state-based congenital anomalies surveillance system

Data collection
Data analysis
Monitor and detect trends in congenital anomalies
Database linkages
Link to state databases
Data dissemination
Timely dissemination of data for utilization
Evaluation for completeness and timeliness

Capacity development
Build congenital anomalies capacity through infrastructure and partnership collaboration

Partner identification
Identify and engage internal and external partners
Action plan development
Develop data-driven prevention and referral action plan to reach target audiences

Prevention
Outreach to target audiences with prevention activities

Prevention messages and activities implemented
Disseminate prevention messages to general and target audiences through partner channels
Evaluation for ongoing improvement to address gaps, opportunities and reach target audiences

Prevention
Outreach to target audiences with prevention activities

Prevention messages and activities implemented
Disseminate prevention messages to general and target audiences through partner channels
Evaluation for ongoing improvement to address gaps, opportunities and reach target audiences

OUTPUTS

Measurable, sustainable, improved congenital anomalies surveillance methodology

Effective transfer of surveillance information for public health uses

Outreach campaigns with prevention messages and activities

Coordinated intervention channels for linking children and families to services

Continuous quality improvement

OUTCOMES

Improved congenital anomalies surveillance

Improved dissemination of accurate and timely information to organizations, agencies and individuals

Increased early identification and linkage to services

Data-driven strategies applied

Early utilization of services

Data inform policy decisions

Prevention of congenital anomalies

Improved birth outcomes

Improved quality of life

Indicators – develop and integrate evaluation measures into the key activities

- Quality and timely data are produced and disseminated
- Quality assurance for completeness of data tested through ongoing improvement efforts using statistical methods

- Matrix identifying capacity-building objectives, strategies and partner list is developed and approved
- Data-driven prevention and referral plans are developed through partnership engagement
- Ongoing partner meetings take place to exchange progress information and make mid-course modifications

- Data-driven list identifying at-risk populations is developed to guide prevention efforts
- Appropriate prevention partners are engaged and a plan to reach target audiences is developed
- Targeted audiences are reached using appropriate prevention/intervention strategies

- Referral protocols are tested for effectiveness and timeliness
- Baseline data are available to indicate changes in number of referrals and number of persons receiving early intervention and special education services
- Timely referral to services is evidenced
- Gaps in referrals are identified using appropriate methods (i.e. qualitative research – focus groups)

Appendix E. Worksheet for capacity development

Examples of potential partners	Surveillance	Referral	Prevention	Examples of potential roles
Ministries of health	X	X	X	Set policies and regulations for health-care services and delivery
Hospitals and, if applicable, hospital associations and clinics	X	X		Serve as data sources; referral sources
Regional and local health departments	X	X	X	Serve as data sources; conduits to audiences for referral and prevention activities
Primary health centres and health-care providers	X	X	X	Serve as data sources and also as sources for prevention and outreach activities
Community health workers/ community health volunteers	X	X	X	Serve as potential data sources because, in many countries, these individuals are present at the delivery; provide prevention information
Congenital anomalies associations, foundations, and other nongovernmental organizations	X		X	Provide advocacy for congenital anomalies infrastructure at national and local levels; serve as dissemination channels for prevention activities and messages; potential sources for outcome data; serve as possible data sources
International organizations	X	X	X	Provide advocacy, technical assistance and expertise
Medical schools/research agencies	X			Provide specialized laboratory services, such as chromosome analyses, or have clinics where individuals with congenital anomalies are seen; can help drive surveillance of congenital anomalies

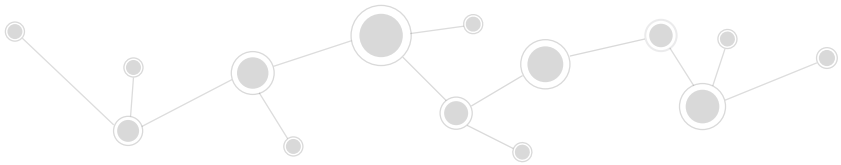
Appendix F

Suggestions for delivering the news of a congenital anomaly diagnosis to a family

Note: it is important to remember that abstractors, those individuals who will be extracting information from hospital logs or medical records for the identification and classification of congenital anomalies, do not give information to parents about a diagnosis or services. **This is to be done by a health-care provider**.

- Parents are told about the diagnosis as soon as possible, even if it is suspected but not yet confirmed.
- The diagnosis is communicated in person, by a health-care professional with sufficient knowledge of the condition. Health-care providers should coordinate the message to ensure consistency in the information provided to the family.
- Begin the conversation with positive words and avoid using value judgments when starting the conversation, such as "I'm sorry", or "Unfortunately, I have bad news". Parents remember the exact words of the first contact with the health-care provider even after many years.
- The family is informed of the diagnosis, treatment and prognosis, in their preferred language. If possible, a professional medical interpreter is present at the time of disclosure.
- Discuss the diagnosis, treatment and prognosis in a private, comfortable setting, free from interruptions. The infant may be present in the room unless he or she is ill. Allow time for questions and make plans for a follow-up conversation. Stop, when possible, to assess for comprehension.
- Parents should be provided with accurate and up-to-date information. Information is normally given with a balanced perspective, including both positive aspects and challenges related to the congenital anomaly.
- Provide the information on diagnosis, treatment and prognosis in a sensitive and caring, yet confident and straightforward manner, using understandable, non-medical terms, and language that is clear and concise.
- Use sensitive language and avoid outdated or offensive terminology. In the neonatal setting, the baby is to be present, and to be referred to by name.
- Assess for knowledge of that specific congenital anomaly, including etiology. Because there may often be guilt or blame associated with congenital anomalies, often placed on the mother, it is important to discuss these issues with the parents.
- Informational resources can be provided, including contact information for local and national support groups, up-to-date printed information or fact sheets, and books. When appropriate, referrals to other specialists may also be helpful (e.g. medical geneticists, genetic counsellors, cardiologists, neonatologists).

Source: suggestions modified from Sheets et al., 2011 *(52)*.



Appendix G

Sample abstraction form ▶

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:

Additional information for autopsy:

Additional information for congenital anomaly:

Appendix H

Potential core variables

The abstraction form can be modified according to the needs of each country. The explanation and instructions that follow can be reviewed accordingly.

The instructions for the abstraction form will help personnel participating in the congenital anomaly surveillance system to clarify doubts about how to fill in the form. Please review the variable column (third column) and the explanation column (fourth column) before completing the form.

Column 1: variable number; useful when designing the database

Column 2: different variable categories

Column 3: variable name

Column 4: instructions for completing the abstraction form for each particular variable

Variable number	Category	Variable name	Instructions
	Report		
1		Case record identification	Each case has a unique identification number. Each country can decide how to create the code, for example, the year and month the baby was born can be part of the unique identification.
2		Date of report	Indicate the date when surveillance staff complete the form; report the day, month and year.
3		Name of health facility	Indicate the name of the hospital where the fetus or neonate with a congenital anomaly was identified.
4		City, province, state or territory	Indicate the city, province, state or territory where the delivery took place.
	Father		
5	Identification information and demographic information	Name(s)	Indicate the father's given name(s) and family name(s), depending on what is commonly used in the country.

6		Father's date of birth, or age if date of birth is not available	Indicate the father's date of birth. If known, please follow the same system as used in the date of report: day, month and year. If only the year is available, use year; if only age is available, use age.
7		Race and ethnicity	Indicate the father's race and ethnicity, if applicable.
	Mother		
8	Identification information and demographic information	Name(s)	Indicate the mother's given name(s) and family name(s), depending on what is commonly used in the country. Make sure to include her maiden name.
9		Mother's date of birth, or age if date of birth is not available	Indicate the mother's date of birth. If known, please follow the same system as used in the date of report: day, month and year. If only the year is available, use year; if only age is available, use age.
10		Race and ethnicity	Indicate the mother's race and ethnicity, if applicable.
11		Primary address during pregnancy	Indicate the primary address for the mother during pregnancy.
12		Current address	Indicate the maternal residence at the time of delivery, such as department and municipality. Use available country coding.
13		Telephone number	Indicate the telephone number where the mother can be contacted.
	Obstetric history		
14		Total number of pregnancies	Indicate the total number of previous pregnancies: live births, stillbirths (fetal deaths), spontaneous abortions, and terminations of pregnancy.
	Fetus/neonate		
15	Identification information and demographic information	Name, if available	Indicate the fetus or neonate's given name and family name(s), depending on what is commonly used in the country.

16		Date of birth	Indicate the fetus or neonate's date of birth. If known, please follow the same system as used in the date of report: day, month and year.
17		Sex	Indicate the sex of the fetus or neonate, if it is known. In case of ambiguous genitalia, indicate as "ambiguous"; if sex cannot be determined during the autopsy, please indicate as "unknown".
18		Date of diagnosis	Indicate the date of diagnosis of congenital anomaly/anomalies; write the date as indicated: day, month and year.
19		Outcome at birth	Indicate whether pregnancy resulted in a live birth, fetal death, spontaneous abortion or termination of pregnancy.
20		Gestational age	Write the gestational age in weeks; estimate the number of weeks based on the first day of the last normal menstrual period or on a sonogram done during the first trimester, if the information is available.
21	Birth measurements	Weight	Register the fetus' or neonate's weight in grams. If the fetus or neonate was stillborn, document the weight (use grams if grams are used in the country).
22		Length	Register the length in centimetres and use a comma to separate decimals.
23		Head circumference	Register the head circumference in centimetres and use a comma to separate decimals.
24		Multiple birth (birth order)	Indicate if the birth was multiple and, if yes, the birth order of the fetus or neonate with a congenital anomaly/anomalies. Complete a form for each fetus or neonate if more than one has a congenital anomaly. Complete only one form if the babies are conjoined twins.

25	Photographs taken	If possible, take at least three photographs: (i) the whole fetus or neonate; (ii) the fetus or neonate's front and back; and (iii) the congenital anomaly/anomalies. Refer to Appendix J for information on taking photographs.
26	Parental consanguinity	Indicate any biological relationship between the parents.
27	If the neonate was born alive and died, include date of death	Write the date as indicated: day, month and year.
28	Autopsy results	Indicate if an autopsy was performed and if the autopsy findings add to the diagnosis of the congenital anomaly. This information can go on the back of the form.
29	Congenital anomaly/anomalies present	Write the name(s) of the congenital anomaly/anomalies; list all congenital anomalies present.
30	Describe in detail each congenital anomaly	Provide a full description for each congenital anomaly identified.
31	Code	Code the congenital anomaly according to the *International classification of disease and related health problems*, 10th revision *(12)*.
32	C or P	Indicate whether the diagnosis is confirmed (C) or possible (P) and whether more tests are needed.
33	Diagnostic tests performed or pending; notes and comments	Indicate what tests were performed or are needed. Include any other relevant comments.
Hospital information		
34	Name and profession of individual completing the form	Identify the name and the profession of the individual completing the form.
35	Contact information	Indicate a name and telephone number if more information is needed to complete the form.

Appendix I

Potential optional variables

Variable number	Category	Variable name	Explanation and instructions
1	**Report**	Source of information	Indicate the different data sources inside the hospital where a fetus or neonate with a congenital anomaly is identified (e.g. delivery room or surgery).
	Father		
2		Occupation/work	Code according to the 1988 *International standard classification of occupations*; see www.ilo.org/public/english/bureau/stat/isco/index.htm.
3		Family health history	Indicate if there is someone in the father's family with a congenital anomaly/anomalies, including the father himself.
	Mother		
4	Demographic information	Civil/marital status	Indicate if the mother is married, single, separated, living with someone but not married, or a widow.
5		Occupation/ work at conception	Code according to the 1988 *International standard classification of occupations*; see www.ilo.org/public/english/bureau/stat/isco/index.htm.
6		Country identification number	This corresponds to any legal document that identifies the mother in each country; use if available in country.
7		Weight (before pregnancy)	Indicate the mother's weight before the pregnancy in kilograms or pounds, according to what the country uses.
8		Education (years or highest level)	Indicate the highest level of education achieved by the mother. Refer to the *International standard classification of education* for 1997 for information; see http://www.unesco.org/education/information/nfsunesco/doc/isced_1997.htm.
9		Religion	Indicate the religious affiliation of the mother, if applicable.
10		Socioeconomic status	Indicate the socioeconomic status of the mother.
	Obstetric history		
11	Health	Chronic diseases	Indicate any illness the mother has (e.g. diabetes, epilepsy or infections).
		Date of last menstrual period	Indicate the first day of the last normal menstrual period. Follow the system: day, month and year.

12		Prenatal tests	Indicate if the mother received prenatal care services and at which month of pregnancy she started receiving the services.
13		Family health history	Indicate if any of the following tests were performed: maternal serum alpha-fetoprotein, amniocentesis, chorionic villus sampling, sonogram or fetal echocardiogram.
14			Indicate if there is someone in the mother's family with a congenital anomaly/anomalies, including the mother herself.
	Infant		
15	Birth information	Type of delivery	Indicate if the delivery was vaginal or a caesarean section; also indicate whether birth was induced.

Appendix J

Suggestions for taking photographs of a fetus or neonate with a congenital anomaly

If parental consent is required for taking the photograph

- Ensure the consent form is signed before taking the photograph.
  - *Note*: if parents do not consent to a photograph, the fetus or neonate can still be included in the surveillance programme.

Prior to taking the photograph

- Have a clean, simple, non-patterned light or dark blue background (no blankets or other things in the bassinet or on the examination table).
- If there are objects on the examination table that affect the photograph, remove them before taking the photograph.

When taking the photograph

- Take a view of the entire fetus or neonate, plus several focused views of the congenital anomaly/anomalies.
- Take a separate view of the face, if possible.
- Take a front or back view, or both, plus a side view, depending on the congenital anomaly.
- Avoid taking photographs at an angle; i.e. take all photographs holding the camera at 90° to the fetus or neonate.
- Use no personal identification; instead use coded identification.
- If more than one photograph is taken, make sure that all photographs can be identified with a code for that particular fetus or neonate.
- Assign identifiers to the photograph files, using a unique code and adding an extra number to indicate the number of photographs taken of the same fetus or neonate (for example: 0001_1; 0001_2, etc.).
- Place a label next to, but not touching, the fetus or neonate, if needed. Similarly, place a ruler or measuring tape next to, but not touching, the fetus or neonate, to help estimate size.
- Ensure that there is adequate lighting and no shadows in the photograph. Use a flash if needed.
- Consider the cost of photograph storage.

When a digital camera is used

- Use high resolution, at least 300 ppi (pixels per inch).
- Review photographs quickly while on site.
- Save the image in jpeg (jpg) format; make sure each photograph is transferred to a computer file or other secure storage before deleting it from the camera.

Tablets or smart phones can also be used to take photographs.

Appendix K

Prevalence of selected anomalies by surveillance programme during 2004–2008 *(53)*

	Prevalence per 10 000 births								
Surveillance programme (alphabetical order)	**Anencephaly**	**Spina bifida without anencephaly**	**Encephalocele**	**Cleft palate without cleft lip**	**Cleft lip with or without cleft palate**	**Limb defects**	**Hypospadias**	**Omphalocele**	**Gastroschisis**
Australia Victoria	5.20	5.73	1.58	7.99	10.13	6.23	38.45	3.32	2.20
Australia Western	5.71	6.83	1.62	8.60	12.33	7.19	31.70	3.80	3.95
Canada Alberta	2.07	4.10	1.15	6.93	12.75	10.72	21.62	2.78	4.90
Canada British Columbia	1.18	2.97	0.33	8.62	7.49	3.63	22.56	2.35	4.66
Canada National	0.90	2.81	0.52	6.89	9.17	3.46	26.28	2.08	4.00
Chile Maule	1.80	1.65	1.20	3.76	10.68	3.91	7.97	1.80	2.11
China Beijing	2.47	1.73	0.91	1.92	9.18	2.51	0.52	0.91	1.43
China CBDMN[a]	2.58	5.26	1.46	2.86	13.49	4.82	4.74	1.46	2.43
Costa Rica	1.32	2.95	0.74	2.75	7.00	4.57	6.80	1.13	1.85
Cuba	3.98	5.12	1.60	1.67	4.49	2.27	9.41	2.16	5.35
Czech Republic	2.60	3.82	1.77	7.78	10.90	6.54	32.42	2.72	3.05
Finland	3.10	4.53	1.84	13.97	11.28	7.02	3.71	5.48	3.10
France Paris	5.24	4.35	2.40	6.25	8.83	6.37	15.46	5.68	1.96
France REMERA[b]	2.76	5.43	1.41	4.85	8.48	5.81	11.32	3.21	1.75
France Strasbourg	4.58	7.26	0.79	8.21	10.42	8.37	18.84	1.57	1.58
Germany Saxony Anhalt	2.53	6.56	1.04	9.21	14.28	7.60	8.29	3.68	5.07
Hungary	1.89	4.07	0.55	4.05	7.65	3.60	26.83	1.32	1.04
India	13.39	11.48	3.90	1.79	5.57	6.22	1.85	2.36	0.63
Iran TRoCA[c]	12.82	1.18	1.57	4.12	5.53	23.41	10.59	0.71	nr

	Prevalence per 10 000 births								
Surveillance programme (alphabetical order)	**Anencephaly**	**Spina bifida without anencephaly**	**Encephalocele**	**Cleft palate without cleft lip**	**Cleft lip with or without cleft palate**	**Limb defects**	**Hypospadias**	**Omphalocele**	**Gastroschisis**
Ireland Dublin	2.44	2.61	1.22	7.25	7.82	3.58	11.49	2.59	3.10
Israel IBDSP[d]	1.74	3.28	0.56	3.84	4.25	2.00	33.40	0.77	0.20
Italy Campania	3.41	3.56	1.01	6.15	7.38	4.48	3.28	1.67	0.19
Italy Emilia Romagna	2.02	3.08	0.85	4.46	6.84	5.57	14.70	1.96	1.27
Italy ISMAC[e]	1.51	2.26	0.25	4.52	4.02	2.76	20.83	nr	0.50
Italy North East	1.30	2.79	0.28	7.43	8.36	3.95	18.43	1.24	0.84
Italy Tuscany	1.73	2.99	0.80	3.65	4.91	4.91	8.50	1.86	1.06
Japan JAOG[f]	1.10	4.96	0.69	4.73	21.09	3.37	4.06	3.89	2.61
Malta	1.52	7.58	2.02	16.17	10.11	6.57	33.36	3.03	1.01
Mexico RYVEMCE[g]	4.70	6.26	1.57	2.97	13.76	6.34	3.62	1.89	5.19
New Zealand	0.36	2.24	0.56	8.44	6.85	2.31	27.52	4.17	nr
Northern Netherlands	1.53	5.36	0.44	7.00	13.56	5.80	23.29	1.86	1.09
Norway	3.85	5.53	1.01	7.22	13.26	4.55	14.40	2.56	3.27
Russia Moscow	2.77	4.00	0.96	4.48	5.88	2.60	13.75	2.09	3.63
Slovak Republic	0.76	2.40	0.80	4.80	8.18	3.35	20.30	0.58	0.87
South America ECLAMC[h]	5.43	9.13	2.91	5.06	13.70	8.11	6.27	4.28	6.43
Spain ECEMC[i]	0.31	1.02	0.21	3.82	3.59	4.45	1.61	0.52	0.63
Sweden	3.53	4.34	1.24	5.58	8.22	4.77	21.33	2.62	1.61
United Kingdom of Great Britain and Northern Ireland (UK), England and	2.68	2.64	0.60	3.24	5.50	3.10	8.70	1.06	2.88

	Prevalence per 10 000 births								
Surveillance programme (alphabetical order)	**Anencephaly**	**Spina bifida without anencephaly**	**Encephalocele**	**Cleft palate without cleft lip**	**Cleft lip with or without cleft palate**	**Limb defects**	**Hypospadias**	**Omphalocele**	**Gastroschisis**
UK, England Wessex	5.30	4.36	1.37	8.47	13.77	1.54	7.78	3.85	4.70
UK, Wales	5.25	6.73	2.01	9.09	11.33	8.50	28.45	4.01	6.85
Ukraine	8.64	10.66	2.09	5.30	8.08	3.48	3.14	1.60	1.60
USA Atlanta	2.46	3.90	1.14	4.96	9.77	3.90	6.06	1.95	4.37
USA Texas	2.35	3.75	0.88	5.64	10.91	5.45	15.83	2.01	5.45
USA Utah	2.43	4.60	0.86	6.85	12.49	6.55	8.57	2.54	5.50
Median	**2.50**	**4.09**	**1.02**	**5.44**	**9.00**	**4.67**	**12.62**	**2.09**	**2.32**
Interquartile range lower	**1.68**	**2.91**	**0.73**	**4.00**	**6.96**	**3.48**	**6.67**	**1.59**	**1.14**
Interquartile range upper	**4.13**	**5.46**	**1.57**	**7.51**	**11.58**	**6.41**	**21.86**	**3.12**	**4.28**

Note: the table gives an idea of variation of the total prevalence among various programmes. The variation may be due to a real prevalence variation, a different frequency of risk factors in the population, or under-registration or over-registration.

[a] CBDMB = Chinese Birth Defects Monitoring Network.

[b] REMERA = Registre Des Malformations en Rhone Alpes.

[c] TRoCA = Tabriz Registry of Congenital Anomalies.

[d] IBDSP = Israel Birth Defects Surveillance Program.

[e] ISMAC = Sicilian Registry of Congenital Malformations.

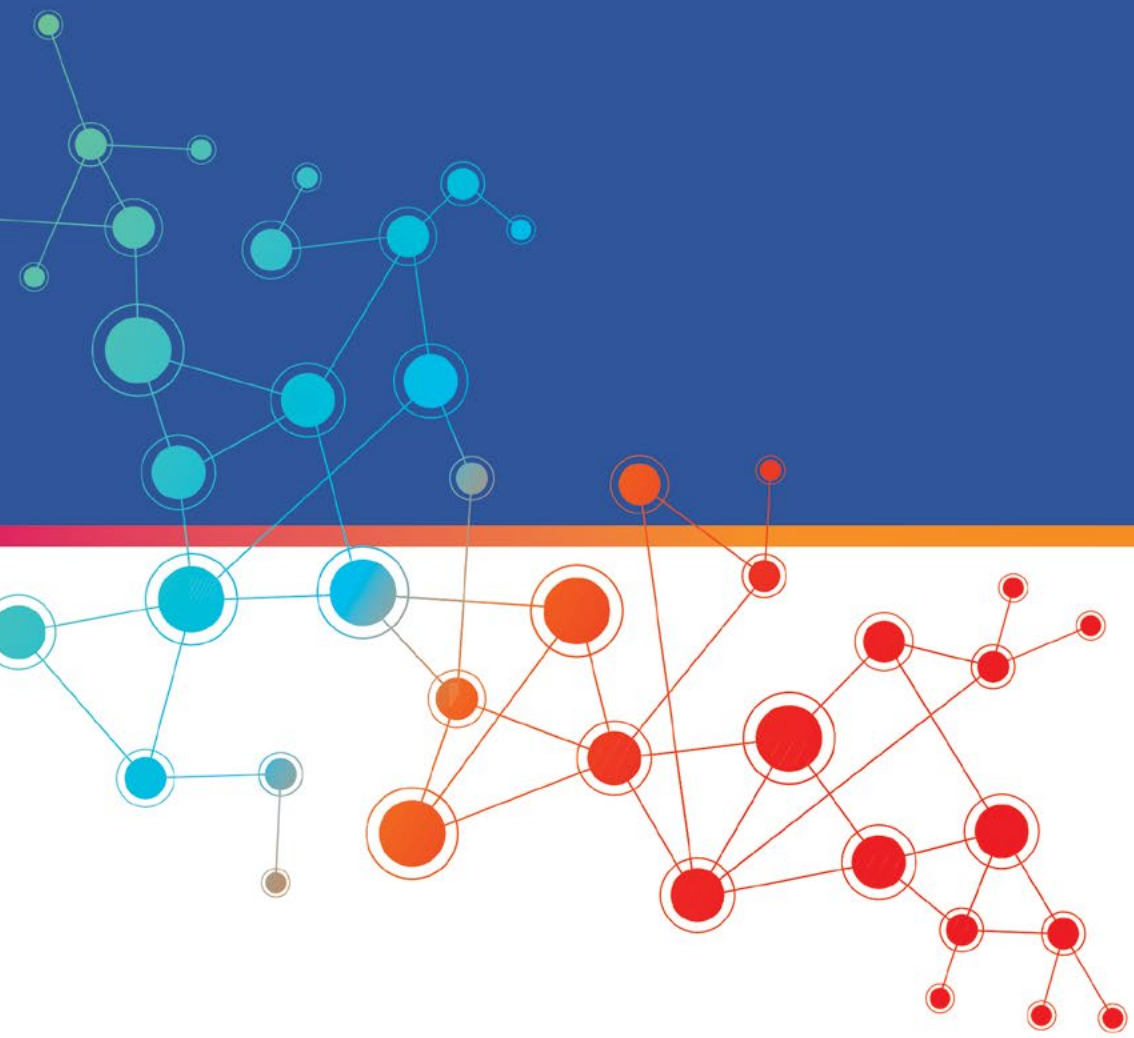
[f] JAOG = Japan Association of Obstetricians and Gynecologists.

[g] RYVEMCE = Mexican Registry and Epidemiological Surveillance of External Congenital Malformations.

[h] ECLAMC = Latin-American Collaborative Study of Congenital Malformations.

[i] ECEMC = Spanish Collaborative Study of Congenital Malformations.

nr = not reported.



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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