Canadian Congenital Anomalies Surveillance Enhancement Initiative (CASE)

INFORMAL GUIDELINES

For

Congenital Anomalies Surveillance

V 1.2 (June 2019)

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Introduction

Document Development

This document has been developed in response to a request at the February 2017 Congenital Anomalies Surveillance Enhancement (CASE) national meeting in Ottawa for a guide to facilitate the standardization of provincial and territorial data collection for submission to the national congenital anomalies dataset. At the meeting it was agreed that a subcommittee of provincial and territorial representatives would develop a comprehensive guide, with final review by the Public Health Agency of Canada. The Newfoundland and Labrador representative created the document format, edited subcommittee member submissions, and developed additional content where needed. The Alberta representative developed case definitions and coding guidelines for congenital anomalies identified as priority by the Public Health Agency of Canada (PHAC).

Although this document has been reviewed by PHAC, it remains an informal guide at present and regular updates will be undertaken to correct and /or add information. Changes to the guide will be addressed by the Data Quality Working Group of the CASE initiative and brought forward to the regular CASE initiative monthly teleconferences. The guide and future amendments will be circulated through the CASE group and will be available on the Perinatal Program Newfoundland Labrador's webpage.

Information for this guide was collated from the following sources:

1. *Guidelines for Congenital Anomalies Surveillance and Reporting to CCASS,*[1]
2. presentations and discussions from CASE annual meetings and monthly teleconferences,
3. Birth defects surveillance: a manual for programme managers,[2]
4. Guidelines for Conducting Birth Defects Surveillance,[3] and
5. various documents from the Malformation Coding Guides page of the EUROCAT website.[4]

For an introductory text on Epidemiology, see *Epidemiology for the Uninitiated, 5th Edition.*[5]

Congenital Anomalies Surveillance in Canada

Congenital anomalies (also called birth defects) include a wide range of structural and functional abnormalities that are present at birth, or that occur later but originate in the prenatal period. Diagnosis may occur before birth, at birth, or months or years after birth. Major anomalies can result in death and/or disability and require substantial medical care throughout life, causing significant economic and social burden (for example, spinal cord and heart anomalies). In Canada, major congenital anomalies occur in about 3% to 5% of newborns and 8% to 10% of stillbirths. They also account for about 23% of neonatal deaths (0 to 27 days after birth), second only to immaturity (1.1 and 1.5 deaths per 1,000 births respectively).[6]

Because the causes of most congenital anomalies are unknown, surveillance is an important step in developing primary preventive measures. A congenital anomalies surveillance system that accurately captures cases of congenital anomalies and exposure and risk information can identify geographical, temporal, and other trends and clusters to facilitate investigation into causes and contributing factors. This information can then be used to

- develop measures to prevent and/or lessen the impact of congenital anomalies,
- evaluate the measures that have been developed, and
- develop maternal health programs and policy to reduce the burden of congenital anomalies.

Examples of successful preventive measures include folic acid supplementation to reduce neural tube defects, pre-pregnancy immunization against rubella, and more recently the avoidance of travel to specific countries to reduce cases of Zika virus-related severe microcephaly.

In Canada, the Canadian Congenital Anomalies Surveillance System (CCASS, established by Health Canada in 1966 in response to the thalidomide tragedy) is a national surveillance system managed by the Maternal and Infant Health Section within the Public Health Agency of Canada. It collects and analyzes select live birth and registered stillbirth data to provide

- birth prevalence rates for selected congenital anomalies in Canada,
- temporal trends at the national level, and
- provincial / territorial and international comparisons.

CCASS ascertains cases of congenital anomalies primarily through the Canadian Institute for Health Information's national *Discharge Abstract Database* (DAD) which captures coded discharge information from hospitals in each province and territory (the DAD was originally developed in 1963). Facilities in all provinces and territories except Québec are required to submit hospital discharge data to the Institute (Québec submits similar data that is appended to the DAD).[7] Although the DAD is a valuable source for case ascertainment, it can have limitations that may impact the surveillance of congenital anomalies. Some examples of limitations impacting congenital anomalies surveillance are listed below:

1. **Missing cases:**
    - Diagnoses on terminations before 20 weeks gestation (before legal registration) may not have a baby record (e.g. neural tube defects).
    - For diagnoses on terminations at 20 weeks gestation or later, the main reason for termination is usually coded. There may be other diagnoses on autopsy, and these would not be included in the discharge abstract data.
    - Query diagnoses that have been removed may be confirmed later (query diagnoses are included automatically in the diagnosis variable and would normally be removed before reporting).
    - Testing results received after discharge are not included (e.g. x-ray, cytogenetic, etc.).
    - Defects not apparent at birth and diagnosed as an outpatient are not included (e.g. renal and cardiac defects).

- When a syndrome is coded, some diagnoses may not be included (e.g. cleft palate in Pierre Robin – coding stops when the syndrome diagnosis has been reached).
- There may be coding errors and/or interpretation differences (coding is not verified).

2. **False cases:**
   - Query diagnoses, if not removed from the diagnosis variable, may be ruled out after discharge (The DAD diagnosis variable contains both established and query diagnoses).
   - Established diagnoses may be changed on further investigation after discharge.
   - Anomalies associated with prematurity may be included (e.g. patent ductus arteriosus).
   - Anomalies acquired immediately following birth may be coded as congenital (e.g. hydrocephalus secondary to a brain bleed in extreme preemies).
   - Duplicate cases may be reported, especially for out-of-province births/admissions (as health numbers often differ between jurisdictions, an already-established case with a different identification number may be counted as a new case).
   - There may be coding errors and/or interpretation differences (coding is not verified).

3. **Information important for analysis that may not be collected or available**, including
   - full postal code (for geographical analysis),
   - longitudinal information on maternal and paternal risk factors and exposures, and
   - longitudinal outcome information such as developmental, parental stress, etc.

The above limitations of the Discharge Abstract Database have the potential to substantially impact data quality when used for congenital anomalies case ascertainment.

The Importance of Data Quality for Usefulness

Guidelines from well-known programs and groups have emphasized the importance of data quality for congenital anomalies surveillance systems. Some examples follow.

- ***Guidelines for Conducting Birth Defects Surveillance, Chapter 7 Data Quality Management:***[8]
  "The credibility of a birth defects surveillance program is built on a foundation of high-quality data. Information and results that are derived from surveillance data should be accurate, complete, and timely. Data quality influences the results of descriptive epidemiologic studies and, therefore, their interpretation. Data quality also affects the extent to which information can be utilized for planning, prevention, and intervention".

- ***Birth defects surveillance: a manual for programme managers*** (2014), page 37:[2]
  "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."

- ***World Health Assembly sixty-third session, agenda item 11.7*** (2010) resolution WHA 63.17 (1)[9] urges member states:

  "(5) to develop and strengthen registration and surveillance systems for birth defects within the framework of national health information systems in order to have accurate information available for taking decisions on prevention and control of these birth defects and to continue providing care and support to individuals affected by birth defects."

- ***Updated guidelines for evaluating public health surveillance systems*****,** Recommendations from the Guidelines Working Group (2001) Task D.2.c. Data Quality:[10]

  "The acceptability . . . and representativeness . . . of a public health surveillance system are related to data quality. With data of high quality, the system can be accepted by those who participate in it. In addition, the system can accurately represent the health-related event under surveillance."

Results obtained from poor-quality data may not accurately reflect the occurrence of congenital anomalies, possibly impacting investigative outcomes and the ability to develop, plan, and evaluate primary preventive efforts. With the causes of most congenital anomalies still unknown, high-quality data is essential for accurately investigating causes and contributing factors to facilitate the continuing development and evaluation of preventive measures.

Enhancing Data Quality through the CASE Initiative

As noted above, Canada currently uses national administrative hospital data collected through the Canadian Institute for Health Information for surveillance of congenital anomalies. However, many larger nations rely on regional (local) registries to collect information for their national systems[11]. Regional/local registries can have more complete data and function more efficiently due to the smaller area of operation, local contacts, and effective practices that ensure high participation rates.[12] These smaller local registries then submit data to the larger surveillance system for national/international reporting. Examples of such registries include

- EUROCAT (European Surveillance of Congenital Anomalies),
- National Congenital Anomaly and Rare Disease Registration Service (NCARDRS),
- National Birth Defects Prevention Network (NBDPN), and
- International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR).

In Canada, not all provinces and territories have a congenital anomalies surveillance system that can submit data to the national system. Furthermore, those that do may differ in their case definitions and data collection protocols, making combining regional data for a national dataset problematic. With the goal of increasing the accuracy and usefulness of the national system, the Public Health Agency of

Canada is taking a more regional approach by supporting provinces and territories in the establishment and development of strengthened, ongoing provincial and territorial congenital anomalies surveillance systems that can contribute data to the national system.

The current initiative, *Congenital Anomalies Surveillance Enhancement (CASE)*, is led by the Agency's *Canadian Perinatal Surveillance System* group (part of Health Canada's initiative to strengthen national health surveillance capacity), and funded by the 2008 federal initiative: *Action Plan to Protect Human Health from Environmental Contaminants*. The goals for the CASE initiative are to

1. support the development and enhancement of regional surveillance activities;

2. maximize comparability across jurisdictions by promoting the use of common procedures for surveillance such as consistent data variables and definitions, data collection methods, and conditions for surveillance;

3. facilitate direct and timely data-sharing for effective national congenital anomalies surveillance reporting; and

4. contribute to more robust datasets for determining risk factors of congenital anomalies and conducting risk assessments.

Informal guidelines for provincial and territorial congenital anomalies surveillance systems and national data submission for the CASE initiative follow.

References

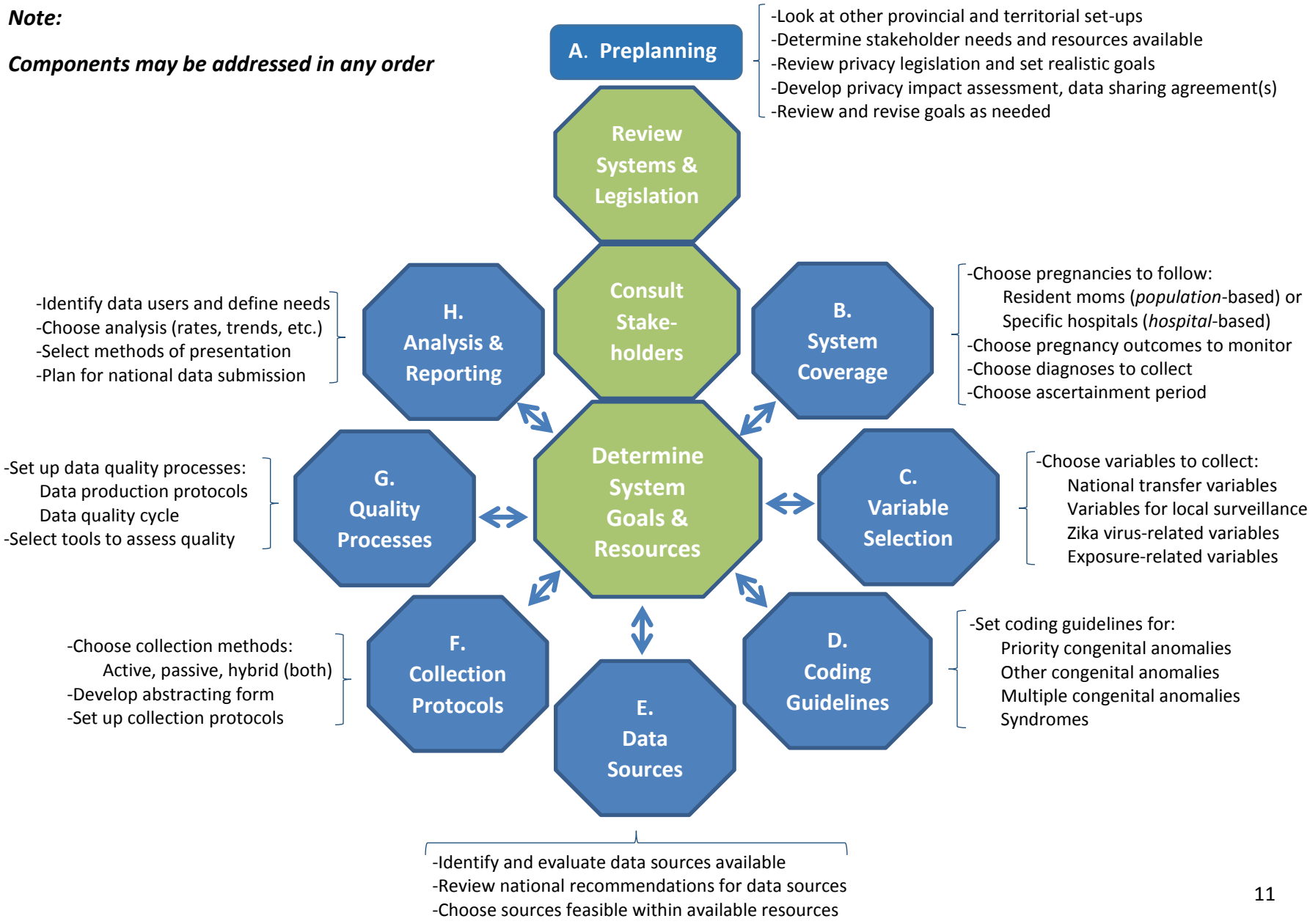
1. Canadian Congenital Anomalies Surveillance Network. Guidelines for congenital anomalies surveillance and reporting to CCASS, May 2011. Ottawa: Public Health Agency of Canada, Maternal and Infant Health Section.
2. WHO/CDC/ICBDSR. Birth defects surveillance: a manual for programme managers. Geneva: World Health Organization; 2014.
3. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised March 2015.
4. EUROCAT-network.eu (n.d.). Malformation coding guides.
5. Coggon D, Rose G, Barker DGP (2003). Epidemiology for the uninitiated, 5th edition. London: BMJ Books. ISBN 0 7279 1604 1.
6. Public Health Agency of Canada. Congenital anomalies in Canada 2013: a perinatal health surveillance report. Ottawa, 2013.
7. Canadian Institute for Health Information. Discharge Abstract Database Metadata (DAD) [Internet]. 2017 Jun [cited 2017].

8. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, Chapter 7 Data Quality Management. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised March 2015.
9. World Health Assembly, 63. (2010). Birth defects. World Health Organization.
10. Centers for Disease Control and Prevention. 2001. Updated guidelines for evaluating public health surveillance systems: recommendations from the Guidelines Working Group. MMWR Recomm Rep 2001;50:1-35.
11. Lowry RB. Congenital anomalies surveillance in Canada. Can J Public Health. 2008;99(6):483-485.
12. Misra T, Dattani N, Majeed A. Evaluation of the National Congenital Anomaly System in England and Wales. Arch Dis Child Fetal Neonatal Ed. 2005;90(5):F365-373.

Overview: Surveillance System Components

Note:

Components may be addressed in any order



A. Preplanning: Systems & Legislation Review

It is important to develop program goals and objectives as early as possible to guide development of a surveillance system that will meet the goals and objectives. This would normally include reviewing legislation, determining resources available, consulting with stakeholders, as well as looking at other programs.

Provincial/Territorial Program Overview

The regional congenital anomalies surveillance systems that currently exist in Canada vary by province and territory. When planning and developing surveillance systems, it may be helpful to talk with representatives from other provinces and territories that have systems developed to learn how they have set up their systems and handled challenges, especially if using a similar system. A quick overview of provincial and territorial systems is provided in Table 1 (updated May 2019) to help determine which systems may be of interest for review.

Table 1: Provincial/Territorial Congenital Anomalies Surveillance System Summaries

	Organization	Start Date	System Coverage*	Collection Methods**	Information Sources and Notes
NL	Perinatal Program NL/Eastern Health (NLCASS)	2013	***Outcomes:*** LB, SB, TOPFA at any GA ***Moms:*** NL residents no matter where delivered	***Hybrid*** (both passive and active)	• Hospital-coded Discharge Abstract Database (DAD) and perinatal data (3M hospital data system) • Audits of DAD with medical records for specific anomalies • Accessing reports: Cytogenetic, autopsy, MFAU, x-ray, etc. • Genetics referral database report • Clinic case reporting: Cardiology, Perinatal, DS/CLP/CP • Home births, deaths, out-of-province deliveries / admissions
PEI	PEI Reproductive Care Program				• Not a participant at present • Exploratory discussions underway
NS	Surveillance of Congenital Anomalies NS SCA-NS	2012	***Outcomes:*** • LB • SB • TOPFA	***Hybrid:*** • Passive	*Confirmed & proposed data sources* • Nova Scotia Atlee Perinatal Database (NSAPD) (Confirmed) • Fetal Anomaly Database (FADB) (Confirmed) • IWK Cardiology Database (Proposed)

	Organization	Start Date	System Coverage*	Collection Methods**	Information Sources and Notes
			Moms: All NS residents, selected out-of-province deliveries		• Canadian Institute for Health Information Discharge Abstract Database, housed at Department of Health and Wellness (Confirmed) • Vital Statistics Database; Service Nova Scotia (Piloted) • IWK Benetech PRA Maternal Serum Screening Database (Proposed) • Laboratory Genetic Database (Piloted) • Medical Services Insurance (MSI) claims (i.e., physician billings), housed at Department of Health and Wellness (Confirmed) • Other databases as they become available.
NB	PerinatalNB	2016	***Outcomes:*** • LB • SB • TOPFA ***Moms:*** All NB residents delivered in NB	***Passive*** – moving towards hybrid similar to NL	• Currently only data collected through 3M (NB DAD with additional perinatal fields coded at discharge as well as all outpatient procedures)
QC	Système de surveillance des anomalies congénitales au Québec	2018			• To be determined (live births file, stillbirths, deaths, hospitalizations, physician billing, pharmacy services file). These datasets will be linked in 2019.
ON	BORN Ontario				• Real time collecting data through The BORN Information System (BIS) suspected or confirmed fetal anomalies in AS (antenatal speciality) encounter data • Suspected or confirmed fetal anomalies in PSOF (prenatal screening follow-up) • Suspected or confirmed newborn anomalies in birth child encounter data, and in NICU encounter data

	Organization	Start Date	System Coverage*	Collection Methods**	Information Sources and Notes
MB	Manitoba Health	2011			• MOA expired 2017 • Renewal MOA 2019-2022 under development
SK	Saskatoon Health Region				• MOA expired 2014 • Contact and collaboration pending for future agreement
AB	Alberta Children's Hospital/ACASS	1980	***Outcomes:*** LB, SB, some fetal deaths < 20 wks, elective terminations ***Moms:*** Province-wide	***Hybrid:*** Secondary sources, with verification for unclear or unconfirmed diagnoses	• Vital records (Births, Deaths, Stillbirths) • Hospitals (delivery, pediatric, tertiary care) – Case reporting form, pathology log book • Clinical genetics centres • Cytogenetics laboratories • Pathology departments • NB screening program
BC	Health Status Registry	1952	***Outcomes***: • LB • SB • TOPFA (≥ 20 weeks/500g)	***Passive***	• Vital Statistics records of live birth, stillbirth, and death. Records of stillbirth include terminations at ≥ 20 weeks/500g • Few clinic case reporting • Other sources have ceased reporting to the registry; data quality has substantially declined
	CA Enhanced Surveillance system (plan)	In development	***Outcomes***: • LB • SB • TOPFA **Moms**: all BC residents regardless of where delivered (see note in information sources)	***Hybrid***: • Passive • Active (terminations data)	• DAD data (includes data on all BC residents regardless of where accessed care) • Vital Statistics (records of LB, SB, Death – only for BC residents) • Terminations data with diagnosis of congenital anomalies to be sent directly from health authorities • Future state to include: provincial laboratory diagnostic data, antenatal care record, information on prescriptions filled, etc. • Possible future integration with Population and Public Health Observatory, with mandate for surveillance of non-communicable diseases and injuries, risk and protective factors, and environmental health.

	Organization	Start Date	System Coverage*	Collection Methods**	Information Sources and Notes
YT	Congenital Anomalies Support Yukon	2018	***Outcomes***: LB, SB ***Moms:*** All YT residents regardless of where delivered	***Hybrid:*** Direct reporting from 1) maternity ward 2)pediatricians	• LB with a defect flagged by maternity care providers at d/c • Child with a birth defect flagged by pediatricians • Q and O-35 ICD 10 codes flagged in 3M at Whitehorse General Hospital
NT	NWT Congenital Anomalies Surveillance System	2011	***All pregnancies***	***Passive***	• Physicians • Includes the NWT 'Congenital Anomalies Reporting Form' • Birth registry
NU	Nunavut Health and Social Services				• MOA expired 2017 • Renewal MOA under review

* LB = live births, SB = stillbirths, TOPFA = termination of pregnancy for fetal anomalies

**See section 'F' for types of collection methods.

Legislation Considerations

Federal, provincial, and territorial legislation establishes rules that custodians must follow when collecting, using, and disclosing (sharing) personal health information. Adherence to personal health information legislation is facilitated by the development and approval of

- Privacy Impact Assessments, and
- Data Sharing Agreements.

Privacy Impact Assessments. Privacy impact assessments are documents used to identify the potential privacy risks of government programs and services, and to describe how those risks will be minimized. They also help reduce risks of privacy breaches to an acceptable level as determined by the Privacy Commissioner.[1]

For the current Congenital Anomalies Surveillance Enhance (CASE) initiative, privacy impact assessments are developed and approved within jurisdictions and may be a part of a jurisdiction's deliverables. For an example related to the current Congenital Anomalies Enhancement (CASE) initiative, contact the Public Health Agency of Canada representative for the initiative, or individual provincial and territorial representatives.

Data Sharing Agreements. Data sharing agreements are written records of understanding between government parties and/or organizations that outline the terms and conditions under which personal information is shared. This may mean that one party is disclosing information while the other party is collecting information, or information may be exchanged.[2]

For the CASE initiative, data sharing agreements are required between each province/territory and the Public Health Agency of Canada for sharing data for national reporting. Some jurisdictions may also require agreements to collate information from regions within their jurisdictions, or for exchanging information with other provinces and/or territories. The *Multilateral Information Sharing Agreement (MLISA)* was developed to support public health information sharing among federal, provincial and territorial governments of Canada.[3] For an example of an agreement currently in use for the CASE initiative, contact the Public Health Agency of Canada representative for the CASE initiative, or individual provincial and territorial representatives.

List of Provincial and Territorial Legislation

Legislation governing personal health information differs by province and territory. See Table 2 for a short summary of current legislation in a number of jurisdictions (updated May 2019).

Table 2: Provincial/Territorial Legislation for Personal Health Information

	Legislation	Comments
NL	*Personal Health Information Act*	• Proclaimed April 1, 2011 • Collection of congenital anomalies data is not mandated
PEI	*Health Information Act*	
NS	*Personal Health Information Act (PHIA)*	• PHIA was proclaimed on December 4, 2012 and came into force on June 1, 2013. A three-year review was completed in 2018.
NB	*Personal Health Information Privacy and Access Act*	• Assented June 2009, current as of May 2018 • Collection of congenital anomalies data is not mandated
QC	*Act Respecting Access to Documents Held by Public Bodies and the Protection of Personal Information*	• Collection of congenital anomalies data is not mandated
ON	*Personal Health Information Protection Act*	• Collection of congenital anomalies data is not mandated
MB	*The Personal Health Information Act*	
SK	*The Health Information Protection Act*	
AB	*Health Information Act* (2001)	• Section 27, subsection 2c allows ACASS the legal and ethical access to identify health information for public health surveillance. • Section 27, subsection 1D allows ACASS legal and ethical access to identify information for conducting research
BC	*Freedom of Information and Protection of Privacy Act*	• Under the Health Act, the Health Status Registry has authority to collect data on congenital anomalies (CAs), but CAs are not reportable in BC.
YT	*Health Information Privacy and Management Act*	• Proclaimed in force as of August 31, 2016 • Yukon's authority to collect CA data falls under the *Public Health and Safety Act* (CMOH is allowed to collect identifiable information for the purpose of surveillance) • Collection of congenital anomalies data is not mandated (except for FASD which is mandated)

	Legislation	Comments
NWT	*NWT Public Health ACT*	• Collection is mandated • The PHA was reviewed in 2010, allowing for the registry to be created.
NU	*Consolidation of Access to Information and Protection of Privacy Act*	

References

1. Office of the Privacy Commissioner of Canada. Privacy Impact Assessments. 2017 Jun [cited 2016 Nov].
2. Government of Canada. Guidance on preparing information sharing agreements involving personal information [Internet]. 2017 Jun [cited 2010 Aug].
3. Public Health Agency of Canada. Overview of the Multi-Lateral Information Sharing Agreement (MLISA) to Support Public Health Information Sharing among Federal, Provincial and Territorial (F/P/T) Governments in Canada [Internet]. 2017 Jun [cited 2014 Sep].

B: System Coverage

To accurately determine congenital anomalies rates and changes over time, the population under study must be clearly defined. Deviations from the defined population must be noted for submissions to the national dataset and for preparing reports and publications.

Pregnancies to Follow (Type of System)

For the CASE initiative, the pregnancies to follow would ideally be the population for the province or territory *(*a *population-based* system). However, under some conditions where it is not feasible to collect data from the whole province or territory, pregnancy outcomes occurring at one or more hospitals may be selected instead *(*a *hospital-based* system*)*. Characteristics of these two systems follow below.

- **Population-based:**
  - Includes moms who are ***resident*** in the defined area at birth/outcome.
  - Residents delivering outside the area are included.
  - Denominator data = birth outcomes for the residents of the defined area.
  - May require significant resources, especially if many births occur outside the area.
  - If unable to capture resident births outside the area, reports and publications should provide the types of births missing to facilitate interpretation of rates.

- **Hospital-based:**
  - Includes moms who ***deliver in specific hospitals*** in the defined area (may include deliveries occurring on the way to hospital if admitted on arrival).
  - Births outside the specific hospitals are not included, regardless of mom's residency.
  - Does not include births at home/birthing centers.
  - Denominator data = birth outcomes for participating hospitals.
  - Requires less resources than a population-based system.
  - Can be biased if a hospital is a tertiary care centre serving complex pregnancies.
  - Cases diagnosed after discharge may be missed.

For the CASE initiative, population-based systems are the goal. Details of the pregnancies being followed must be clearly stated when submitting data to the national system as well as for preparing reports and publications.

Outcomes to Follow

Ideally, systems should monitor live births, stillbirths, and terminations for fetal anomalies (TOPFA). The following definitions are provided from the CASE guidelines developed for the initiative in 2011 (*Guidelines for Congenital Anomalies Surveillance and Reporting to CCASS,* May 2011):[1]

- **Live birth:** A complete expulsion or extraction from the mother, irrespective of the duration of the pregnancy, of a fetus in which, after expulsion or extraction, there is breathing, beating of the heart, pulsation of the umbilical cord or definite movement of a voluntary muscle, whether or not the umbilical cord has been cut or the placenta is attached.

- **Stillbirth**: A complete expulsion or extraction from the mother, after at least 20 weeks of pregnancy, or after attaining a weight of 500 grams or more of a fetus in which, after expulsion or extraction, there is no breathing, beating of the heart, pulsation of the umbilical cord or unmistakable movement of voluntary muscle. (Note: some discussion has happened around revising this definition; however, there are no plans to do so at this time).

- **Termination of Pregnancy** [for fetal anomalies]: Any induced delivery (usually less than 20 weeks of gestational age) regardless of the status of the fetus, i.e., either intrauterine fetal death or therapeutic termination for fetal anomalies.
    - ***Less than 20 weeks****:*
      Many terminations for congenital anomalies will be done prior to 20 weeks and hence are not captured under the Vital Statistics Act [in such cases, all screening and investigation results will be maintained on the mother's medical record; the enhanced system should capture these if possible].

    - ***20 weeks or later:***
      A small number of terminations may be done between 20 and 23 completed weeks for serious or lethal anomalies. Such events are then registered as a stillbirth, or as a live birth if meeting the live-birth criteria, even though labour started as a termination procedure.

      It is useful to have a field on the computer which helps to classify post 20 weeks events, e.g., stillbirth – termination, live birth – termination. This is to provide a clearer picture whether a temporal trend is due to treatment, e.g., folic acid fortification, natural temporal trends, or unnatural ones (e.g., terminations). The PHAC recommended variables in the following section includes a flag for termination due to diagnosis of fetal anomalies (variable #9b).

- **Spontaneous loss/miscarriage**:
    - If a spontaneous loss/miscarriage happens to have fetal anomalies it should be coded as a spontaneous abortion and the anomalies coded if known. [Note: in such cases, all screening and investigation results will be maintained on the mother's medical record.]

    - If an intrauterine fetal death occurs greater than 20 weeks and is then induced, it is coded as a stillbirth.

When submitting data to the national system and preparing reports and publications, types and definitions of outcomes captured should be clearly stated.

Diagnoses to Collect

The Canadian Congenital Anomalies Surveillance System recommends the following:[1]

1. The congenital anomalies surveillance at the national level includes all codes in the section Q of the International Classification of Diseases, 10th revision (ICD-10) developed by the World Health Organization (WHO 1996). All anomaly conditions coded in the section Q of the ICD 10 should be included in congenital anomaly surveillance.

2. Jurisdictions with the ability to expand the scope of surveillance should do so, e.g. to include inborn errors of metabolism, hereditary muscle and blood disorders.

Case Ascertainment Period

Bower et al. (2010),[2] using data from the Western Australian (WA) Birth Defects Registry, reported that about 87% of congenital anomalies were diagnosed by one year of age, and 99% by six years of age. Timing of diagnoses included

- 18.7% prenatally,
- 47.8% between birth and 1 month,
- 20.4% between 1 month and 1 year, and
- 12.1% between 1 year and 6 years.

Cases diagnosed after one year included urogenital, central nervous system, cardiovascular, and muscular congenital anomalies, Fetal Alcohol Syndrome (FAS), and congenital hearing loss. Number of cases diagnosed after one year was particularly large for both Fetal Alcohol Syndrome (66.7%), and congenital hearing loss (24.1%).

Recommendations for the case ascertainment period. The case ascertainment period may vary with jurisdiction based on resources available. The Canadian Congenital Anomalies Surveillance System (CCASS) recommends that[1]

1. The case ascertainment period should be a minimum of one year of age.

2. Ideally, ascertainment should be up to 6 years of age, as this will allow for the collection of data relating to such conditions as Fetal Alcohol Spectrum Disorder (FASD), autism, sensory impairment and other disorders.

3. Lifetime ascertainment is desirable, but is currently not feasible in all jurisdictions (it may be feasible in some jurisdictions).

Zika virus-related pregnancies. Subsequent to the development of the above guidelines, the Public Health Agency of Canada recommended increasing the follow-up period to two years after birth for pregnancies positive for Zika virus exposure.

References

1. CCASS. Guidelines for congenital anomalies surveillance and reporting to CCASS May 2011. The Public Health Agency of Canada, Maternal and Infant Health Section.
2. Bower C, Rudy E, Callaghan A, Quick J, Nassar N. Age at diagnosis of birth defects. Birth Defects Res A: Clin Mol Teratol. 2010;88(4):251-255.

C: Variable Selection

When choosing the information to collect, consideration should be given to the deliverables required for the CASE initiative, the jurisdiction's objectives, and the interests of other stakeholders.

Selection of variables can also be affected by available data sources, birth outcomes monitored, method of case ascertainment, case definitions, and program resources. The National Birth Defects Prevention Network recommends that the following data characteristics be considered when choosing variables:[1]

- *Availability* - The data must be retrievable and easily accessed by the program.
- *Consistency* - The data must have a consistent meaning across data sources and time.
- *Accuracy* - The data must accurately represent the condition.
- *Uniqueness* - The information is not already being collected.
- *Definability* - Each variable needs a clear definition.
- *Collectability* – Data can be quickly abstracted with a high degree of accuracy by available staff.
- *Comparability* - Data should be comparable with other programs/jurisdictions.

For further information on variable selection, see the NBDPN *Guidelines for Conducting Birth Defects Surveillance, Chapter 4 Data Variables.*[1]

Recommended Variables

CCASS has developed an extensive list of core and recommended variables for collection based on scientific literature review and review of surveillance systems in Canada, the United States, and Europe. The list has 36 core variables (considered a minimum for a useful provincial/territorial system) plus 53 recommended variables.[2] See details of both core and recommended variables in Table 3.

Table 3: Core and Recommended Variables for Congenital Anomalies Surveillance

Var. #	Variable Type	Variable Name	Format	Use
		Infant/Fetus		
1	Core	Personal (unique) Health Number	Char 9	Unique ID for linkage
2	Core	Birth Registration Number (BRN)	Char 11	Unique ID in Vital Statistics
3	Core	Date of Birth/Termination	mm/dd/yyyy	Linkage and age calculation
4	Core	Place of Birth (Province)	Text	
5	Core	Hospital ID of Birth	Char 12	Linkage and identification
6	Core	Death Registration Number (DRN)	Char 11	Unique ID in Vital Statistics
7	Core	Date of Death	mm/dd/yyyy	Linkage and survival analysis
8	Core	Place of Death	Text	Analysis and reporting
9	Core	a) outcome Live/Stillbirth/abort b) Termination-fetal anomalies (TOPFA) flag	Char 1 Char 1	Linkage and analysis
10	Core	Sex	Char 1	Linkage and analysis
11	Core	Family name	Char 50	Linkage and identification

Var. #	Variable Type	Variable Name	Format	Use
12	Core	First name	Char 40	Linkage and identification
13	Core	Middle name	Char 50	Linkage and identification
		Mother		
14	Core	Personal Health Number (PHN)	Char 9	Unique ID for linkage
15	Core	Date of Birth	mm/dd/yyyy	Linkage and age calculation
16	Core	Family Name	Char 50	Linkage and identification
17	Core	Maiden Name	Char 50	Linkage and identification
18	Core	First Name	Char 40	Linkage and identification
19	Core	Middle Name	Char 50	Linkage and identification
20	Core	a) Residence Postal Code at Delivery b) Residence Province Code at Delivery	Char 2 Char 7	Linkage and analysis
21	Core	Residence Standard Geographic Code	Char 7	Linkage and analysis
22	Core	Residence Health Region	Char 2	Linkage and analysis
		Case Ascertainment and Reporting		
23	Core	Source of Report	Char 7	Analysis and reporting
24	Core	Name/code of Reporting Agency	Text	Analysis and reporting
25	Core	Single or Multiple Anomalies	Char 1	Analysis and reporting
26	Core	Syndrome flag	Yes/No	Analysis and reporting
27	Core	Diagnostic code(s), ICD10-CA, ICD9-CM	Char 7	Analysis and reporting
28	Core	ICD version indicator	09 or 10	Analysis and reporting
29	Core	Baby's Birth Weight (grams)	Numeric	Analysis and reporting
30	Core	Gestational Age (weeks)	Numeric	Analysis and reporting
31	Core	Plurality (singleton, twins, etc.)	Char 2	Analysis and reporting
32	Core	Date of Reporting	mm/dd/yy	Analysis and reporting
		Provider Contact Information		
33	Core	Name of Responsible Party (physician)	Text	
34	Core	Mailing address of Responsible Party	Text	
35	Core	Tel and Fax Number of Responsible Party	Numeric	
36	Core	Email Address of Responsible Party	text	
		Personal Identification and Administrative Information		
37	Recommended	Baby name change flag	Char 1	Linkage and identification
38	Recommended	Adopted Name (AKN)	Text	Linkage and identification
39	Recommended	Mother's Hospital Chart ID	Char 12	Linkage and identification
40	Recommended	Mother's Mailing Address at Birth or Termination	Text	Linkage and identification
41	Recommended	Mother's Place/Country of Birth	Text	Analysis and reporting
42	Recommended	Marital Status of Mother	Char 2	Analysis and reporting
43	Recommended	Immigrant from other countries (mom)	Yes/No	Analysis and reporting
44	Recommended	Mother's Ethnicity	Char 2	Linkage and analysis
45	Recommended	Father's Date of Birth	mm/dd/yyyy	Linkage and age calculation

Var. #	Variable Type	Variable Name	Format	Use
46	Recommended	Father's ethnicity	Char 2	Linkage and analysis
Diagnostic Information – Infant/Fetus				
47	Recommended	Text Description of Congenital Anomaly	Text	Analysis and reporting
48	Recommended	Birth Length (cm)	Numeric	Analysis and reporting
49	Recommended	Weight Centile	Numeric	Analysis and reporting
50	Recommended	Head Circumference (cm)	Numeric	Analysis and reporting
51	Recommended	Apgar Score	Numeric	Analysis and reporting
52	Recommended	Birth Order	Numeric	Analysis and reporting
53	Recommended	Cytogenetic Analyses Performed	Yes/No	Analysis and reporting
54	Recommended	Diagnostic Tests and Procedures Performed	Yes/No	Analysis and reporting
55	Recommended	Autopsy Performed	Yes/No	Analysis and reporting
56	Recommended	Date of Diagnosis or Test(s)	mm/dd/yyyy	Analysis and reporting
Diagnostic Information - Mother				
57	Recommended	Medical Record Number(s)	Char 12	Linkage and identification
58	Recommended	Receiving Prenatal Care Flag	Yes/No	Analysis and reporting
59	Recommended	Participate Prenatal Class Flag	Yes/No	Analysis and reporting
60	Recommended	Number of Prenatal Visits	Numeric	Analysis and reporting
61	Recommended	Date of Last Menstrual Period (LMP)	mm/dd/yyyy	Analysis and age calculation
62	Recommended	Number of Ultrasounds in 1st Trimester 2nd Trimester	0, 1-2, 3-4, 5+ 0, 1-2, 3-4, 5+	Analysis and reporting
63	Recommended	Mother's Pre-pregnancy weight (grams)	Numeric	Analysis and reporting
64	Recommended	Mother's Height (cm)	Numeric	Analysis and reporting
65	Recommended	Number of Pregnancies	Numeric	Analysis and reporting
66	Recommended	Parity	Numeric	Analysis and reporting
67	Recommended	Number of Pregnancy Losses: Stillbirth, Spontaneous, Induced	Numeric	Analysis and reporting
68	Recommended	Maternal Conditions: ○ Chronic Hypertension, ○ Pre-Pregnancy Diabetes, ○ Obesity, ○ Pre-Pregnancy Heart Disease, ○ Pre-Pregnancy Renal Disease, ○ Blood-borne pathogens (HIV/AIDS, HBV, HCV, Other), ○ Other (specify)	Yes/No	Analysis and reporting
69	Recommended	Pregnancy Complications: ○ Pre-eclampsia, ○ Pregnancy-induced HBP, ○ Gestational Diabetes, ○ Prenatal Bleeding, ○ Premature Rupture of Membranes,	Yes/No	Analysis and reporting

Var. #	Variable Type	Variable Name	Format	Use
		○ Pregnancy Infections (Syphilis, Rubella, TB, Other), ○ Other (specify)		
70	Recommended	Mode of Delivery (Vaginal vs. Caesarean)	Vag/CS	Analysis and reporting
71	Recommended	Complications of Delivery: ○ Prolonged Labor, ○ Shoulder Dystocia, ○ Forceps Delivery, ○ Birth Asphyxia, ○ Other (Specify)	Yes/No	Analysis and reporting
72	Recommended	Prenatal Screening (also capture modality using text, as screening systems may differ): ○ Down Syndrome, Trisomy 18 & 13 ○ Neural Tube Defects, ○ Other Anomalies (Specify), ○ Other Screening (Specify) Newborn Screening: ○ Congenital metabolic disorders ○ Pulse oximetry screening	Screen: Yes/No Modality: Text	Analysis and reporting
Parental Social Class and Socio-economic Information				
73	Recommended	Mothers Education: ○ Highest University Degree, ○ Highest College Degree, ○ Trade Certificate or Diploma, ○ Completed High School, ○ Less than Grade 12 education	Text, Yes/No	Analysis and reporting
74	Recommended	Mother on Social-Family Welfare Flag	Yes/No	Analysis and reporting
75	Recommended	Mother on Health Care Subsidy Flag	Yes/No	Analysis and reporting
76	Recommended	Mother's Occupation at Conception	Text	Analysis and reporting
77	Recommended	Father's Occupation at Conception	Text	Analysis and reporting
Parental Risk or Protective Factors				
78	Recommended	Maternal Alcohol Use prior/during Pregnancy	Yes/No	Analysis and reporting
79	Recommended	Prescription Drug Use during Pregnancy	Yes/No	Analysis and reporting
80	Recommended	Non-Prescription Drug Use prior/during Pregnancy	Yes/No	Analysis and reporting
81	Recommended	Maternal Smoking prior/during Pregnancy	Yes/No	Analysis and reporting
82	Recommended	Other Maternal Exposure to: ○ Environmental hazards, ○ Radiations, ○ Medications	Yes/No	Analysis and reporting
83	Recommended	Family History of Malformations	Yes/No	Analysis and reporting
84	Recommended	Family History of Genetic Diseases	Yes/No	Analysis and reporting
85	Recommended	Use of Folic Acid at periconceptional period	Yes/No	Analysis and reporting

Var. #	Variable Type	Variable Name	Format	Use
86	Recommended	Assisted Reproductive technology (ART)	Yes/No	Analysis and reporting
87	Recommended	ART – drug administered: clomiphene, gonadotropin, other, none, unknown	Yes/No	Analysis and reporting
88	Recommended	ART –intervention: IVF only, IVF with ICSI, unknown	Yes/No	Analysis and reporting
89	Recommended	ART/IVF – embryo frozen and thawed	Yes/No/Unk	Analysis and reporting

In addition to the above variables, CCASS also suggests that jurisdictions consider creating an 'OMIM code' variable (May 2011 guidelines).[2] OMIM codes, the codes for certain single gene disorders listed in Online Mendelian Inheritance in Men (OMIM), are available free from the National Center for Biotechnology Information.

Variables for Transfer

CCASS has developed a list of anonymized variables to be transferred to the national system to improve national congenital anomalies surveillance. See details in Table 4.[2]

Table 4: Anonymized National Variables for Transfer to the Public Health Agency of Canada

Variable	Description	Values/Codes	Reason required/Notes
BATCHID	Unique ID# for each batch	*10 characters:* PTYYYY#### (e.g. batch 1 = NL20130001, batch 2 = 0002)	Batch ID's should not be duplicated.
CASEID	Unique encrypted ID per case	10 characters, YYYY###### (e.g. Case 1 = 2013000001, 2013000002)	Unique #; To identify case for contacting program for quality/research purposes.
PROV_ BIRTH	Province of birth outcome	*2 characters:* ## 10 = NL, 11 = PEI, 12=NS, 13=NB, 24=QC, 35=ON, 46=MB, 47=SK, 48=AB, 59=BC, 50=YT, 61=NT,62=NU, 09=Unknown, 99=Outside Canada	To find duplicate moms/babies who sought care outside the area of residence, or moved during baby's first year of life.
PROV_ RES	Mother' province of residence at birth outcome	*2 characters:* As above for PROV_BIRTH	For provincial/territorial presentation of congenital anomaly rates.
BIRTH_ DATE	Date of birth outcome	*8 characters:* DDMMYYYY -use 99 for missing month -If not able to send full DOB, set day to 99 (e.g. 99042013)	To produce statistics and reports, examine trends, and identify the time period when diagnosis was made.
PC_RES	Mother's residence postal code at birth outcome	*6 characters:* L#L#L#	To identify geographical clusters, allow precise mapping for environmental factors, or linkage to demographic data (e.g. census).

SGC_RES	Mother's residence SGC code at birth outcome	*7 characters:* ####### -9999999 = unknown	As above for PC_RES.
MAT_ DOB	Mother's date of birth	*8 characters:* DDMMYYYY -use 99 for missing month -If not able to send full DOB, set day to 99 (e.g. 99041990)	To determine mother's age at pregnancy outcome.
OUTCOME	Pregnancy outcome	*1 character:* # 1 = Live Birth 2 = Fetal death >/= 20 weeks gestation (CCASS stillbirth definition) 3 = Fetal death < 20 weeks gestation (CCASS miscarriage/abortion definition) 9 = Unknown	Used to help identify outcomes from terminations vs natural causes, evaluate trends in prenatal diagnosis and impact of prevention strategies, help determine need for services.
TOP	Termination of pregnancy at any age after prenatal diagnosis of congenital anomalies	*1 character: #* 1 = Yes 2 = No 9 = Unknown/missing	To increase accuracy of frequency estimates by identifying live births/still births from natural causes vs terminations.
NUMFET	# of babies/ fetuses at birth/ termination	*2 character: ##* 1 = Singleton; 2 = Twins, 3 = Triplets, 4 =Quads; 99 = Unknown/missing -Conjoined twin = 1 record -Fetal reduction = record # babies born	To evaluate differences in congenital anomaly rates for single vs multiple births.
SEX	Sex of baby/fetus	*1 character:* # 1 = Male; 2 = Female; 3 = Indeterminate 9 = Unknown/missing	To evaluate differences in congenital anomaly rates by sex.
BIRTHWT	Weight of baby/fetus at birth outcome in grams	*4 numeric:* #### (grams) 9999 = Unknown/missing ***-Note: Do not use 99 or 999 for missing #*** -Multiples are recorded separately -Conjoined twins = one record	To assist in assessing small for gestational age; For birth weight-specific rates of live births and stillbirths with congenital anomalies
GESTAGE	Completed weeks gestation at outcome	*2 numeric:* ## (completed weeks) 99 = Unknown/missing	To determine if case meets set definitions (some cases excluded if premature)
DEATH-DATE	Date of death (for ***live births*** only)	*8 characters:* DDMMYYYY -Use 99 for month if unknown/missing -If unable to send full date, set day to 99 (e.g. 99042013) 222222 = confirmed alive at 1 year 333333 = unknown status at 1 yr.	To determine survival rates for specific congenital anomalies
DXCODE1-DXCODE20	Diagnostic codes for congenital anomalies	*5 characters*: Q#### -'Q' plus 4-digit code -If more than 20 congenital anomalies, additional fields may be added	Allows for easy identification and analysis; standardizes anomalies and allows for comparability
DXPREFIX01-DXPREFIX##	Prefix code for ICD 10-CA code	*1 character: #* 'Q' indicates this is a query (suspected) case	To determine number of established vs suspected cases

Zika Virus-Related Variables

In 2016, the Public Health Agency of Canada requested that additional variables be collected by provincial/territorial congenital anomalies surveillance systems for pregnancies testing positive for Zika virus exposure. These variables include

- maternal Zika virus testing results,
- travel information (maternal and partner),
- a variable indicating that suspected Zika-virus related anomalies are present,
- a description of suspected Zika-virus related anomalies, and
- detailed follow-up information for up to *two years after birth*.

Environmental Exposure Variables

One of the recommended variables for collection for the CASE initiative is '*Other Maternal Exposure - Environmental Hazards'* (see Table 3, variable # 82). There are many possible variables that can be collected for environmental exposure, depending on the area of interest, data available, and resources for collection. Some areas to consider follow.

- **Socio-economic variables:**
  - Neighborhood characteristics

- **Ambient air quality:**
  - Fine particulate matter ($PM_{2.5}$)
  - Sulphur oxides (SO_x),
  - Oxides of nitrogen (NO_x)
  - Ozone (O_3)
  - Carbon monoxide (CO)
  - Volatile organic compounds (VOCs)

- **Public drinking water parameters**:
  - Disinfection by-products such as
    - Trihalomethanes (THMs)
    - Haloacetic acids (HAAs)
  - Heavy metals (lead, arsenic, etc.)

- **Well water/ground water contaminants**:
  - Herbicides
  - Pesticides
  - Heavy metals (lead, arsenic, etc.)

- **Soil contaminants**:
  - Polychlorinated biphenyls (PCBs)
  - Heavy metals

- **The built environment** (buildings, roadways, etc.) for:
  - Chemical exposures
  - Heavy metal exposures

References

1. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, Chapter 4 Data Variables. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised Mar 2015.
2. CCASS. Guidelines for congenital anomalies surveillance and reporting to CCASS May 2011. The Public Health Agency of Canada, Maternal and Infant Health Section.

D: Coding Guidelines

CCASS recommends collecting cases of congenital anomalies as listed in the ICD-10-CA, Chapter 'Q'.[1] Congenital anomalies should be coded using the most specific ICD-10-CA code available for the congenital anomaly under review. Most of the time this will be a 4-digit code (QXXX). In a few cases, there is only a three-digit code (e.g. Q02 microcephaly, Q36 cleft lip), and sometimes there is a five-digit code (e.g. Q5031 accessory ovary, Q6471 double urethra). In all cases, ***the most specific code that applies should be used*** (e.g. for unspecified renal agenesis use code Q602; *do not use code Q60*).

The ICD-10-CA does not, however, contain detailed instructions on how to define the congenital anomalies to be coded. Generally, a case definition is a set of uniform criteria used to define a disease for public health surveillance and enables public health officials to classify and count cases consistently across reporting jurisdictions.[2] Instructions on defining cases to facilitate the standardization of coding follow below.

Priority Congenital Anomalies

The Public Health Agency of Canada has identified specific cases of congenital anomalies for national reporting that should be prioritized for collection and verification by provinces/territories. To facilitate comparability between jurisdictions, detailed coding instructions have been developed for these particular anomalies and are contained in Appendix 1. The priority congenital anomalies and the corresponding ICD-10_CA Chapter 'Q' codes are as follows:

1. **Neural tube defects:**
    a. Anencephaly and similar anomalies — Q00
    b. Spina bifida without anencephaly — Q05 if not Q00.0
    c. Encephalocele — Q01

2. **Selected central nervous system defects:**
    a. Microcephaly — Q02
    b. Hydrocephaly — Q03
    c. Arhinencephaly/holosprosencephaly — Q04.1, Q04.2

3. **Selected sense organ defects:**
    a. Anophthalmos/microphthalmos — Q11.0, Q11.1, Q11.2
    b. Anotia/microtia — Q16.0, Q17.2
    c. Choanal atresia — Q30.0

4. **Selected congenital heart defects:**
    a. Common truncus — Q20.0
    b. Transposition of great vessels — Q20.1, Q20.3, Q20.5
    c. Endocardial cushion defects/AVSD — Q21.2
    d. Tetralogy of Fallot — Q21.3
    e. Hypoplastic left heart syndrome — Q23.4

	f. Coarctation of aorta	Q25.1
5.	**Oro-facial clefts:**	
	a. Cleft palate (only)	Q35 excluding Q35.7
	b. Cleft lip (only)	Q36
	c. Cleft lip with or without cleft palate	Q36, Q37
6.	**Selected gastrointestinal anomalies:**	
	a. Oesophageal atresia/stenosis, tracheoesophageal fistula	Q39.0—Q39.4 (inclusive)
	b. Small intestine absence/atresia/stenosis	Q41
	c. Ano-rectal absence/atresia/stenosis	Q42.0—Q42.3 (inclusive)
	d. Hirschsprung disease	Q43.1
	e. Atresia of bile ducts	Q44.2
7.	**Selected urinary tract anomalies:**	
	a. Renal agenesis	Q60.0—Q60.2 (inclusive)
	b. Cystic kidney	Q61.1—Q61.5 (inclusive), Q61.8, Q61.9
	c. Bladder and cloacal exstrophy	Q64.1
	d. Lower urinary tract obstruction	Q64.2, Q64.3
8.	**Selected genital anomalies:**	
	a. Cryptoorchidism/undescended testes (excluding infants <35weeks gestational age)	Q53.1, Q53.2, Q53.9
	b. Hypospadias	Q54 excluding Q54.4
	c. Epispadias	Q64.0
	d. Indeterminate sex	Q56
9.	**Limb deficiency defects:**	Q71—Q73
10.	**Diaphragmatic hernia:**	Q79.0
11.	**Prune belly sequence:**	Q79.4
12.	**Selected abdominal wall defects:**	
	a. Omphalocele/exomphalos	Q79.2
	b. Gastroschisis	Q79.3
13.	**Selected chromosomal defects:**	
	a. Down syndrome	Q90
	b. Trisomy 13 (Patau)	Q91.4—Q91.7
	c. Trisomy 18 (Edwards)	Q91.0—Q91.3
	d. Turner syndrome	Q96

Other Congenital Anomalies

For congenital anomalies not on the priority list, there is no universally-accepted list of coding instructions, and practices may differ among programs and jurisdictions. See guidelines from the well-known programs below:

- *NBDPN Guidelines for Conducting Birth Defects Surveillance, Chapter 3 Case Definition*[3]
- EUROCAT *Detailed Congenital Anomaly Coding Guidelines*[4]

Definitions used for coding must be clearly stated when submitting data for the national system and for preparing reports and publications.

Zika-Related Congenital Anomalies

For surveillance of potentially Zika-related congenital anomalies, the National Birth Defects Prevention Network (NBDPN) has published a case inclusion guide. See *Guidelines for Conducting Birth Defects Surveillance, Appendix 3.5 Case Inclusion Guidance for Potentially Zika-related Birth Defects* (updated 12/16).[5]

Multiple Congenital Anomalies and Syndromes

A congenital anomaly may occur as an isolated defect or with other anomalies, and distinguishing between these cases can be important for investigative purposes. The core variables suggested for the CASE initiative include a variable for single/multiple anomalies (see Table 3, variable # 25) as well as a variable for syndromes (see Table 3, variable # 26). However, no definitions are provided.

Discussions during the CASE initiative monthly teleconferences have referenced Garne et al.'s cause-related ('etiologic') classification variable that grouped anomalies as isolated, syndromes, and multiples.[6] However, classification can be complex, requiring consideration of dysmorphologic mechanisms, underlying cause, and whether or not more than one body system is involved. For this reason, the expertise of dysmorphologists and/or geneticists is recommended if possible. See a description and some examples for each classification below:

- **Isolated anomalies:** Cases with one congenital anomaly or a known sequence where multiple anomalies are considered a ***consequence of a single primary anomaly***. These are usually multifactorial in origin and are primarily included in evaluation studies (e.g. determining the impact of folic acid fortification). Examples of 'isolated' anomalies include:

    - a single congenital anomaly (e.g. a case with gastroschisis);
    - a major anomaly with one or more minor anomalies (e.g. absent kidney and a single umbilical artery);
    - an anomaly resulting from a single primary anomaly (e.g. clubfoot due to spina bifida);

- multiple major anomalies due to a single mechanism (e.g. Tetralogy of Fallot [consists of pulmonary stenosis, VSD, overriding aorta and right ventricular hypertrophy] with no other anomalies or recognized syndromic cause, and thus is considered due to the single mechanism of anterior malalignment of the conal septum).

- **Syndromes:** Patterns of unrelated congenital anomalies due to a single cause. These include:
  - chromosomal syndromes (cases with a chromosomal anomaly),
  - monogenic syndromes (cases due to a single gene defect), and
  - environmental syndromes (cases due to a known environmental teratogen).

  For detailed information on syndromes, the *EUROCAT Syndrome Guide* provides a list of syndromes, descriptions, and ICD-10 coding instructions.[7]

- **Multiple major congenital anomalies:** Two or more major congenital anomalies (anomalies having surgical, functional and/or significant cosmetic consequence requiring intervention) that have not been recognized as part of a syndrome or sequence. These cases can be more sensitive for detecting new teratogens (agents that can disturb the development of an embryo or fetus) than monitoring all anomalies or cases with isolated anomalies.[8] Some examples follow:

  - Major anomalies in two or more systems if neither a sequence nor a syndrome can be identified (e.g. cleft lip and hypoplastic left heart);

  - Multiple major defects in a single system if no dysmorphologic link or underlying syndrome can be identified (e.g. a split hand and diaphragmatic hernia, a missing thumb and vertebral segmentation defects, holoprosencephaly and lumbosacral spina bifida).

In cases where congenital anomalies are attributable to a specific cause or recognized pattern, Evans[9] recommends documenting all individual anomalies in addition to the code for the specific cause / recognized pattern, particularly when the anomalies have their own code. For example, in a case with Trisomy 21 (Q90.0) and atrioventricular septal defect (AVSD, Q21.2), coding only the trisomy 21 would underestimate the rate of AVSD. In addition, coding of the AVSD allows further analysis, such as the rate of AVSD attributable to Trisomy 21.[9] Other considerations for coding multiple congenital anomalies include:

- **Discharge Abstract Data:** For jurisdictions using Discharge Abstract Database codes for case ascertainment, it is important to note that the coding of multiple congenital anomalies follows a decision tree, and other congenital anomalies may not always be coded when there is also a syndrome code. You can view the decision tree in the *Canadian Coding Standards for Version 2018 ICD-10-CA and CCI*, *Chapter XVII Congenital Malformations, Deformations and Chromosomal Abnormalities,* page 424.[10]

- **Complex cases:** Cases with multiple major congenital anomalies may be part of a well-established pattern or specific cause, or may be complex and related to an unknown or previously unrecognized grouping of anomalies.[9] For complex cases which are difficult to define, it is recommended that

    - the expertise of dysmorphologists and/or geneticists be consulted, and

    - if the malformations represent a pathogenetically-related disorder but the nature is unclear, *'Q89.7-Multiple congenital malformations, not elsewhere classified'* can be used for easy retrieval and further case review at a later date.[9]

Minor Congenital Anomalies

Minor congenital anomalies are defined as structural changes that pose no significant health problem and usually have limited social or cosmetic consequences for the affected individual.[2] The defining, collecting, and reporting of minor anomalies may differ among programs and data sources. When deciding to include or exclude minor anomalies for collection and reporting, the following questions should be considered:

- **Are there other congenital anomalies present?**
    - As noted above, Evans[9] recommends that all anomalies be captured when there are syndromes or major anomalies present.

- **How will the information be used?**
    - For example, Maternal Fetal Medicine may like to know the types of congenital anomalies associated with single umbilical artery.

- **Is ascertainment feasible?**
    - In the case of Discharge Abstract Database information, minor anomalies that do not meet the criteria for significance may not be coded (see pages 26-27 and 423 in *Canadian Coding Standards for Version 2018 ICD-10-CA and CCI*).[10]

    - Will health professionals consider some anomalies too minor to report (for example, tongue-tie)?

    - If full ascertainment is possible, are there sufficient resources to achieve it?

Jurisdictions may find reviewing the following lists of minor anomalies from other programs helpful:

- *Birth defects surveillance: a manual for programme managers*, Appendix B, page 89 (*External minor congenital anomalies*)[2]

- *NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.3 Examples of Conditions Considered to Be Minor Anomalies*[11]
- EUROCAT *Minor Anomalies for Exclusion*)[12]
- *MACDP's Birth Defects and Genetic Diseases Branch 6-Digit code*, page A-93 (*Exclusion List for the MACDP Non-reportable birth defects*)[13]

References

1. CCASS. Guidelines for congenital anomalies surveillance and reporting to CCASS May 2011. The Public Health Agency of Canada, Maternal and Infant Health Section.
2. WHO/CDC/ICBDSR. Birth defects surveillance: a manual for programme managers. Geneva: World Health Organization; 2014.
3. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, Chapter 3 Case Definition. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised March 2015.
4. EUROCAT (2013). EUROCAT Guide 1.4, Section 3.5: Detailed Congenital Anomaly Coding Guidelines. EUROCAT Central Registry, University of Ulster.
5. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, *Appendix 3.5 Case Inclusion Guidance for Potentially Zika-related Birth Defects*. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised March 2015.
6. Garne E, Dolk H, Loane M, Wellesley D, Barisic I, Calzolari E, Densem J, and a EUROCAT Working Group. Paper 5: Surveillance of multiple congenital anomalies: implementation of a computer algorithm in European registers for classification of cases. Birth Defects Res A Clin Mol Teratol. 2001;91:S44-S50.
7. EUROCAT (2004). EUROCAT Syndrome Guide: Definition and coding of syndromes, Version July 2017. EUROCAT Central Registry, University of Ulster.
8. Khoury MJ, Botto L, Mastoiacovo P, Skjaerven R, Castilla E, Erickson D. Monitoring for multiple congenital anomalies: an international perspective. Epidemiol Rev. 1994;16(2):335-350.
9. Evans JA. Multiple congenital anomalies: issues for birth defects surveillance. J Registry Manag. 2014;41(1):7-12.
10. Canadian Institute for Health Information. Canadian Coding Standards for Version 2018 ICD-10-CA and CCI. Ottawa, ON: CIHI; 2015.
11. NBDPN Guidelines for Conducting Birth Defects surveillance. Appendix 3.3 Examples of Conditions Considered to Be Minor Anomalies.
12. The European Surveillance of Congenital Anomalies. 3.2 Minor anomalies for exclusion [Internet]. 2017 Jun [cited Oct 2014].
13. The Metropolitan Atlanta Congenital Defects Program (MACDP). Birth Defects and Genetic Diseases Branch 6-Digit Code (Revised 5/07)

E: Data Sources

Provinces and territories should evaluate available information sources and determine which will produce a reliable and comprehensive case listing and other variables required within the resources available. Ideally, this would be done through a systematic evaluation of sources or a literature review. See Appendix 2 for some suggestions of literature to review. To facilitate data source review, see the NBDPN's *Data Sources Descriptive Assessment Tool*.[1]

Recommendations

Using multiple sources for case ascertainment as well as documenting the sources will facilitate the collection of good-quality information. CCASS guidelines[2] recommend that, at a minimum, jurisdictions should use the following data sources for case ascertainment:

- vital statistics (including live births, deaths, and stillbirths),
- hospital discharges (including pregnancy terminations), and
- physician notices of birth.

CCASS also recommends the following additional sources for case ascertainment, if feasible:

- genetics clinics,
- cytogenetic laboratories,
- provincial newborn screening programs, and
- maternal screening programs.

Understanding Case Ascertainment Sources

Sources for case ascertainment can be grouped as follows:

- **Administrative Data**: This is secondary information containing diagnoses and other information that has been collected for another purpose. Although it has potential for errors, it is a common, efficient approach that can be consistent across jurisdictions and has potential for linkage to other data[3]. Some examples include:
    - hospital discharge data,
    - vital statistics records,
    - physician billing data, and
    - ambulatory care reporting systems information.

- **Diagnostic testing results**: Sources for results from diagnostic testing include cytogenetic laboratories, newborn screening programs, non-invasive prenatal testing, diagnostic imaging, pathologist's reports, or maternal serum screening programs.

- **Outpatient clinics/programs data:** These can include specialized clinics such as pediatric cardiology, maternal fetal assessment, genetic referral, and newborn screening programs.

- **Case reports:** Some jurisdictions may receive reports of cases of congenital anomalies from physicians, laboratories, hospitals, clinics, and medical examiners/coroners.

Table 5 provides details of the characteristics of these sources.

Table 5: Sources for Congenital Anomalies Case Ascertainment

Source	Characteristics
Hospital Discharge Abstract Data (DAD) (Administrative data)	• Administrative/clinical/demographic information on hospital discharge • Greatest capacity for ascertainment although has weaknesses • Discharge abstract information is usually available in a database • Adequately captures major and obvious anomalies[4,5] • Maternal 'O35' codes can flag terminations for congenital anomalies • May be possible to have the system capture perinatal information as well • Cost effective and efficient • Supports the aggregation of data • May assist in monitoring temporal and geographical trends and variations, strategic planning, or program evaluation[6] ***Disadvantages:*** • Produces a high number of false positives and false negatives • Captures 'cases' better than specific defects • Biased towards major anomalies detected at birth • Does not capture outpatient diagnoses • Has errors, as some testing results that could change the discharge diagnosis may not be available until after discharge • Coding is not verified and may contain errors and/or practice differences
Vital Statistics Records (Administrative data)	• Legislated collection, usually birth, stillbirth, and death information • Can increase sensitivity of data collection[7] ***Disadvantages:*** • Quality depends on jurisdiction process; some are more/less rigorous • Can significantly underestimate prevalence if used alone as more likely to detect major anomalies easily diagnosed at birth; Alberta found it to under-report by about 20% • Jurisdictions may differ in registration requirements, e.g. stillbirth definitions
Physician Billing Data (Administrative data)	• Reports ICD codes for fee-for-service physician billings • Contains a wealth of information • Can be used to cue a medical record review for possible cases ***Disadvantages:*** • ICD coding version and information may vary by jurisdiction • Can result in many false positives if not verified[4]

Source	Characteristics
Ambulatory Care Reporting Systems (Administrative data)	• Includes information on all hospital and community-based ambulatory care (Surgery, ER, Community clinics, etc.) • Useful for capturing cases diagnosed after birth and as an outpatient • Can cue a medical record review for possible cases ***Disadvantages:*** • Not all jurisdictions have this system, and scope can differ if they do • Can have poor validity even for obvious anomalies[3] • Coded by medical coders with the same limitations as for the 'DAD' data
Diagnostic Testing Reports	• Includes cytogenetic testing, newborn screening, prenatal testing, diagnostic imaging, pathology, maternal serum screening, etc. • Some places may have a central database that can be accessed • If no central database, may need to connect directly with public or private laboratories. ***Disadvantages:*** • Certainty of diagnosis depends on anomaly, gestational age, and test performed; some follow-up may be needed to confirm diagnoses • May be complicated to access in some jurisdictions
Clinic/Program Records	• May be accessible through an ambulatory care database. • Other access possibilities include being copied in on referrals to pediatric specialty clinics (Alberta) or providing bulk data transfers (Newfoundland and Labrador). • Can include all medical records available in clinic charts • Highly reliable (particularly for specialty pediatric clinics, MFAU clinics, coroner records, etc.) • Useful for capturing cases diagnosed as an outpatient • Can verify cases that were initially a query diagnosis • Can be used to seek out information on specific categories of defects ***Disadvantages:*** • For some clinics/programs, accessing records may take some work due to volume - a cue may be needed to indicate which records to review • Collating the records in a dataset format can be challenging
Case Reports	• Reporting of cases by individuals or institutions (physicians, laboratories, hospitals, clinics, medical examiners, coroners) • Access through paper, online reporting, or electronic data transfers • Value depends on level of participation; best if reporting is 'mandatory' • Participation can be encouraged by disseminating surveillance reports, visiting data sources (e.g. a children's hospital), delivering conference presentations, or developing publications for academic journals ***Disadvantages:*** • Likely underreports cases, especially if not mandatory • Staff must remember, and have time, to send the information • Staff may be unsure which cases meet the criteria for submission • Can be viewed negatively as an imposed burden on a busy clinic

Sources for Environmental Exposure Data

The collection of environmental exposure data is recommended to provide information for the investigation of causes and contributing factors of congenital anomalies (see Table 2, variable # 2). Exposure information may be collected at the individual or population level, as follows:

- **Individual-level**: May be reported by the mom to her health care provider and may be found on the prenatal record or clinic/hospital medical records.

- **Population-level:** For example, by community, site, etc. These may be reported by the mom to her health care provider if she is aware of an exposure, or identified through review of data reports from public health and other monitoring programs.

Most jurisdictions should have some publically-available sources for population-level environmental exposure information. Some suggestions are shown in Table 6.

Table 6: Population-level Data on Environmental Pollutants

Element	Possible Data Sources
Air Quality	• Environment Canada National Air Pollution Surveillance Program (NAPS) • Provincial/territorial (P/T) ambient air monitoring reports • P/T air zone management reports • Refinery emission reports • Local/regional studies
Public Drinking Water	• Municipal drinking water monitoring programs • P/T annual drinking water reports • P/T special parameters monitoring reports • Local/regional studies
Well Water/ Ground Water	• Provincial/territorial monitoring program reports • Local/regional studies
Waste Water	• Industrial effluent monitoring programs • Local/regional studies
Soil and Sediments	• Federal Contaminated Sites Inventory • Provincial contaminated sites inventory • Site-specific contamination remediation reports • Road-side spraying programs • Local/regional studies
Built Environment	• Local/regional studies

References

1. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, Appendix 7.1 Data Sources Descriptive Assessment Tool. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised Mar 2015.
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F: Collection Protocols

Types of Collection Methods

There are three methods for collecting information on congenital anomalies:

1. **Active** – Medical records are directly accessed and cases are ascertained from the records.
2. **Passive** – Cases are ascertained from secondary sources collected for other purposes or from case reports.
3. **Hybrid** – A combination of active and passive methods.

Characteristics and challenges for each method are detailed in Table 7.

Table 7: Methods for Data Collection and Case Ascertainment

Method	Method Characteristics
Active	• Cases are ascertained from medical information directly accessed by trained personnel. • Can be achieved through site visits or viewing electronic medical records. • Multiple sources are used (e.g. logbooks, death records, medical reports). • Both maternal and infant records are reviewed, improving data quality. ***Disadvantages:*** • Requires significant resources, personnel and time. ***Example:*** • Metropolitan Atlanta Congenital Defects Program *(MACDP)*

Passive	• Cases are either:
	  ○ ascertained from administrative databases (vital statistics, discharge data, etc.), or
	  ○ ascertained from case reports by medical staff based on a specific criteria.
	• Case report. The importance of reporting can be publicized through site visits, presentations, surveillance reports, publications, etc.
	• Requires less resources and personnel than other methods.
	
	Disadvantages:
	• Data quality depends on others who collected and input the information.
	• Documentation may be incomplete.
	
	Examples: CCASS, Florida Birth Defects Registry *(FBDR)*

Hybrid	• Cases are passively ascertained, then verified with medical records. • Can allocate resources based on priorities – only anomalies of interest get verified further. ***Examples:*** Alberta Congenital Anomalies Surveillance System *(ACASS);* Newfoundland and Labrador Congenital Anomalies Surveillance system *(NLCASS);* Utah Birth Defect Network *(UBDN)*

Data Abstraction Form

When collecting data directly from a medical record, an abstraction form will be required to record the information for later entry into the surveillance system. Ideally, this form will be developed locally to ensure the data collected will be sufficient to meet the system's goals.

In general, the following type of information should be included in an abstraction form:

- **Case ascertainment information:**
  - Text description of the congenital anomalies (CCASS recommends the use of text to describe conditions rather than relying on codes alone)[1]
  - The type of diagnosis (established, suspect/query)
  - Date and source of report and/or test confirming the diagnoses
  - ICD-10-CA code / OMIM code

- **Variables for transfer to PHAC:**
  - Mom's resident postal code at the time of outcome
  - Type of outcome (live birth, stillbirth, aborted, terminated for fetal anomalies)
  - Birth information (weight, gestational age, number of babies/fetuses)
  - Date of death

- **Baby Identification** (for linking information and ruling out duplicates):
  - Name
  - Medical record #
  - Date of birth/outcome
  - Sex

- **Mom Information** (for linking information and ruling out duplicates):
  - Name
  - Medical record #
  - Date of birth
  - Place of residence

- **Other variables**:
  - Other information chosen by the program
  - Collection information (source, date collected, person collecting)

This form can also be used by medical professionals when making case reports to the system, or for ascertainment from secondary sources if data is not directly downloaded. An example of an abstraction form is provided in *Birth defects surveillance – a manual for programme mangers*, Appendix G.[2]

Collecting from Hospital Discharge Data

The main source for ascertaining congenital anomalies from the Discharge Abstract Database (DAD) is the ICD-10-CA 'Chapter 'Q' diagnosis codes. However, the DAD diagnosis variable (*Dx*) contains all diagnoses, including query diagnoses.

To establish the status of the diagnosis as a query or rule-out diagnosis, a 'Q' is entered as a prefix to the diagnosis (see the *Canadian Coding Standards for Version 2018 ICD-10-CA* (pp.77 to 78).[3] The prefix is contained in a second prefix variable (*DxPrefix)*. As query diagnoses should not be included in prevalence data, it is imperative that the prefix variable be collected along with the associated diagnosis variable in order to remove the query diagnoses before reporting. If resources permit, the query diagnosis should be investigated to determine if a diagnosis was ultimately established.

Figures 1 and 2 show the diagnoses variables in a 3M hospital data system, and on an SPSS spreadsheet.

Figure 1: 3M Hospital Data Management System Diagnosis Variables

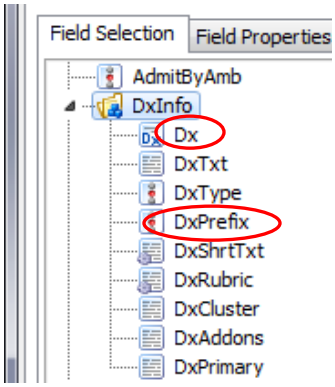


Figure 2: Diagnosis Variables in an SPSS Spreadsheet

	Dx1	Dx2	Dx3	DxPrefix1	DxPrefix2	DxPrefix3
1	Q058	Q070	Q658			
2	P285	Q639	Q058		Q	Q
3	Q224	Q059	Q270		Q	
4	Q059	Z3800		Q		

Record 1: All Dx are established (no Q);
Record 2: Q639 and Q058 are query diagnoses (Q in Prefix2 and Prefix3)
Record 3: Q059 is a query diagnosis (Q in prefix2);
Record 4: Q059 is a query diagnosis (Q in prefix1).

In Figure 1: The diagnosis is contained in 'Dx'; the Query status is contained in 'DxPrefix'.
In Figure 2: The four diagnoses with a 'Q' in the 'DxPrefix' (circled) are not established.

In Figure 2, the diagnoses with a 'Q' in the associated DxPrefix variable are not established (circled codes), and should not be included in prevalence data unless further investigated to verify (note: Quebec does not use query diagnosis codes in their discharge data).

Other baby diagnoses codes. If feasible, it may be useful to review other baby diagnoses codes with possible associated congenital anomalies to ascertain cases that may not have been coded in the Discharge Abstract data. These include:

- **Congenital syphilis** (A50)

- **Fetus/newborn affected by maternal use/exposure to:**
  - Tobacco (PO4.2)
  - Alcohol (P04.3), not including FASD (Q86.0)
  - Drugs of addiction (P04.4), not including withdrawal symptoms (P96.1)
  - Nutritional chemical substances (P04.5)
  - Environmental chemical substances (P04.6)
  - Other maternal noxious influences (P04.8, P04.9)

- **Congenital infections:**
  - Rubella syndrome, including congenital rubella pneumonitis (P35.0)
  - Cytomegalovirus infection (P35.1)
  - Herpes viral [herpes simplex] infection (P35.2)
  - Viral hepatitis (P35.3)
  - Other viral diseases, including varicella/chickenpox (P35.8)
  - Congenital viral disease, unspecified (P35.9)

- **Syndrome of infant of a diabetic mother** (P70.1):
  - Includes: Fetus or newborn with hypoglycaemia affected by pre-existing maternal diabetes mellitus

Using Mom Hospital Discharge Diagnoses

Some maternal diagnosis codes can suggest that a pregnancy outcome has been affected by congenital anomalies. These are '*O35'* codes - *Maternal care for known or suspected fetal abnormalities and damage.* The codes and related anomalies follow.

- **Suspected congenital anomalies:**
  - Anencephaly (O35.001 to .009)
  - Spina bifida (O35.011 to .019)
  - Hydrocephalus (O35.021 to .029)
  - Spina bifida with hydrocephalus (O35.031 to .039)

- Other neural tube defects (O35.081 to .089)
- Central nervous system malformation, unspecified (O35.091 to .099)
- Chromosomal abnormality (O35.101 to .109)
- Hereditary disease (O35.201 to .209)

- **Suspected damage to fetus:**
  - Viral disease in mother (O35.301to .309)
  - Alcohol (O35.401 to .409)
  - Drugs (O35.501 to .509)
  - Radiation (O35.601 to .609)
  - Other medical procedures (O35.701 to .709)

- **Other fetal abnormality and damage:** (O35.801 to O35.909)

The 'O35' codes are particularly useful for identifying early terminations for fetal anomalies (TOPFA) that did not result in a baby record and therefore would not be captured in the Discharge Abstract data. Additional information is required, however, to establish the final diagnosis/diagnoses before being included in prevalence data.

Accepted Prenatal Diagnoses

CCASS recommends that if prenatal diagnoses in terminations or stillbirths cannot be verified because of the method of termination and/or condition of the specimen, lack of autopsy or post termination/ postnatal investigation (karyotyping, x-rays, etc.), ***no data entry is to be done.***[1]

In some obvious cases, unverified prenatal diagnoses can be accepted. For the priority congenital anomalies identified for national reporting (listed in Section D), see instructions for each specific anomaly in the case definitions in Appendix 1. For other congenital anomalies, the Canadian Congenital Anomalies Surveillance System (CCASS) guidelines suggests the following may be accepted:[1]

- **Prenatal *diagnostic* tests:** These are diagnostic tests used to detect chromosomal abnormalities and can be used for case ascertainment purposes. These include
  - Amniocentesis (Amnio), which tests cells from the amniotic fluid, and
  - Chorionic villus sampling (CVS), which tests cells from the placenta.

- **Clearly defined ultrasound diagnoses:** These can be accepted as definitive if:
  - The diagnosis has been made in a special prenatal diagnostic centre, e.g., Maternal Fetal Medical Unit etc.
  - Such diagnoses include obvious neural tube anomalies, anencephaly, definitive hydrocephaly, hydranencephaly, clearly demarcated limb defects, certain heart defects, and absence defects, e.g., renal, limb, etc.

  - For an ultrasound diagnosis of definitive hydrocephalus, no measurement is required (this may differ from other programs such as EUROCAT which only accepts an ultrasound diagnosis of hydrocephalus if it is >15 mm).

- **Ultrasound *'query'* diagnoses for verification:** Some diagnoses (but not the soft marker signs) may be accepted on a *temporary basis* for verification:
  - These would be coded with an ICD code, with the letter "U" added at the end of the code (for example, Q35.0U) to indicate the diagnosis is uncertain.
  - These would be followed up to either confirm or refute the diagnosis.
  - If the diagnosis cannot be confirmed, a decision has to be made to either keep it with the U code or discard it.
  - ***Any entry with a U code is not to be included in prevalence data.***

Rejected **prenatal diagnoses.** ***Do not include*** the following for case ascertainment:

- **Undefined ultrasound diagnoses.** For example, ***do not include***
  - renal, pelvi-calyceal or ureteral dilatation (this may differ from other programs such as EUROCAT which accepts renal dilatation of >10 mm);
  - ventriculomegaly;
  - soft marker signs, e.g., lemon, banana, nuchal thickening, echogenic foci in heart or bowel. [Note: Soft markers are an indicator of risk and may lead to additional testing].

- **Prenatal *screening* tests** (these assess risk only). For example, ***do not include*** the following:
  - First Trimester combined screening (FTS) - Nuchal translucency measurement and blood sample between 10 – 13 weeks gestation;
  - Second trimester quad screen (Blood samples taken between 15 – 20 weeks gestation);
  - Serum integrated prenatal screen (SIPS) - Two blood tests taken between 10-13 weeks and 15-20 weeks gestation;
  - Integrated prenatal screen (IPS) - Nuchal translucency measurement and blood work in the first trimester and second trimester;
  - Non Invasive Prenatal Screening (NIPS) detects fetal DNA in maternal blood and identifies an increased or decreased risk for having a baby with chromosomal anomalies.

When Sources Conflict

When using more than one source for case ascertainment, conflicting records may emerge. In such cases the following should be considered:

1. Congenital anomalies reported on surgical/operative notes or autopsy/pathology reports are considered more accurate than those reported on diagnostic imaging records, particularly prenatal ultrasounds.

2. Clinic diagnoses should be trusted even if not recorded in the Discharge Abstract Database (DAD) birth information. For example, a pediatric urology clinic diagnosis of a kidney malformation would likely only become apparent as the child aged and therefore may not be recorded on the DAD record from the birth.

3. Diagnoses recorded on the Physician Notice of Birth should be trusted even if not recorded in a subsequent readmission. For example, polydactyly diagnosed at birth may not be recorded in a readmission DAD record because it had been corrected surgically prior to admission, or it may not meet the ICD-10-CA Coding Standards for inclusion in the DAD for that particular admission.

4. A medical geneticist or another subject matter expert should be consulted to resolve particularly challenging cases.

References

1. Canadian Congenital Anomalies Surveillance Network. Guidelines for congenital anomalies surveillance and reporting to CCASS, May 2011. Ottawa: Public Health Agency of Canada, Maternal and Infant Health Section.
2. WHO/CDC/ICBDSR. Birth defects surveillance: a manual for programme managers. Geneva: World Health Organization; 2014.
3. Canadian Institute for Health Information. Canadian Coding Standards for Version 2018 ICD-10-CA and CCI. Ottawa, ON: CIHI; 2015.

G: Quality Processes

High-quality data is essential for accurately monitoring and investigating causes and contributing factors for congenital anomalies. The Canadian Congenital Anomalies Surveillance System (CCASS) guidelines[1] for the CASE initiative provide the following instructions for data quality:

1. Data maintenance and validation will be performed at the provincial/territorial level.

2. For national reporting, in the event that the CCASS identifies inconsistencies with the data
    a. discrepancies will be flagged,
    b. the reporting party will be notified of the discrepancy, and
    c. the national data will be updated. Notification that this has been done will be forwarded to the jurisdiction so that the local system can be updated.

Some general information on quality processes and protocols follows.

Producing Quality Data

Many issues can impact data quality. These include[2]

- missing values (empty data fields),
- diagnosis errors (description or coding errors),
- duplicate cases (especially if ascertaining from multiple sources), and
- biased representation (not all cases are included). For example, including cases from outside the system's geographic limits, or limiting cases to those that are very severe, from urban settings, or from the private sector.

Methods for ensuring and improving data quality will vary depending on the type of case ascertainment used. However, regardless of the type of case ascertainment, an ongoing data quality improvement cycle is essential for preventing errors, correcting errors when they do occur, and minimizing future errors. In general, a continuous quality improvement cycle should have the following protocols:

- **Error prevention protocols** (sometimes called quality assurance): These are protocols for producing good quality data and preventing errors:
    - Use standardized case definitions.
    - Develop multiple sources for case ascertainment.
    - Develop clear case ascertainment methods and protocols.
    - Ensure coding instructions are clearly defined.
    - Train data collection staff appropriately.

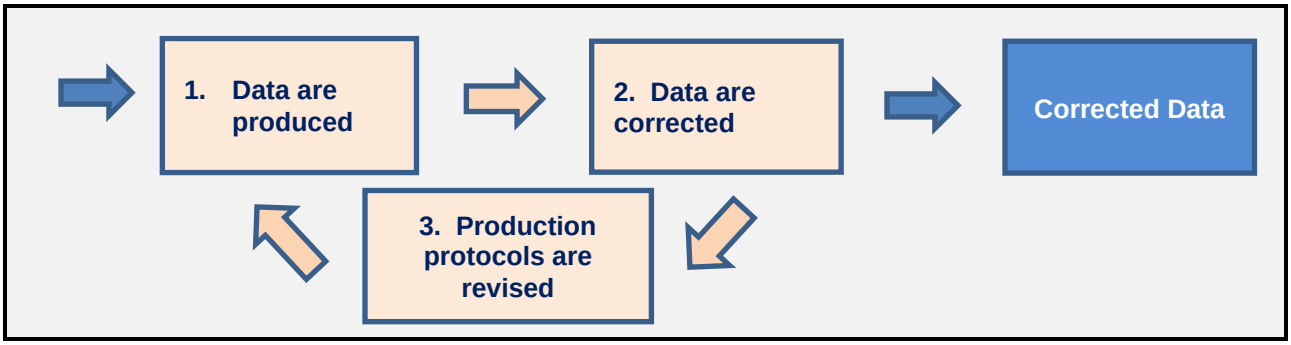
- **Error detection protocols** (sometimes called quality control): These are protocols for reviewing and correcting data that has been produced to ensure the final result is accurate:[3]

- Repeat case finding activities and compare results (re-case finding).
- Repeat abstracting activities and compare results (re-abstracting).
- Compare medical record to reported cases (validity audits/medical records reviews).
- Have case records reviewed by a clinical expert and compare results (clinical review).
- Perform reliability and inter-rater agreement checks.
- Calculate timeliness measures.
- Evaluate data sources.
- Compare and verify data with multiple sources.
- Use computer technology.

- **Error reduction protocols** (sometimes called quality improvement): These are protocols to improve data quality by revising data production protocols based on the nature of errors detected:
  - Determine the cause of the error detected (root cause analysis).
  - Identify possible solutions and feasibility of each.
  - Implement chosen solution. Some examples:
    - Clarification of coding instructions when ambiguous wording has resulting in coding errors and/or inconsistency, and
    - Additional staff training when there are specific recurring errors.

The data quality improvement cycle is illustrated in Figure 3:

Figure 3: Continuous Quality Improvement Cycle



For more detailed information on improving data quality, including specific methods, outcome measurements, and frequency, see the NBDPN *Guidelines for Conducting Birth Defects Surveillance, Chapter 7 Data Quality Management* (Subsection 7.8).[3]

Assessing Data Quality

Quality indicators can provide a means to quickly evaluate data quality and identify areas that need improvement, as well as provide a means for evaluating and comparing registries. Indicators may be prioritized based on program objectives and the resources available. As systems may vary in methods, scope, purpose, objectives, and resources, attributes important to one system may be less important to another. Programs will, therefore, differ on the attributes monitored. A comparison of some well-known systems shows the following three attributes in common:

- Completeness
- Accuracy
- Timeliness

See Table 8 for other indicators used by these systems.

Table 8: Comparison of Suggested Data Quality Indicators by Program[2,3,4]

Suggested Data Quality Indicators	WHO/CDC/ ICBDSR[2]	NBDPN[3]	EUROCAT[4]
Completeness (all-inclusive and comprehensive data)	x	x	x
Accuracy (exact, correct, and valid data)	x	x	x
Timeliness (data collected and analyzed in a timely manner)	x	x	x
Oriented (focused, targeted and intended)		x	
Measureable (quantifiable, calculable, and objective)		x	
Applicable (relevant)		x	
Comparability (how one dataset conforms with others)		x	
Thoroughness (meticulous and exhaustive data collection)		x	
Outcome measurements (evaluate targets, goals, benchmarks)		x	
Ascertainment (regions compared to system average)			x
Denominator information (completeness of denominator data)			x

Data quality assessment tool. The National Birth Defects and Prevention Network (NBDPN) has developed an easy-to-use *Data Quality Assessment Tool*[5] that focuses on the three common attributes from Table 8:

- Completeness
- Accuracy
- Timeliness.

The tool can provide a broad measure of the congenital anomalies surveillance system for the CASE initiative, even though it is designed for systems in the United States. See summary measures for the tool in Table 9.[5]

Table 9: Summary Items for the NBDPN Quality Assessment Tool[5]

No.	Quality Assessment Item	System Level*				Total
		0	1	2	3	
Completeness						
1.1	Types of data sources used systematically and routinely to identify potential cases at a population-based level					
1.2	Birth defects included using standard NBDPN case definitions					
1.3	Pregnancy outcomes included					
1.4	Systematic and routine identification of cases during ascertainment period					
1.5	Data elements collected					
Timeliness						
2.1	Time of case data completion for NBDPN "core" list					
2.2	Time of case data completion for NBDPN "recommended" list					
Accuracy						
3.1	Data quality procedures for verification of cases diagnosis					
3.2	Scope of birth defects verified					
3.3	Level of expertise for individuals who perform case diagnosis verification					
Total Possible Score (for 10 questions) = 30						

* System Levels: 1 = Rudimentary; 2 = Essential; 3 = Optimal

Measuring the quality of regional submissions. EUROCAT (European Surveillance of Congenital Anomalies) has developed measures to quickly assess the quality of regional data submissions to the national system. Each measure is applied to the regional submissions, and the results are compared to the average of all submissions in the system; any significant differences require an explanation.[4] Table 10 provides details of the indicators and associated measures.

Table 10: List of EUROCAT Data Quality Indicators [4]

EUROCAT Data Quality Indicators *(compares each registry to system average)*	
Ascertainment	1. Total number of cases 2. Total congenital anomaly prevalence per 10,000 births 3. Spina bifida: Anencephaly ratio 4. Neural tube defect prevalence per 10,000 5. Selected cardiac anomalies prevalence per 10,000 6. Selected postnatal diagnosis prevalence per 10,000 7. Non-chromosomal syndrome prevalence per 10,000 8. Down syndrome: Observed: Expected ratio by maternal age 9. Prevalence malformed fetal death or stillbirth 20-27 weeks gestation per 10,000 10. Prevalence malformed stillbirth ≥ 28 weeks gestation per 10,000 11. Missing stillbirth gestational age (%)
Accuracy of diagnosis	12. Multiple malformations among non-syndromes or non-chromosomal (%) 13. Stillbirths with postmortem examination (%) 14. Stillbirths with postmortem examination – results known (%) 15. TOPFA with postmortem examination (%) 16. TOPFA with postmortem examination – results known (%) 17. Chromosomal anomalies with karyotype performed (%) 18. Chromosomal anomalies with karyotype performed – results known (%) 19. Chromosomal anomalies with karyotype text (%) 20. Non-chromosomal multiple malformations with known karyotype (%) 21. Down syndrome with congenital heart defects or duodenal atresia (live births) (%) 22. Prevalence selected ICD-10 Q-chapter codes per 10,000 23. Prevalence selected unspecified ICD-10 Q-chapter codes per 10,000
Completeness of information	24. Average number of core variables 90% complete 25. Average number of noncore variables 80% complete 26. Syndrome text complete (%) 27. Malformation text complete (%) 28. Number of registries with no other text information available
Timeliness	29. Number of registries that transmitted data between a defined period
Denominator information	30. Number of registries with 80% of maternal age denominators by 5-year groups 31. Number of registries with monthly denominators

Evaluating System Effectiveness

To ensure a surveillance system is monitoring events of public health importance efficiently and effectively, the whole system should be evaluated periodically.

The Centers for Disease Control and Prevention has developed a step-by-step checklist for Evaluating Public Health Surveillance Systems describing the tasks needed to perform a comprehensive evaluation (see *Updated Guidelines for Evaluating Public Health Surveillance Systems*, Appendix A)[6]. When using the checklist, programs should focus on the attributes that are most important for meeting their objectives.

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1. Canadian Congenital Anomalies Surveillance Network. Guidelines for congenital anomalies surveillance and reporting to CCASS, May 2011. Ottawa: Public Health Agency of Canada,cy of Canada, Maternal and Infant Health Section.
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H: Analysis & Reporting

A discussion of epidemiology is beyond the scope of this document. For a short, introductory text, see *Epidemiology for the Uninitiated, 5th Edition.*[1] Some basic information on the analysis and reporting of congenital anomalies surveillance data is provided below.

Identifying Targeted Users

To ensure that the data collection, analysis and reporting plan will provide the information needed to meet program objectives, it is important to identify targeted users and their needs during the program development stage. Although programs will differ among provinces and territories, considerations should include:

- **Targeted/potential users of the information**. These may include:
  - Health care providers
  - Program epidemiologists
  - Researchers
  - Health Departments
  - Specialty programs
  - Others

- **User information needs**. These may include:
  - Counts, rates, and trends of specific congenital anomalies
  - Cluster analysis
  - Associations with individual characteristics, prenatal conditions, environmental exposures
  - Others

- **Preferred reporting format**. Results can be presented in a wide variety of formats, for example:
  - Raw data (for those doing research)
  - Charts, tables, graphs, and diagrams
  - Infographics
  - Mapped data
  - Videos
  - Others

- **Preferred dissemination method.** These may include:
  - Data transfer (for researchers)
  - Reports, pamphlets and posters (paper or electronic)
  - Presentations at events, webcasts, YouTube videos
  - Media and social media venues
  - Others

The NBDPN *Guidelines for Conducting Birth Defects Surveillance, Appendix 11.3 Data Users Matrix*[2] provides detailed examples of types of users, questions each type of user may need answered, and types of information that may be required to answer the questions.

Calculating Rates

Basic congenital anomalies reporting commonly consists of counts, rates, ratios, and trends over time. Calculation of clusters, as well as risk related to individual characteristics, prenatal health, and exposures may also be undertaken depending on user needs.

An in-depth discussion of statistical procedures is outside the scope of this document. However, some basic calculations commonly used for reporting on congenital anomalies follow.

- **Number of cases of congenital anomalies:**
  - For ***specific*** anomalies, count one case for ***every occurrence***.
  - For ***grouped*** anomalies, count one case for ***every baby/*fetus**, regardless if the baby/ fetus has more than one anomaly in that group (e.g. heart defects – many babies/fetuses have more than one congenital heart defect, but should be counted only once for the group *Congenital Heart Defects)*.

- **Prevalence at birth:** A common surveillance rate is the '*prevalence at birth'* (the term *'incidence'* is not used because of unknown spontaneous losses).[3] Prevalence at birth is commonly calculated as follows:
  - $\frac{\text{\# cases with anomaly}}{\text{\# live births}} \times 10{,}000 \text{ (sometimes 1000)}$
  - Prevalence can be standardized for the unequal distribution of variables in the population (for example, sex, maternal age at delivery, etc.)
  - See the NBDPN *Guidelines for Conducting Birth Defects Surveillance*, *Chapter 8 Statistical Methods* (Subsection 8.6), for more information on calculating standardized rates.[3]

- **Relative risk**: The '*Relative Risk'* calculation can be used to compare the rate of having a specific condition for one group (for example, smokers) with the rate of having the same condition in a comparison group (for example, non-smokers). It is calculated as follows:
  - $\frac{\text{(\# cases in group)}}{\text{(total \# in group)}}$ divided by $\frac{\text{(\# cases in comparison group)}}{\text{(total \# in comparison group)}}$
  - A result of 1 = similar risk, > 1 = greater risk, and < 1 = less risk.

- **Confidence intervals:**
  - Are designed to compensate for measuring only a sample of the population.
  - Provide a range to ***estimate*** the outcomes if different samples were measured.

- For larger samples, measures may be closer to the true measure so range is narrower.
- For smaller samples, measures may be further from the true measure so range is wider.
- The confidence ***level*** reflects how often the true value is expected to lie in the range.
- At a 95% confidence level, the true value should fall within the range 95% of the time.
- As the confidence level increases, the range gets wider.

Technically, if measuring the whole population (and not a sample), the result is the true measure of the population and confidence intervals are not applicable. Some epidemiologists maintain that even if the whole population is measured, the population is necessarily a subsample of a larger population. However, the subsample may not be a random sample, allowing for systematic error or bias and complicating interpretation. There is, therefore, some controversy over the use of confidence intervals with population data and practices may differ by jurisdiction.

Because confidence intervals provide a range of values, they can be useful when extremely small numbers cause rates to be unstable[3] (this can happen when a change of only a few cases can cause a large change in rates). For more information and resources on using and calculating confidence intervals, see the NBDPN *Guidelines for Conducting Birth Defects Surveillance*, *Chapter 8 Statistical Methods* (Subsection 8.6)[3]

Interpreting Results

Caution should be used when interpreting results, as there are a number of issues than can cause differences in rates other than a true difference in prevalence. Some examples follow.

- **Measures used.** When comparing calculations, whether with prior years or other jurisdictions, it is important to ensure that comparable measures are used. Considerations include:
  - ***Case ascertainment*:** Does the period differ (at birth, up to one year of age, etc.)?
  - ***Case definition**:* Are the same case definitions used for coding/counting?
  - ***Numerator**:* Are the same outcomes used for case ascertainment (for example, live births, stillbirths, termination for fetal anomalies)?
  - ***Denominator**:* Are the same outcomes used to calculate the denominator (Live births, stillbirths)?
  - ***Multiplier**:* Is the same multiplier used (is it per 1,000 births, per 10,000 births, other)?

- **Changes in medical diagnoses and technologies**. For example:
  - The birth prevalence of some disorders may increase due to new technologies facilitating diagnosis.
  - Because diagnosis and coding changes can affect prevalence rates, such changes should be recorded in detail and reported with the data to facilitate interpretation.

- **Changes in reporting and case ascertainment.** For example:
  - As hospitals shift to computerized diagnostic indices, conditions that appear to be minor to medical records staff may be omitted if there are a limited number of ICD-9-CM codes retained in the index (due to a limited number of fields for codes).

- **Changes in populations at risk.** For example:
  - Population demographics such as maternal age, prevalence of pre-existing diabetes, etc. can change over time.
  - Current, detailed population estimates will need to be examined to identify possible changes.

- **Random variation:**
  - This can be especially noticeable when case numbers are small.
  - Using confidence intervals may be helpful when dealing with small numbers.

Presenting Data

The methods for presenting and disseminating results should be developed based on user needs. In the case of multiple users, multiple reporting and dissemination methods may be developed. For detailed information, see the NBDPN *Guidelines for Conducting Birth Defects Surveillance, Chapter 11 Data Presentation*.[4]

In addition to user needs and dissemination methods, maintaining privacy and facilitating interpretation must be considered when developing plans for presentation and dissemination.

Privacy. All reporting and/or sharing of data must follow the guidelines set out in the *Privacy Impact Assessments* and *Data Sharing agreements* of the reporting jurisdiction (these may vary among provinces and territories). For small numbers:

- In general, the Public Health Agency of Canada does not report numbers between 0 and 5 (values from 1 to 4).
  - Provinces and territories may differ in their limitations.

- There are various methods to deal with small numbers prohibited from reporting:
  - Omit small counts, as well as another cell to ensure the suppressed cell cannot be recalculated (cell suppression).
  - Combine cells from several groups.
  - Combine years.

For additional information see the NBDPN *Guidelines for Conducting Birth Defects Surveillance, Appendix 11.1 Data Suppression.*[5]

Interpretation. Reports should include enough information to ensure that users can accurately interpret the information provided. This includes:

- ***Details of the program***. For example:
  - Pregnancies followed (resident mothers, hospital outcomes)
  - Outcomes collected (live births, stillbirths, terminations for fetal anomalies)
  - Ascertainment period (fiscal or calendar year of outcomes, follow-up period)
  - Case ascertainment sources (Discharge Abstract Database, medical records, etc.)
  - Data collection methods (direct ascertainment, case reporting, etc.)

- ***Congenital anomaly definitions***. This includes:
  - Case definitions
  - Coding used
  - Changes in diagnosis and reporting in the period of the report (including changes in ICD version/coding practices)

- ***Data analysis methods*** including:
  - Case linkages
  - Denominator data
  - Treatment of missing information
  - Analysis procedures
  - Cell suppression
  - Analysis limitations

Submitting Data to the National System

The following submission recommendations have been collated from CASE documents, presentations, and discussions.

- **Data Submission Protocols:**
  - An ASCII file format is preferred (other formats may be accepted).
  - Data will normally be transferred through PHAC's secure file transfer protocol (FTP).
  - Other methods may be used if agreed-upon (for example, the Newfoundland and Labrador transfer is through Eastern Health's secure email).

- **Data Submission Information.** Data submission should be accompanied by the following information (in either Word, Excel, WordPerfect, or PDF format):
  - Denominator aggregate data (for all births for each year of data)
  - CCASS National Variables Record Layout Form (describes the variable characteristics)
  - CCASS Data Transfer Form (describes the data submitted)

See examples of the above in Tables 11, 12, and 13 below.

Table 11: Denominator Data Form for National Submission

CCDASS Denominator Data Record Layout								
Reporting Calendar Year: YYYY								
Date of Extraction: MM-DD-YYYY								
Reported numbers must include both babies born with or without Congenital Anomalies								
Variable	*Categories*	*Live births*			*Stillbirths*			
		Males	*Females*	*Undetermined*	*Males*	*Females*	*Undetermined*	Notes
Maternal age (years)	<20							
	20-24							
	25-29							
	30-34							
	35-39							
	40-44							
	≥45							
	Unknown							
	Total							
Month of delivery	January							
	February							
	March							
	April							
	May							
	June							
	July							
	August							
	September							
	October							
	November							
	December							
	Unknown							
	Total							
Gestational age (weeks)	< 22							
	22-27							
	28-31							
	32-36							
	37-41							
	≥42							
	Unknown							
	Total							
Birth weight (grams)	<500							
	500-999							
	1,000-1,499							
	1,500-1,999							
	2,000-2,499							
	2,500-2,999							
	3,000-4,499							
	≥ 4,500							
	Unknown							
	Total							

Table 12: CCASS National Variables Record Layout for Submission

No	Variable	Description	Start Position	End Position	Length	Type	Coding Notes	Submission Notes
1	BATCHID	Unique ID# for each batch	1	10	10	character	PPYYYY#### (e.g. NL20130001, NL20130002)	
2	CASEID	Unique encrypted ID per case	11	20	10	character	YYYY###### (e.g. 2013000001, 2013000002)	
3	PROV-BIRTH	Province of birth outcome	21	22	2	character	10 = NL, 11 = PEI, 12=NS, 13=NB, 24=QC, 35=ON, 46=MB, 47=SK, 48=AB, 59=BC, 50=YT, 61=NT, 62=NU, 09=Unknown, 99=Outside Canada	
4	PROV-RES	Mother's province of residence at birth outcome	23	24	2	character	10 = NL, 11 = PEI, 12=NS, 13=NB, 24=QC, 35=ON, 46=MB, 47=SK, 48=AB, 59=BC, 50=YT, 61=NT, 62=NU, 09=Unknown, 99=Outside Canada	
5	BIRTH_DATE	Date of birth outcome	25	32	8	character	DDMMYYYY. If not able to send full DOB, set day to 99 (e.g. 99042013)	
6	PC_RES	Mothers residence postal code at birth outcome	33	38	6	character	L#L #L# - Sending full postal code as per revised Privacy Impact Assessment and amendment to Data Sharing Agreement	
7	SGC_RES	Mom's residence SGC code at birth outcome	39	45	7	character	SGC code derived based on town 9999999 = unknown	
8	MAT_DOB	Mother's date of birth	46	53	8	character	DDMMYYYY. If not able to send full DOB, set day to 99 (e.g. 99041990)	
9	OUTCOME	Pregnancy outcome	54	54	1	character	1 = Live Birth; 2 = Fetal death >/= 20 weeks gestation (still birth); 3 = Fetal death < 20 weeks gestations (miscarriage/aborted); 9 = unknown	

No	Variable	Description	Start Position	End Position	Length	Type	Coding Notes	Submission Notes
10	TOP	Termination of pregnancy at any age after prenatal diagnosis of congenital anomalies	55	55	1	character	1 = Yes; 2 = No; 9 = Unknown/missing	
11	NUMFET	# of babies/fetuses at birth/termination	56	57	2	character	1 = Singleton; 2 = Twins; 3 = Triplets; 4 = Quadruplets; 99 = Unknown/missing	
12	SEX	Sex of baby/fetus	58	58	1	character	1 = Male; 2 = Female; 3 = Indeterminate; 9 = Unknown/missing	
13	BIRTHWT	Weight of baby/fetus at birth outcome in grams	59	62	4	numeric	9999 = Unknown/missing	
14	GESTAGE	Completed weeks of gestation at birth outcome	63	64	2	numeric	99 = Unknown/missing	
15	DEATH_DATE	Date of death (for live births only)	65	72	8	character	DDMMYYYY. If not able to send full date, set day to 99 (e.g. 99042013) 22222222=confirmed alive at 1 yr. 33333333=unknown status at 1 yr.	
16	DXCODE01-DXCODE##	ICD 10-CA code	73	77	5	character	'Q' plus 4-digit code	
17*	DXPREFIX01-DXPREFIX##	Prefix code for ICD 10-CA code	78	78	1	character	''Q' indicates this is a query (suspected) case	

*Note: Given the discussions at the November 2018 External Advisory Committee Meeting it has been decided to include the diagnosis prefix in order to identify potential query codes

Table 13: CCASS Data Transfer Form

CCASS Data Transfer Form **Accompanying Information Form** In order to answer questions related to the data submission, please provide the following information with each file transfer.	
Province/Territory:	
Filename: Date: Cases from: to (MM-DD-YYYY) Denominator from: to (MM-DD-YYYY) Date of Extraction (Denominator) (MM-DD-YYYY)	
Questions	**Responses**
In what format did you submit the data (for example fixed format text file (ASCII, excel format, comma delimited)	
If the variable position file is provided, does it correspond with the case and denominator files for this submission? (Y/N)	
Did you use the same format as previous submissions? (Y/N) – if no, provide details (for example the record layout)	
Indicate the data source(s) used to acquire data/ascertain cases for the most recent submission by data item **Error! Bookmark not defined.**	
Are the data submitted as calendar or fiscal year – please provide details	
Does this submission include revised data from a previous submission? (Y/N) – if yes, provide details	
Are any of the seventeen variables missing (Y/N) – if yes, provide details	
Are any data suppressed (including denominator data) (Y/N) – if yes, provide details	
Are the data based on definitions from the enclosed CCASS Example of a Data File Information Sheet (Excel)? (Y/N) – if no, have you included a data dictionary with the submission	
Were the denominator and numerator data extracted during approximately the same time period? (Y/N) –if no list the time gap and explain why	
Have you coded the multiple congenital anomalies using the decision tree? [i]	
Specify the case follow-up period (should be a minimum of one year of age, and ideally up to 6 years of age)	
Have you submitted any data for which a variable code was not available (Y/N) – if yes, provide details	
Was the denominator data included in the most recent data submission (Y/N) – if no, provide details	
Are any denominator data suppressed (Y/N) – if yes, provide details	
Did you respond to the CCASS Survey and Fill in the Notes Section of the CCASS National Variable and Denominator Record Layouts?	

[1] Canadian Institute for Health Information. Canadian Coding Standards for Version 2018 ICD-10-CA and CCI. Ottawa, ON: CIHI; 2015.Chapter XVII Congenital Malformations, Deformations, and Chromosomal Anomalies, page 424.

- **Submission Follow-up**:
    - Following submission, CCASS will review the data and information.
    - A CCASS Data Quality Report will be issued to the jurisdiction through PHAC's secure file transfer protocol (FTP) with
        - data submission details,
        - summary of outstanding items and questions,
        - data frequencies, and
        - denominator data.
    - Upon receipt of the Data Quality Report, issues identified should be reviewed and addressed with CCASS as appropriate.

References

1. Coggon D, Rose G, Barker DGP (2003). Epidemiology for the uninitiated, 5th edition. London: BMJ Books. ISBN 0 7279 1604 1.
2. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, Appendix 11.3 Data Users Matrix. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised March 2015.
3. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, Chapter 8 Statistical Methods, Subsection 8.6. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised March 2015.
4. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, Chapter 11 Data Presentation. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised March 2015.
5. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance, Appendix 11.1 Data Suppression. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc., June 2004, revised March 2015.

Appendix 1: Case Definitions for National Reporting

CONGENITAL ANOMALY DEFINITIONS AND ICD-10-CA CODES FOR THE PUBLIC HEALTH AGENCY OF CANADA CONGENITAL ANOMALIES SURVEILLANCE REPORT

Images and definitions are from the following sources unless otherwise stated:

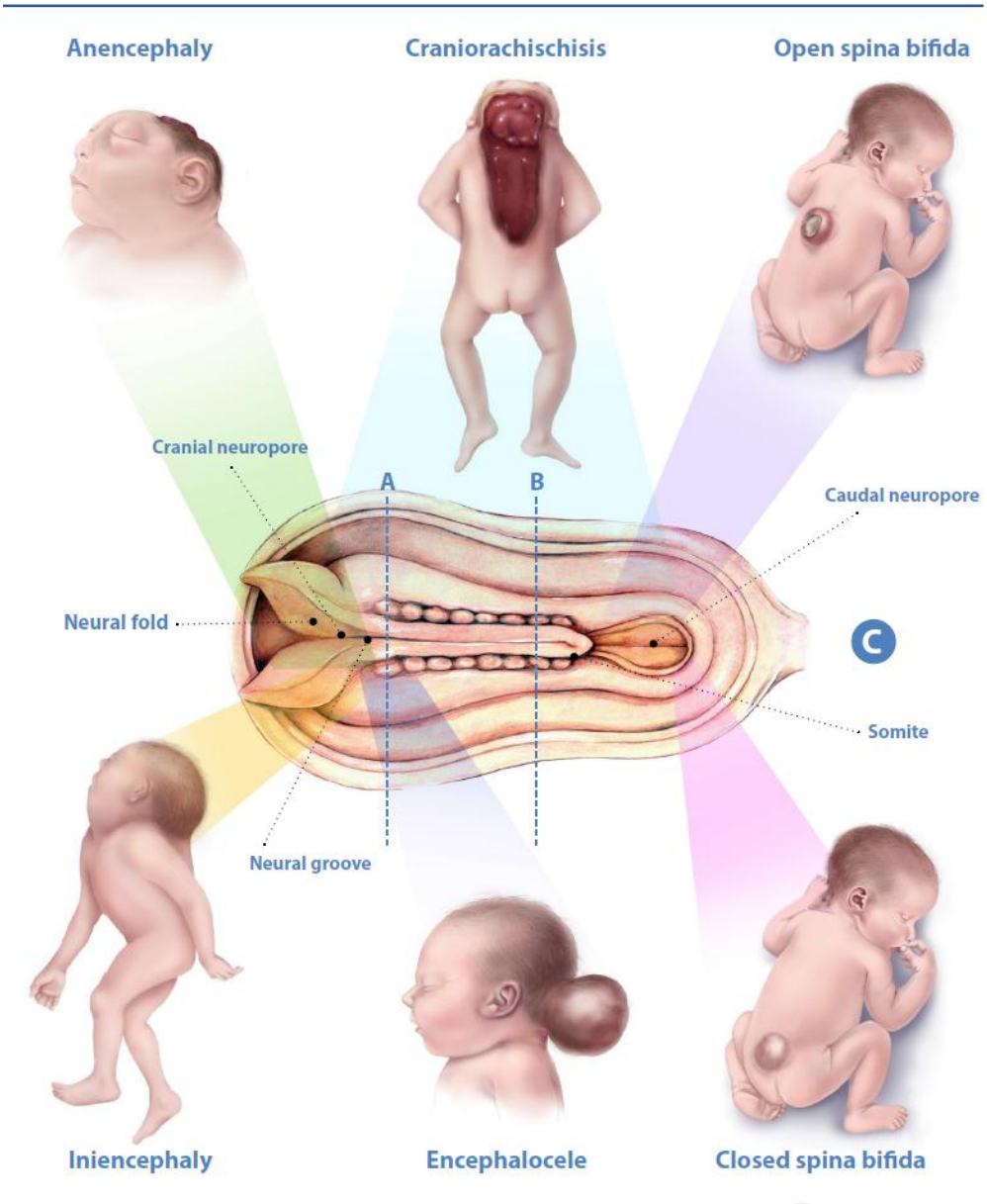
Birth defects surveillance: a manual for programme managers

Appendix 3.1 Birth Defects Descriptions for NBDPN Core, Recommended, and Extended Conditions

Centers for Disease Control and Prevention, Specific Birth Defects

1. Neural Tube Defects (NTDs)

These anomalies affect the brain and spinal cord. Very early in the development of an embryo, certain cells form a tube (called the neural tube) that will later become the spinal cord, the brain, and the nearby structures that protect them, including the backbone (also called the spinal column or vertebra). As development progresses, the top of the tube becomes the brain and the remainder becomes the spinal cord. A neural tube defect occurs when this tube does not close completely somewhere along its length, resulting in a hole in the spinal column or another type of defect. These defects occur in the first month of pregnancy, often before a woman even knows that she is pregnant. See additional information about neural tube defects on the NIH Eunice Kennedy Shriver National Institute of Child Health and Human Development.



A. Anenecephaly and similar anomalies (Q00*): a congenital malformation characterized by the total or partial absence of the cranial vault, the covering skin, and the brain missing or reduced to a small mass (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** Anencephaly can be accepted as a definitive anomaly and coded appropriately if the diagnosis has been made in a special prenatal diagnostic centre, e.g. Maternal Fetal Medical Unit (Surveillance and Standards Working Group, May 2007 and ACASS). However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data (NBDPN, 2017).

- ***IDC-10-CA codes:***
  - ***Q00.0*** Anencephaly. Includes:
    - Acephaly
    - Acrania
    - Amyelencephaly
    - Hemianencephaly
    - Hemicephaly
  - ***Q00.1*** Craniorachischisis
  - ***Q00.2*** Iniencephaly

- ***Illustrations:***

Q00.0 (Anencepahly)

Q00.1 (Craniorachischisis)

Q00.2 (Iniencephaly)

B. Spina bifida without anencephaly (Q05*) without (Q00.0): a family of congenital malformation defects in the closure of the spinal column characterized by herniation or exposure of the spinal cord and/or meninges through an incompletely closed spine (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** Spina bifida can be accepted as a definitive anomaly and coded appropriately if the diagnosis has been made in a special prenatal diagnostic centre, e.g. Maternal Fetal Medical Unit (Surveillance and Standards Working Group, May 2007). However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of spina bifida on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***Includes:***
  - Hydromeningocele (spinal)
  - Meningocele (spinal)
  - Meningomyelocele
  - Myelocele
  - Myelomeningocele
  - Rachischisis
  - Spina bifida (aperta)(cystica)
  - Syringomyelocele

- ***Excludes:*** Arnold Chiari Malformation (Q07.0) and spina bifida occulta (Q76.0).

- ***ICD-10-CA coding:*** Do not code spina bifida when associated with anencephaly – code anencephaly only. Codes:
  - ***Q05.0*** Cervical spina bifida with hydrocephalus
  - ***Q05.1*** Thoracic spina bifida with hydrocephalus. Includes:
    - Spina bifida dorsa with hydrocephalus
    - Thoracolumbar with hydrocephalus
  - ***Q05.2*** Lumbar spina bifida with hydrocephalus. Includes:
    - Lumbosacral spina bifida with hydrocephalus
  - ***Q05.3*** Sacral spina bifida with hydrocephalus
  - ***Q05.4*** Unspecified spina bifida with hydrocephalus
  - ***Q05.5*** Cervical spina bifida without hydrocephalus
  - ***Q05.6*** Thoracic spina bifida without hydrocephalus. Includes:
    - Spina bifida, dorsal NOS
    - Thoracolumbar NOS)
  - ***Q05.7*** Lumbar spina bifida without hydrocephalus. Includes:
    - Lumbosacral spina bifida NOS)
  - ***Q05.8*** Sacral spina bifida without hydrocephalus

- ***Q05.9*** Spina bifida, unspecified

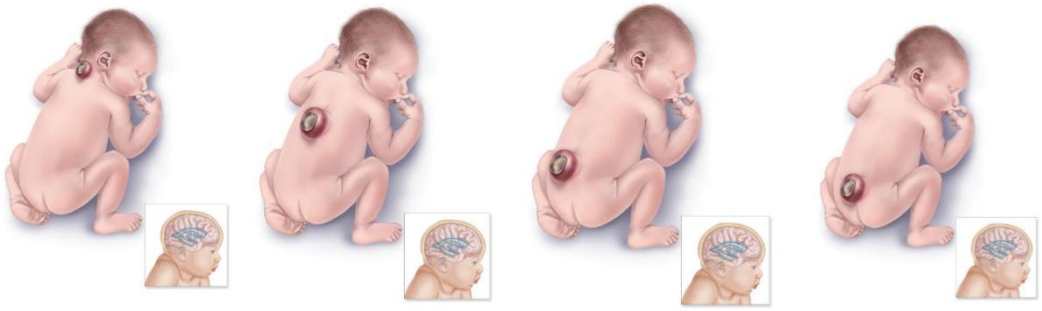
- ***Illustrations:***

<u>Spina bifida with hydrocephalus:</u>

Q05.0 (Cervical) **Q05.1 (Thoracic)** **Q05.2 (Lumbar)** **Q05.3 (Sacral)**

<u>Spina Bifida without hydrocephalus:</u>

Q05.5 (Cervical) **Q05.6 (Thoracic)** **Q05.7 (Lumbar)** **Q05.8 (Sacral)**



C. Encephalocele (Q01): a congenital malformation characterized by the herniation of the brain and/or meninges through a defect in the skull (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** Encephalocele may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. In addition, the absence of a small encephalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***Includes:***
  - Encephalomyelocele
  - Hydroencephalocele
  - Hydromeningocele, cranial
  - Meningocele, cerebral
  - Meningoencephalocele

- ***ICD-10-CA coding:***
  - ***Q01.0*** Frontal encephalocele
  - ***Q01.1*** Nasofrontal encephalocele
  - ***Q01.2*** Occipital encephalocele
  - ***Q01.8*** Encephalocele of other sites
  - ***Q01.9*** Encephalocele, unspecified

- ***Illustrations:***

Q01.0 (Frontal) **Q01.1 (Nasofrontal)** **Q01.2 (Occipital)**

2. Selected Central Nervous System Defects

A. Microcephaly (Q02): a congenitally small cranium, defined by an occipito-frontal circumference (OFC) 3 standard deviations below the age and sex appropriate distribution curves (ICBDSR definition). (Note: Ideally, head circumference measurements should be recorded and used, but may not be available in all jurisdictions).

- ***Includes:***
  - Hydromicrocephaly
  - Micrencephalon

- ***Illustrations:***

Centers for Disease Control: Congenital Zika Syndrome & Other Birth Defects / Microcephaly, Measuring Head Circumference

B. Hydrocephaly (Q03): a congenital malformation characterized by dilatation of the cerebral ventricles, not associated with primary brain atrophy, with or without enlargement of the head, diagnosed at birth (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** Definitive hydrocephaly can be accepted as a definitive anomaly and coded appropriately if the diagnosis has been made in a special prenatal diagnostic centre, e.g. Maternal Fetal Medical Unit (Surveillance and Standards Working Group, May 2007).

- ***ICD-10-CA coding:***
  - ***Q03.0*** Malformations of aqueduct of Sylvius. Includes:
    - Anomaly
    - Obstruction, congenital
    - Stenosis

- ***Q03.1*** Atresia of foramina of Magendie and Luschka. Includes:
  - Dandy-Walker syndrome
- ***Q03.8*** Other congenital hydrocephalus
- ***Q03.9*** Congenital hydrocephalus, unspecified

C. Arhinencephaly/Holoprosencephaly (Q04.1, Q04.2): a congenital malformation of the brain, characterized by various degrees of incomplete lobation of the brain hemispheres. Olfactory nerve tract may be absent (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** Holoprosencephaly may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. For example, clear diagnoses of cyclopia, ethmocephaly, or cebocephaly are virtually always associated with holoprosencephaly, but prenatal diagnoses of lobar holoprosencephaly and middle interhemispheric variants are more problematic without postnatal imaging or autopsy confirmation (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q04.1*** Arhinencephaly
  - ***Q04.2*** Holoprosencephaly

3. Selected Sense Organ Defects

A. Anophthalmos/microphthalmos (Q11.0, Q11.1, Q11.2): apparently absent or small eyes. Some normal adnexal elements and eyelids are usually present. In microphthalmia, the corneal diameter is usually less than 10 mm and the atero-posterior diameter of the globe is less than 20mm (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anophthalmia or microphthalmia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q11.0*** Cystic eyeball
  - ***Q11.1*** Other Anophthalmos. Includes:
    - Agenesis of eye
    - Aplasia of eye
  - ***Q11.2*** Microphthalmos. Includes:
    - Cryptophthalmos NOS
    - Dysplasia of eye
    - Hypoplasia of eye
    - Rudimentary eye

- ***Illustrations:***

Anophthalmia **Micropthalmia**

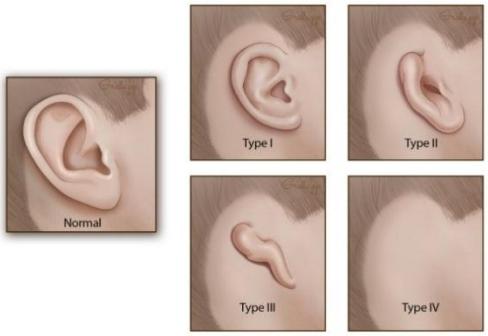
Centres for Disease Control and Prevention: Facts about Anophthalmia/Microphthalmia

B. Anotia/microtia (Q16.0, Q17.2): a congenital malformation characterized by absent parts of the pinna (with or without atresia of the ear canal) commonly expressed in grades (I-IV) of which the extreme form (grade IV) is anotia, absence of pinna. Exclude small, normally shaped ears (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally:*** While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of anotia or microtia on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q16.0*** Congenital absence of (ear) auricle
  - ***Q17.2*** Microtia

- ***Illustrations:***

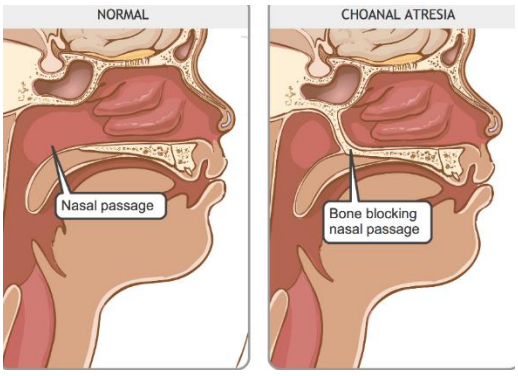


Stanford Children's Health: Microtia – The four grades of microtia

C. Choanal atresia (Q30.0): congenital obstruction (membranous or osseous) of the posterior choana or choanae (ICBDSR definition).

- ***ICD-10-CA coding:***
  - ***Q30.0*** Choanal atresia. Includes:
    - Atresia of nares (posterior) (anterior)
    - Congenital stenosis on nares (posterior) (anterior)

- ***Illustrations:***



About Kids Health: Choanal Atresia

4. Selected Congenital Heart Defects

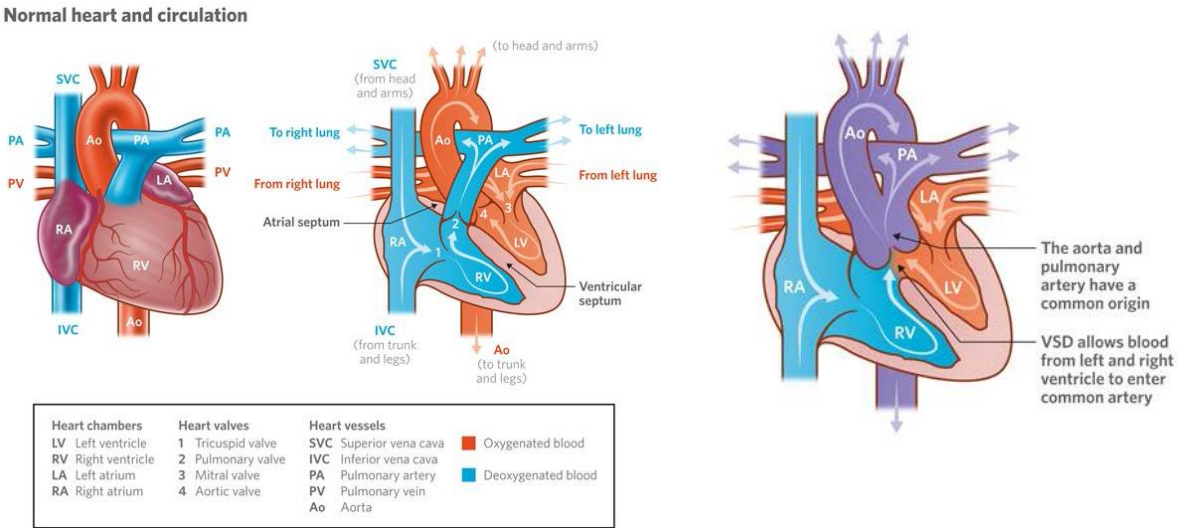
A. Common Truncus (Q20.0, Q21.4): failure of separation of the aorta and the pulmonary artery, resulting in a single common arterial trunk carrying blood from the heart to both the body and the lungs (Conotruncal) (NBDPN description).

- ***Prenatal diagnoses not confirmed postnatally*:** These conditions may be included as cases when only diagnosed prenatally by a pediatric cardiologist through fetal echocardiography. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q20.0*** Common arterial trunk. Includes:
    - Persistent truncus arteriosus
  - ***Q21.4*** Aortopulmonary septal defect. Includes:
    - Aortic septal defect
    - Aortopulmonary window

- ***Illustrations:***

Normal Heart **Q20.0 (Common truncus)**



[The Royal Children's Hospital Melbourne: Heart defects]

B. The group of anomalies: 'Transposition of great vessels' (Q20.1, Q20.3, Q20.2, Q20.5). These anomalies are grouped for reporting purposes, since these defects are considered to be on the same spectrum of congenital heart anomalies.

- ***Prenatal diagnoses not confirmed postnatally*:** These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - **Double Outlet Right Ventricle (Q20.1)**: a cardiac defect where both the aorta and the pulmonary artery originate from the right ventricle and blood from the left ventricle passes across a VSD into the RV to reach the great arteries. The lung circulation is often exposed to very high pressure and increased blood flow (as with a large VSD). There are many different varieties of this abnormality.
    - ***Q20.1*** Double outlet right ventricle. Includes:
      - Incomplete (partial) transposition of great vessels
      - Taussig-Bing syndrome

  - **Double Outlet Left Ventricle (Q20.2)**: a cardiac defect where both the aorta and the pulmonary artery originate from the left ventricle. This is a very rare condition.
    - ***Q20.2*** Double outlet left ventricle

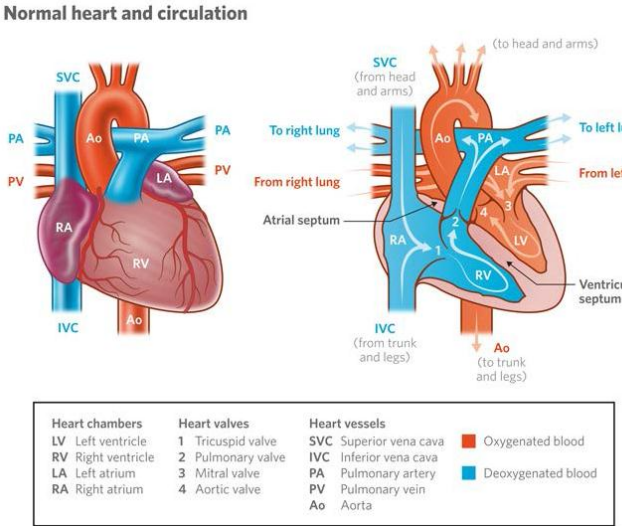
  - **Transposition of great vessels (Q20.3)**: a cardiac defect where the aorta exits from the right ventricle and the pulmonary artery from the left ventricle, with or without other cardiac defects (ICBDSR definition).
    - ***Q20.30*** Dextratransposition of aorta
    - ***Q20.31*** Complete transposition of great vessels
    - ***Q20.32*** Congenitally corrected transposition of great vessels
    - ***Q20.38*** Other transposition of great vessels NEC

  - **Discordant atrioventricular connection (Q20.5):** where the ventricle on the right side of the heart has the anatomic appearance of the left ventricle, and the ventricle on the left side of the heart has the anatomic appearance of the right ventricle (ventricular inversion). The pulmonary artery arises from the anatomic left ventricle and the aorta arises from the anatomic right ventricle (hence the designation of transposition). Because blood from the ventricle on the right flows through the pulmonary artery, and that from the ventricle on the left flows through the aorta, circulation is normal as long as there are no other defects (NBDPN description).

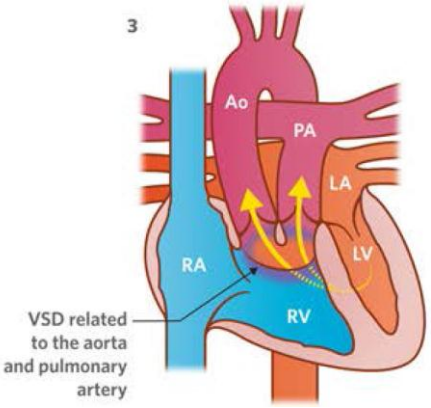
- ***Q20.50*** Discordant atrioventricular connection with corrected transposition. Includes:
  - Corrected transposition of atrioventricular connection
- ***Q20.58*** Discordant atrioventricular connection NEC. Includes:
  - Laevotransposition
  - Ventricular inversion

- ***Illustrations:***

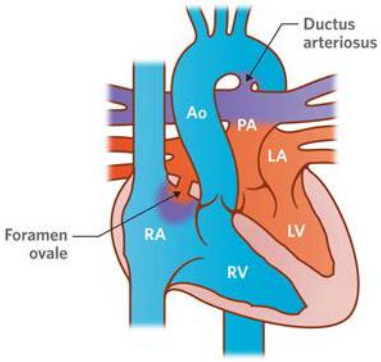
Normal heart



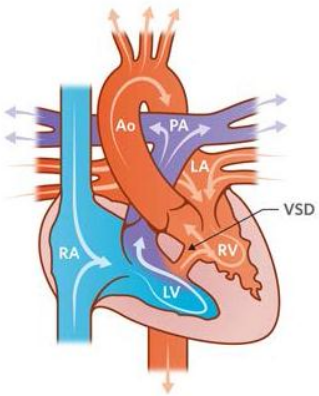
Q20.1 (Double outlet right ventricle)



Q20.3 (Transposition great vessels)



Q20.5 (Discordant atrioventricular connection)



[The Royal Children's Hospital Melbourne: Heart defects]

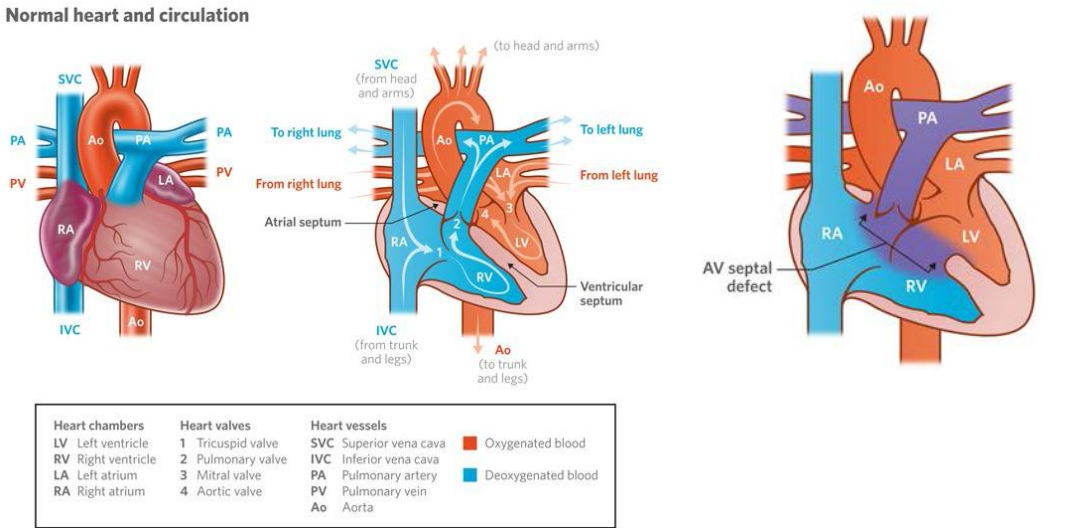
C. Endocardial cushion defects/atrioventricular septal defect (Q21.2): a defect in the lower portion of the atrial septum and the upper portion of the ventricular septum, producing a large opening (canal) in the central part of the heart. The adjacent parts of the mitral and tricuspid valves may be abnormal also, resulting in a single common atrioventricular valve. In extreme cases, virtually the entire atrial and ventricular septae may be missing (NBDPN description).

- ***Prenatal diagnoses not confirmed postnatally*:** These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish this condition from other abnormalities of the cardiac septae prenatally. Live-born children who survive should always have confirmation of the defect postnatally (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q21.2*** Atrioventricular septal defect. Includes:
    - Common atrioventricular canal
    - Endocardial cushion defect
    - Ostium primum atrial septal defect (type I)

- ***Illustrations:***

Normal Heart **Q21.2 (Atrioventricular septal defect)**



The Royal Children's Hospital Melbourne: Heart defects

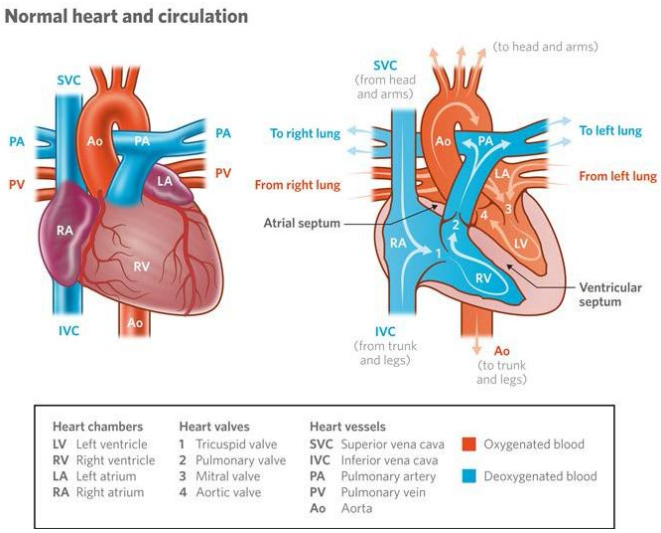
D. Tetralogy of Fallot (Q21.3): The simultaneous presence of a ventricular septal defect (VSD), pulmonic stenosis (valve/right ventricular outflow), a malpositioned aorta that overrides the ventricular septum, and right ventricular hypertrophy (NBDPN description)

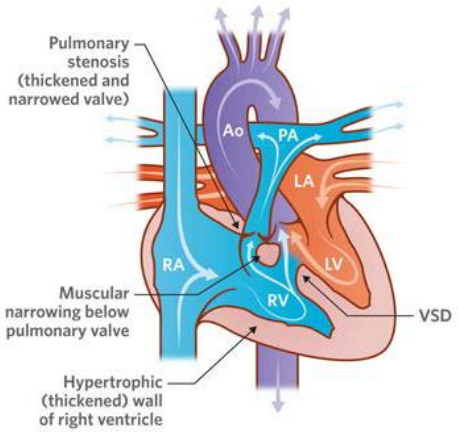
- ***Prenatal diagnoses not confirmed postnatally*:** These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q21.3*** Tetralogy of Fallot. Includes:
    - Ventricular septal defect with pulmonary stenosis or atresia
    - Dextroposition of aorta and hypertrophy of right ventricle

- ***Illustrations:***

Normal Heart **Q21.3 (Tetralogy of Fallot)**





The Royal Children's Hospital Melbourne: Heart defects

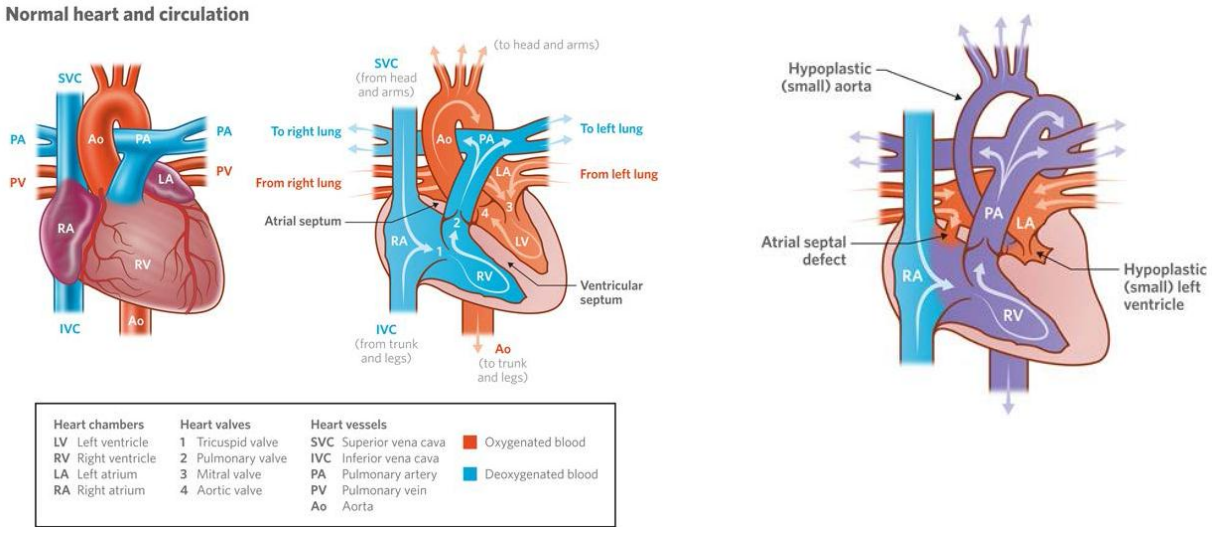
E. Hypoplastic left heart syndrome (Q23.4): a condition in which the structures on the left side of the heart and the aorta are extremely small. Classically, this condition includes hypoplasia of the left ventricle, atresia or severe hypoplasia of the mitral and arotic valves, and hypoplasia and coarctation of the aorta (NBDPN description).

- ***Prenatal diagnoses not confirmed postnatally*:** These conditions may be included as cases when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish this condition from other abnormalities of the left ventricle prenatally. Live-born children who survive should always have confirmation of the defect postnatally before being included (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q23.4*** Hypoplastic left heart syndrome. Includes:
    - Atresia, or marked hypoplasia of aortic orifice or valve, with hypoplasia of ascending aorta and defective development of left ventricle (with mitral valve stenosis or atresia).

- ***Illustrations:***

Normal Heart **Q23.4 (Hypoplastic left heart syndrome)**



[The Royal Children's Hospital Melbourne: Heart defects]

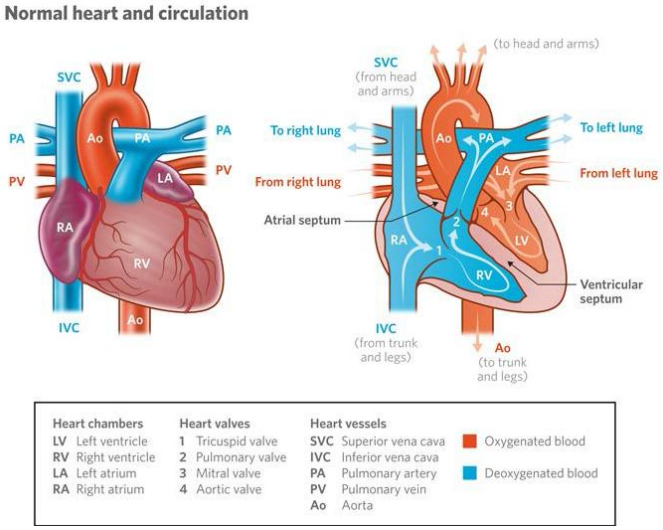
F. Coarctation of aorta (Q25.1): narrowing of the descending aorta, which may obstruct blood flow from the heart to the rest of the body. The most common site of coarctation occurs distal to the origin of the left subclavian artery in the region of the ductus arteriosus (NBDPN description).

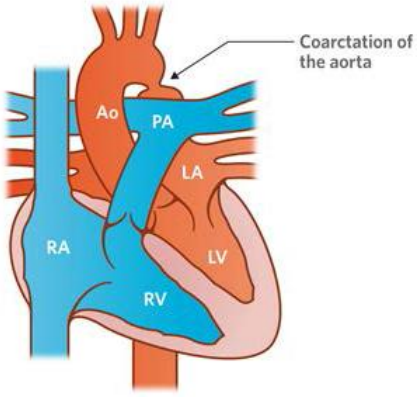
- ***Prenatal diagnoses not confirmed postnatally*:** While this condition may be identified by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of coarctation of the aorta on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q25.1*** Coarctation of aorta. Includes coarctation of aorta:
    - Preductal
    - Postductal

- ***Illustrations:***

Normal Heart **Q25.1 (Coarctation of aorta)**





[The Royal Children's Hospital Melbourne: Heart defects]

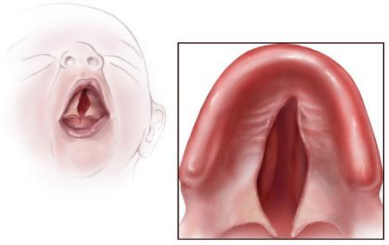
5. Oro-Facial Clefts

A. Cleft palate only (Q35 excluding Q35.7, cleft uvula): a congenital malformation characterized by a closure defect of the hard and/or soft palate behind the foramen incisivum without cleft lip. Include submucous cleft palate (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** This condition should not be included in birth defects surveillance data without postnatal confirmation (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q35.1*** Cleft hard palate
  - ***Q35.3*** Cleft soft palate
  - ***Q35.5*** Cleft hard palate with cleft soft palate
  - ***Q35.9*** Cleft palate, unspecified. Includes:
    - Cleft palate NOS
    - Submucous cleft palate

- ***Illustrations:***



B. Cleft lip only (Q36): a congenital malformation characterized by partial or complet clefting of the upper lip, with or without clefting of the alveolar ridge (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** While this condition may be identified by prenatal ultrasound, it should not be included in birth defects surveillance data without postnatal confirmation. In addition, the absence of cleft lip on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q36*** Cleft lip. Includes:
    - Cheiloschisis
    - Congenital fissure of lip
    - Harelip
    - Labium leporinum

- ***Illustrations:***

Bilateral cleft lip

Unilateral cleft lip

C. Cleft lip with cleft palate (Q37): a congenital malformation characterized by partial or complete clefting of the upper lip, with clefting of the palate (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** While this condition may be identified by prenatal ultrasound, it should not be included in birth defects surveillance data without postnatal confirmation. In addition, the absence of cleft lip on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q37*** Cleft palate with cleft lip

- ***Illustrations:***

Bilateral cleft lip and palate

Unilateral cleft lip and palate

6. Selected Gastrointestinal Anomalies

A. Oesophageal atresia/stenosis, tracheoesophageal fistula (Q39.0, Q39.1, Q39.2, Q39.3, Q39.4): a congenital malformation characterized by absence of continuity or narrowing of the oesophagus, with or without tracheal fistula (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included (NBDPN, 2017).

- ***ICD-10-CACodes:***
  - ***Q39.0*** Atresia of oesophagus without fistula. Includes:
    - Atresia of oesophagus NOS
  - ***Q39.1*** Atresia of oesophagus with tracheo-oesophageal fistula. Includes:
    - Atresia of oesophagus with broncho-oesophageal fistula
  - ***Q39.2*** Congenital tracheo-oesophageal fistula without atresia. Includes:
    - Congenital tracheo-oesophageal fistula NOS
    - ***Excludes***: that with atresia of oesophagus (Q39.1)
  - ***Q39.3*** Congenital stenosis and stricture of oesophagus
  - ***Q39.4*** Oesophageal web

- ***Illustrations:***

Normal oesophagus

Abnormal oesophagus

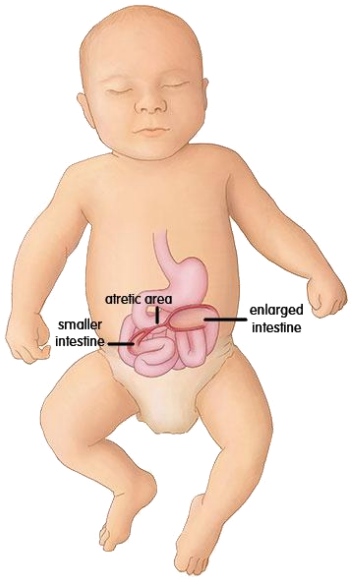
The upper part of the esophagus has a closed end (atresia) and the lower part of the esophagus is attached to the trachea (fistula).

B. Small intestine absence/atresia/stenosis (Q41): complete or partial occlusion of the lumen of a segment of the small intestine. It can involve a single area or multiple areas (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** While these conditions may be suspected by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation; postnatal diagnosis of the small intestinal atresia or stenosis requires a surgical or autopsy report (i.e., ultrasound or abdominal x-ray studies, such as an upper GI or barium enema, are not sufficient). In addition, the absence of small intestinal atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q41.0*** Congenital absence, atresia and stenosis of duodenum
  - ***Q41.1*** Congenital absence, atresia and stenosis of jejunum. Includes:
    - Apple peel syndrome, imperforate jejunum
  - ***Q41.2*** Congenital absence, atresia and stenosis of ileum
  - ***Q41.8*** Congenital absence, atresia and stenosis of other specified parts of small intestine
  - ***Q41.9*** Congenital absence, atresia and stenosis of small intestine, part unspecified. Includes:
    - Congenital absence, atresia and stenosis of intestine NOS

- ***Illustrations:***

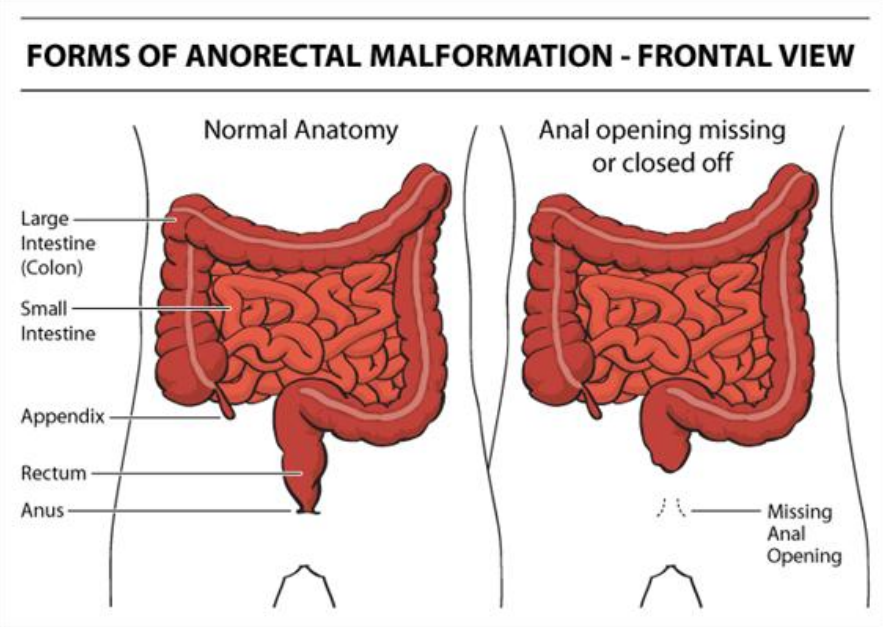


C. Ano-rectal absence/atresia/stenosis (Q42.0, Q42.1, Q42.2, Q42.3): characterized by absence of continuity of the anorectal canal or of communication between rectum and anus, or narrowing of anal canal, with or without fistula to neighbouring organs (ICBDSR definition

- ***Prenatal diagnoses not confirmed postnatally*:** While these conditions may be identified by prenatal ultrasound, they should not be included in surveillance data without postnatal confirmation. In addition, the absence of intestinal, rectal or anal atresia or stenosis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CAcodes:***
  - ***Q42.0*** Congenital absence, atresia and stenosis of rectum with fistula
  - ***Q42.1*** Congenital absence, atresia and stenosis of rectum without fistula. Includes:
    - Imperforate rectum
  - ***Q42.2*** Congenital absence, atresia and stenosis of anus with fistula
  - ***Q42.3*** Congenital absence, atresia and stenosis of anus without fistula. Includes:
    - Imperforate anus

- ***Illustrations:***



University of California San Francisco Pediatric Surgery: Anorectal Malformation

D. **Hirschsprung's disease (Q43.1)**: characterized by the absence of particular nerve cells (ganglions) in a segment of the bowel in an infant which causes the muscles in the bowels to lose their ability to move stool through the intestine (peristalsis) (National Organization for Rare Disorders: Hirschsprung Disease).

- ***ICD-10-CA coding:***
  - ***Q43.1*** Hirschsprung's disease. Includes:
    - Aganglionosis
    - Congenital (aganglionic) megacolon

E. **Atresia of bile ducts (Q44.2)**: characterized by destruction or absence of all or a portion of the bile duct resulting in the abnormal accumulation of bile in the liver (National Organization for Rare Disorders: Biliary Atresia).

- ***Prenatal diagnoses not confirmed postnatally*:** While biliary atresia may be suspected by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of biliary atresia on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q44.2*** Atresia of bile ducts

7. Selected Urinary Tract Anomalies

A. Renal agenesis (Q60.0, Q60.1, Q60.2): characterized by complete absence of kidneys (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally*:** Bilateral renal agenesis may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q60.0*** Renal agenesis, unilateral
  - ***Q60.1*** Renal agenesis, bilateral
  - ***Q60.2*** Renal agenesis, unspecified

B. Cystic Kidney (Q61.1, Q61.2, Q61.3, Q61.4, Q61.5, Q61.8, Q61.9): characterized by multiple cysts in the kidney (ICBDSR definition).

- ***ICD-10-CA coding:***
  - ***Q61.1*** Polycystic kidney, autosomal recessive. Includes:
    - Polycystic kidney, infantile type
  - ***Q61.2*** Polycystic kidney, autosomal dominant. Includes:
    - Polycystic kidney, adult type
  - ***Q61.3*** Polycystic kidney, unspecified
  - ***Q61.4*** Renal dysplasia. Includes:
    - Multicystic dyplastic kidney
    - Multicystic kidney (developmental)
    - Multicystic kidney disease
    - Multicystic renal dysplasia
    - ***Excludes:*** Polycystic kidney disease (Q61.1-Q61.3)
  - ***Q61.5*** Medullary cystic kidney. Includes:
    - Sponge kidney NOS
  - ***Q61.8*** Other cystic kidney diseases. Includes:
    - Fibrocystic: kidney, renal degeneration or disease
  - ***Q61.9*** Cystic kidney disease, unspecified. Includes:
    - Meckel-Gruber syndrome

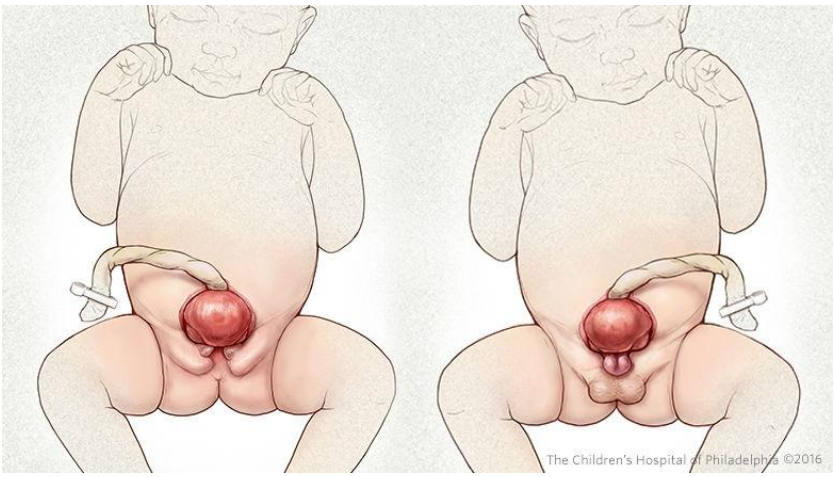
C. Bladder and cloacal exstrophy (Q64.1): a complex malformation characterized by a defect in the closure of the lower abdominal wall and bladder. As the bladder is developing the

abdominal wall does not fully form, leaving the pubic bones separated and the bladder exposed to the outside through an opening in the lower abdominal wall. Urine produced by the kidneys drains into this open area and is not stored normally in the bladder (Children's Hospital of Philadelphia: Bladder Exstrophy).

- ***Prenatal diagnoses not confirmed postnatally*:** These conditions may be included when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data, as it may be difficult to distinguish bladder exstrophy from cloacal exstrophy. Live-born children who survive should always have confirmation of the defect postnatally before being included (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q64.1*** Exstrophy of urinary bladder
  - ***Q64.10*** Cloacal exstrophy of urinary bladder
  - ***Q64.18*** Other exstrophy of urinary bladder. Includes:
    - Ectopica vesicae
    - Exstrophy of bladder NOS
    - Extroversion of bladder

- ***Illustrations:***
  This illustration shows both a female (left) and male (right) baby with an exposed bladder caused by bladder exstrophy.



Children's Hospital of Philadelphia: Bladder Exstrophy

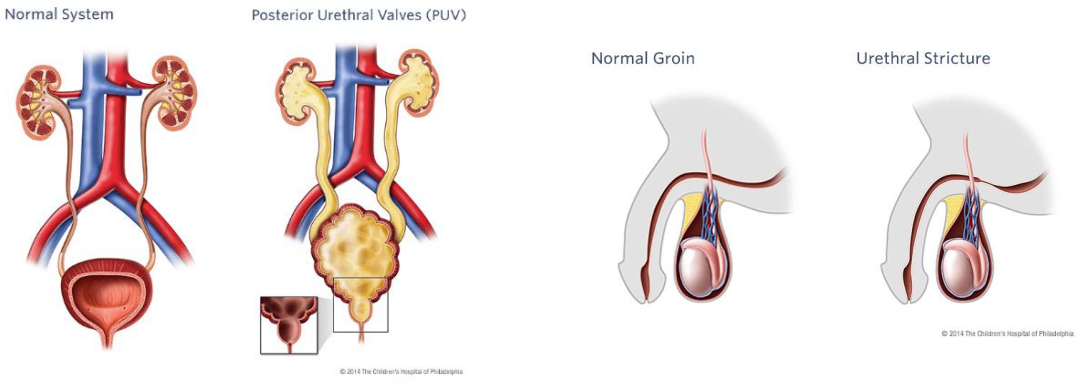
D. Lower urinary tract obstruction (Q64.2, Q64.3): Posterior urethral valves (PUV) are obstructive membranes that develop in the urethra (tube that drains urine from the bladder), close to the

bladder. The valve can obstruct or block the outflow of urine through the urethra. When this occurs, the bladder, ureters and kidneys become progressively dilated, which can lead to damage.

- ***Prenatal diagnoses not confirmed postnatally:*** While obstructive genitourinary defects including congenital PUV may be identified by prenatal ultrasound, many lesions diminish or resolve spontaneously prior to birth. For this reason, PUV should not be included in surveillance data without postnatal confirmation. In addition, the absence of genitourinary obstruction on prenatal ultrasound does not necessarily mean that an obstructive defect such as PUV will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q64.2*** Congenital posterior urethral valves
  - ***Q64.3*** Other atresia and stenosis of urethra and bladder neck
  - ***Q64.30*** Congenital bladder neck obstruction. Includes:
    - Urethrovesical obstruction (stricture)
  - ***Q64.31*** Congenital stricture of urethra
  - ***Q64.32*** Congenital stricture of urinary meatus
  - ***Q64.38*** Other congenital atresia and stenosis of urethra and bladder neck. Includes:
    - Iimpervious urethra

- ***Illustrations:***
  In boys, the urethra starts at the lower portion of the bladder and continues through the penis. A urethral stricture is a narrowing in the urethra. This narrowing makes it difficult for urine to drain out.



Children's Hospital of Philadelphia: Posterior Urethral Valves (PUV)

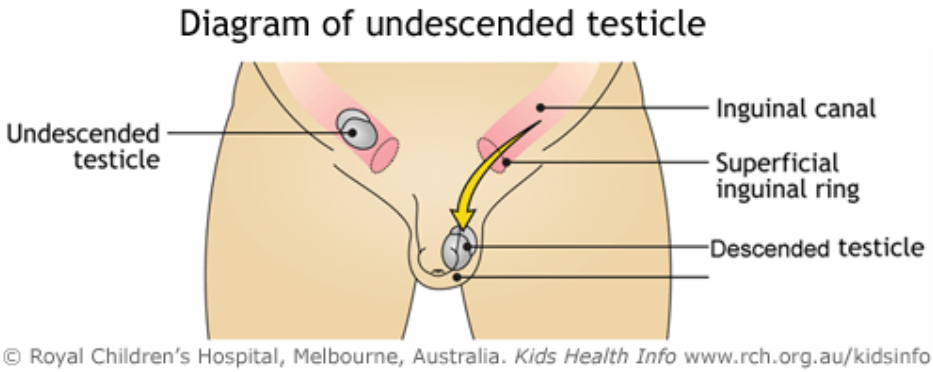
Children's Hospital of Philadelphia: Urethral Stricture

8. Selected Genital Anomalies

A. Cryptorchidism/undescended testes (Q53.1, Q53.2, Q53.9): When the testes (one or both) do not move down into the scrotum it is called 'undescended testes'. It is also known as Cryptorchidism. The Royal Children's Hospital Melbourne: Undescended Testes

- ***ICD-10-CA coding:***
  - ***Q53.1*** Undescended testicle, unilateral
  - ***Q53.2*** Undescended testicle, bilateral
  - ***Q53.9*** Undescended testicle, unspecified. Includes:
    - Cryptorchism NOS

- ***Illustrations:***

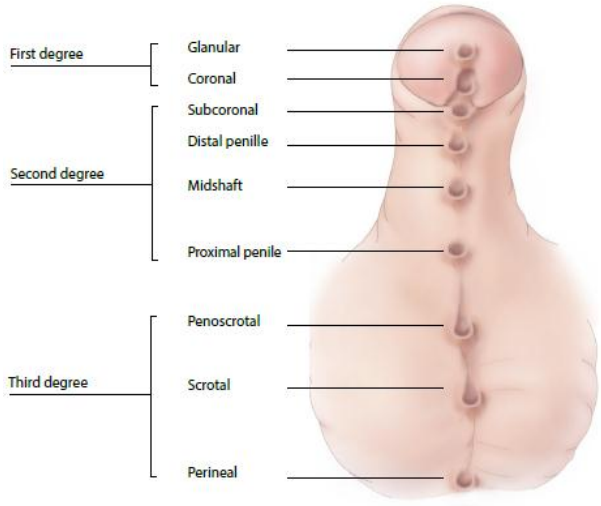


B. Hypospadias (Q54, excluding Q54.4): displacement of the urethral meatus ventrally and proximally from the tip of the penis. It is classified according to the position of the meatus on the penis (Birth defects surveillance: atlas of selected congenital anomalies).

- ***Prenatal diagnoses not confirmed postnatally*:** While this condition may be diagnosed by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of hypospadias on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q54.0*** Hypospadias, balanic. Includes:
    - Hypospadius: Coronal, glandular
  - ***Q54.1*** Hypospadias, penile
  - ***Q54.2*** Hypospadias, penoscrotal
  - ***Q54.3*** Hypospadias, perineal
  - ***Q54.8*** Other hypospadias
  - ***Q54.9*** Hypospadias, unspecified

- ***Illustrations:***



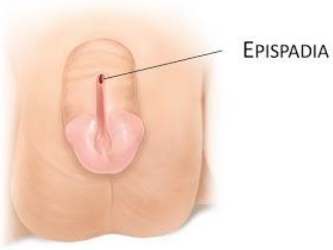
Note: illustration indicates all possible locations for the malformation, but one case will not have all.

C. Epispadias (Q64.0): a congenital malformation characterized by the opening of the urethra on the dorsal surface of the penis (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally***: While this condition may be diagnosed by prenatal ultrasound, it should not be included in surveillance data without postnatal confirmation. In addition, the absence of hypospadias on prenatal ultrasound does not necessarily mean that they will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q64.0*** Epispadias. ***Excludes:*** hypospadias (Q54.-)

- ***Illustrations:***



D. Indeterminate sex (Q56): genital ambiguity at birth that does not readily allow for phenotypic sex determination (ICBDSR definition).

- ***Excludes**:*
    - Female, with adrenocortical disorder (E25.-)
    - Male, with androgen resistance (E34.5)
    - Specified chromosomal anomaly (Q96-Q99)

- ***ICD-10-CA coding:***
    - ***Q56.0*** Hermaphroditism, not elsewhere classified. Includes:
        - Ovotestis
    - ***Q56.1*** Male pseudohermaphroditism, not elsewhere classified. Includes:
        - Male pseudohermaphroditism NOS
    - ***Q56.2*** Female pseudohermaphroditism, not elsewhere classified. Includes:
        - Female pseudohermaphroditism NOS
    - ***Q56.3*** Pseudohermaphroditism, unspecified
    - ***Q56.4*** Indeterminate sex, unspecified. Includes:
        - Ambiguous genitalia

9. Limb Deficiency Defects

Limb deficiency defects (Q71 to Q73): Congenital malformations characterized by total or partial absence or severe hypoplasia of skeletal structures of the limbs (ICBDSR definition). (See Birth defects surveillance: atlas of selected congenital anomalies).

- ***Prenatal diagnoses not confirmed postnatally***: While these conditions may be identified by prenatal ultrasound, they generally should not be included in surveillance data without postnatal confirmation. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Lack of visualization of a bone or limb on prenatal ultrasound does not necessarily mean that the bone or limb truly is not present. Live-born children who survive should always have confirmation of the defect postnatally before being included (NBDPN, 2017).

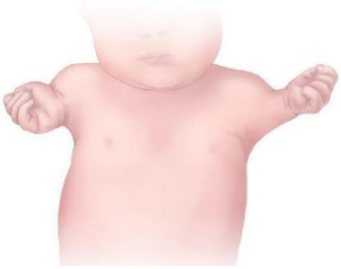
- ***ICD-10-CA coding:***
  - ***Q71.0*** Congenital complete absence of upper limb(s)
  - ***Q71.1*** Congenital absence of upper arm and forearm with hand present
  - ***Q71.2*** Congenital absence of both forearm and hand
  - ***Q71.3*** Congenital absence of hand and finger(s)
  - ***Q71.4*** Longitudinal reduction defect of radius. Includes:
    - Clubhand (congenital)
    - Radial clubhand
  - ***Q71.5*** Longitudinal reduction defect of ulna
  - ***Q71.6*** Lobster-claw hand
    - Complete or partial absence of central fingers and metacarpals.
    - Also known as split hand and congenital cleft hand.
    - The terms lobster claw and ectrodactyly, used by some, should be discouraged (Birth defects surveillance: atlas of selected congenital anomalies).
  - ***Q71.8*** Other reduction defects of upper limb(s). Includes:
    - Congenital shortening of upper limb(s)
  - ***Q71.9*** Reduction defect of upper limb, unspecified
  - ***Q72.0*** Congenital complete absence of lower limb(s)
  - ***Q72.1*** Congenital absence of thigh and lower leg with foot present
  - ***Q72.2*** Congenital absence of both lower leg and foot
  - ***Q72.3*** Congenital absence of foot and toe(s)
  - ***Q72.4*** Longitudinal reduction defect of femur. Includes:
    - Proximal femoral focal deficiency
  - ***Q72.5*** Longitudinal reduction defect of tibia
  - ***Q72.6*** Longitudinal reduction defect of fibula

- ***Q72.7*** Split foot
  - Complete or partial absence of central toes and metatarsals.
  - The term ectrodactyly, used by some, should be discouraged (Birth defects surveillance: atlas of selected congenital anomalies).
- ***Q72.8*** Other reduction defects of lower limb(s). Includes:
  - Congenital shortening of lower limb(s)
- ***Q72.9*** Reduction defect of lower limb, unspecified
- ***Q73.0*** Congenital absence of unspecified limb(s). Includes:
  - Amelia NOS
- ***Q73.1*** Phocomelia, unspecified limb(s). Includes:
  - phocomelia NOS
- ***Q73.8*** Other reduction defects of unspecified limb(s). Includes:
  - Ectromelia NOS
  - Hemimelia NOS
  - Reduction defect
  - Longitudinal reduction deformity of unspecified limb(s)

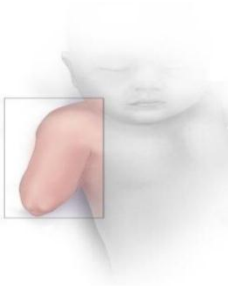
- ***Illustrations:***

Q71.0 (Complete absence upper limb)

Q71.1 (Absent upper arms & forearms)

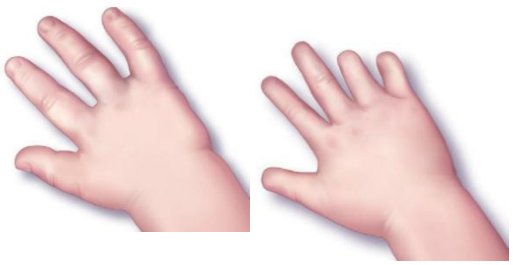


Q71.2 (Absent forearm & hand)



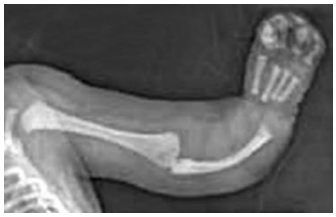
Q71.3 (Absence of hand)

Q71.3 (Absence of fingers)



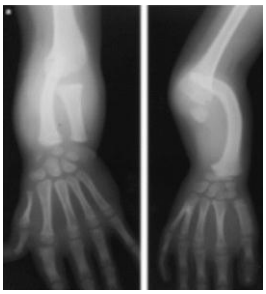
Q71.4

(Longitudinal reduction of radius)

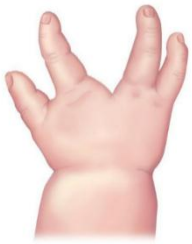


Q71.5

(Longitudinal reduction of ulna)

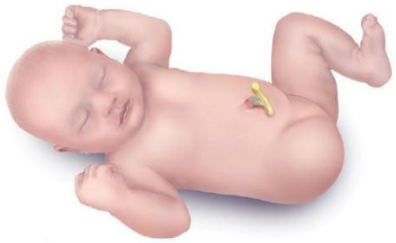


Q71.6 (Lobster-claw hand)

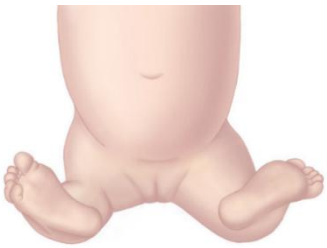


Complete or partial absence of central fingers and metacarpals. Also known as split hand and congenital cleft hand. The terms lobster claw and ectrodactyly, used by some, should be discouraged (Birth defects surveillance: atlas of selected congenital anomalies).

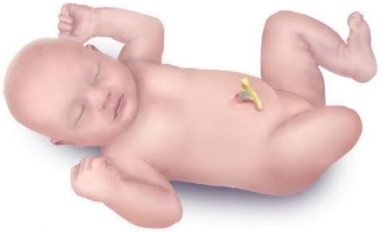
Q72.0
(Complete absence lower limb)



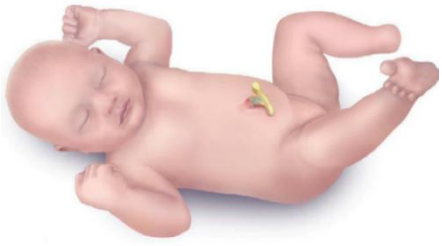
Q72.1
(Absent thighs & lower legs)



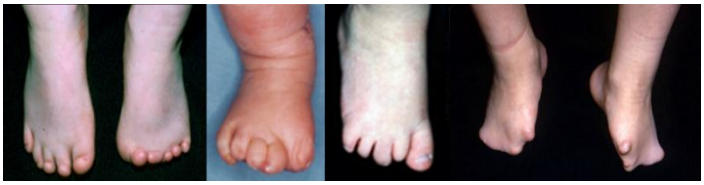
Q72.2
(Absent lower leg and foot)



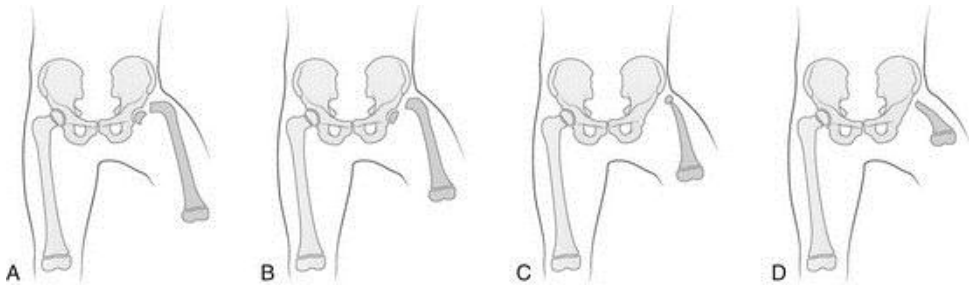
Q72.3 (Absent foot & toes)



Q72.3 (Absent toes)



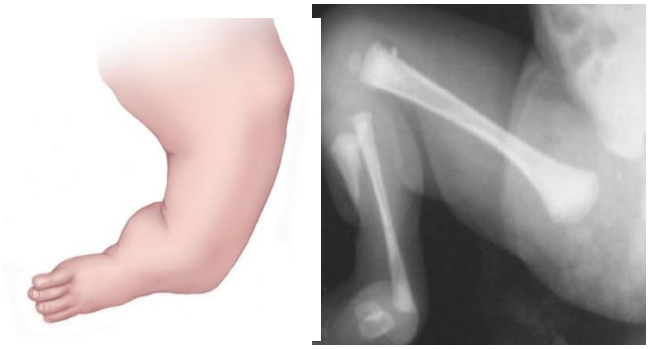
Q72.4 (Longitudinal reduction of femur)



Radiology Key: Congenital Anomalies of the Bone

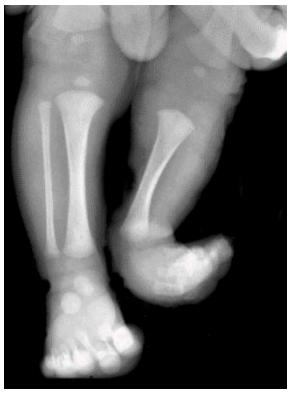
Q72.5

(Longitudinal reduction of tibia)



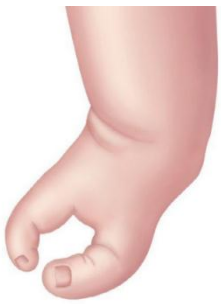
Q72.6

(Longitudinal reduction of fibula)



Indmedica Current Pediatric Research Case Report: Congenital absence of tibia – Type 2

Q72.7 - Split foot



Complete or partial absence of central toes and metatarsals. The term ectrodactyly, used by some, should be discouraged (Birth defects surveillance: atlas of selected congenital anomalies).

10. Diaphragmatic Hernia

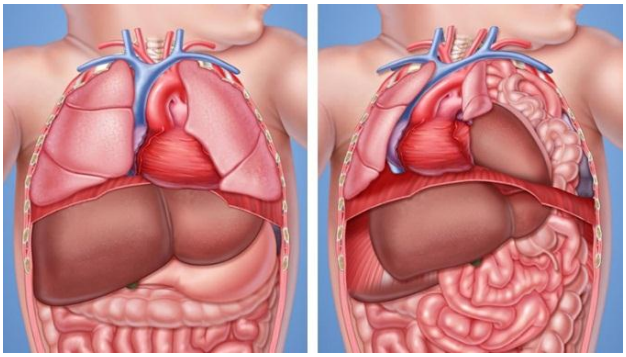
Diaphragmatic hernia (Q79.0): A congenital malformation characterized by herniation of abdominal contents into the thorax through a defect in the diaphragm (ICBDSR definition).

- ***Prenatal diagnoses not confirmed postnatally***: Diaphragmatic hernia may be included in surveillance data when only diagnosed prenatally. However, if it is possible to ascertain the degree of certainty of the prenatal diagnosis, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q79.0*** Congenital diaphragmatic hernia
  - ***Excludes:*** congenital hiatus hernia (Q40.1)

- ***Illustrations: Left:*** The diaphragm is a sheet of flat muscle that separates the heart and lungs from the abdominal cavity.

Right: In congenital diaphragmatic hernia, a hole in the diaphragm allows abdominal organs to move into the chest and restrict lung development.



Children's Hospital of Philadelphia: Congenital Diaphragmatic Hernia (CDH)

11. Prune Belly Sequence

Prune belly sequence (Q79.4): A complex congenital malformation characterized by deficient abdominal muscle and urinary obstruction/distention. It can be caused by urethral obstruction secondary to posterior urethral valves or urethral atresia. In the affected fetus, the deficiency of the abdominal muscle may not be evident. It can be associated with undescended testes, clubfoot and limb deficiencies (ICBDSR definition).

- ***IDC-10 Codes:***
  - ***Q79.4*** Prune belly syndrome

12. Selected Abdominal Wall Defects

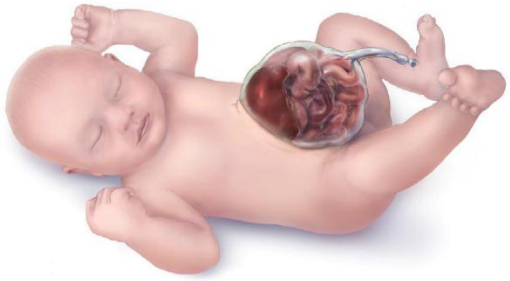
A. Omphalocele/exomphalos (Q79.2): Congenital anomaly of the anterior abdominal wall, in which the abdominal contents (gut, but at times also other abdominal organs) are herniated in the midline through an enlarged umbilical ring. The umbilical cord is inserted in the distal part of the membrane covering the anomaly. The herniated organs are covered by a membrane consisting of the peritoneum and amnion (but this membrane can be ruptured) (Birth defects surveillance: atlas of selected congenital anomalies).

- ***Prenatal diagnoses not confirmed postnatally***: Omphalocele may be included when only diagnosed prenatally. However, it may be difficult to distinguish omphalocele from gastroschisis on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of omphalocele on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***Excludes:***
  - Umbilical hernia (K42.-)

- ***ICD-10-CA coding:***
  - ***Q79.2*** Exomphalos. Includes:
    - Omphalocele

- ***Illustrations:***

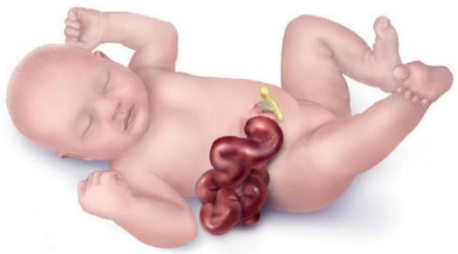


B. Gastroschisis (Q79.3): Gastroschisis is a congenital anomaly of the anterior abdominal wall, accompanied by herniation of the gut and occasionally other abdominal organs. The opening in the abdominal wall is lateral to the umbilicus, and the herniated organs lack a protective membrane. Note that the extruded abdominal contents can be matted and covered by a thick fibrous material, but this membrane does not resemble skin (Birth defects surveillance: atlas of selected congenital anomalies).

- ***Prenatal diagnoses not confirmed postnatally***: Gastroschisis may be included when only diagnosed prenatally. However, it may be difficult to distinguish gastroschisis from omphalocele on prenatal ultrasound, and the terms sometimes are used interchangeably. If it is possible to ascertain the degree of certainty of the prenatal diagnosis and the location of the umbilical cord insertion relative to the abdominal defect, this should factor into the decision as to whether or not to include an individual case in the surveillance data. Live-born children who survive should always have confirmation of the defect postnatally before being included. In addition, the absence of gastroschisis on prenatal ultrasound does not necessarily mean that it will not be diagnosed after delivery (NBDPN, 2017).

- ***ICD-10-CA coding:***
  - ***Q79.3*** Gastroschisis

- ***Illustrations:***



13. Selected Chromosomal Defects

A. Down syndrome, Trisomy 21 (Q90): Typically, a baby is born with 46 chromosomes. Babies with Down syndrome have an extra copy of one of these chromosomes, chromosome 21. A medical term for having an extra copy of a chromosome is 'trisomy.' Down syndrome is also referred to as Trisomy 21. This extra copy changes how the baby's body and brain develop, which can cause both mental and physical challenges for the baby.

- ***Prenatal diagnoses not confirmed postnatally***: Down syndrome may be included when only diagnosed through direct analysis of fetal chromosomes or molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization) of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 21 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth since the placenta may contain mosaic cell lines not present in the fetus. Mosaic trisomy 21 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples (NBDPN, 2017).

- ***ICD-10-CA coding:***

  - ***In all cases:*** Common physical features of Down syndrome should not be coded and include (Centers for Disease Control and Prevention: Facts about Down Syndrome):
    - A flattened face, especially the bridge of the nose
    - Almond-shaped eyes that slant up
    - A short neck
    - Small ears
    - A tongue that tends to stick out of the mouth
    - Tiny white spots on the iris (colored part) of the eye
    - Small hands and feet
    - A single line across the palm of the hand (palmar crease)
    - Small pinky fingers that sometimes curve toward the thumb
    - Poor muscle tone or loose joints
    - Shorter in height as children and adults

  - ***Q90.0*** Trisomy 21, meiotic nondisjunction - When the two copies of chromosome 21 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 21 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+21 or 47,XY,+21. This is the most common type of trisomy 21 and is associated with advanced maternal age, particularly of 35 years or greater (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).

- ***Q90.1*** Trisomy 21, mosaicism (mitotic nondisjunction) - Mosaic trisomy 21 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 21. In this instance, the karyotype is written as 46,XY/47,XY,+21, for example (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).

- ***Q90.2*** Trisomy 21, translocation - Translocation trisomy 21 occurs when two separate copies of chromosome 21 are present, but a third copy of part of chromosome 21 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 21. The karyotype is written as 46,XY,der(14;21)(q10;q10),+21, for example (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).

- ***Q90.9*** Down's syndrome, unspecified – use this code when unable to determine whether nondisjunction or translocation. E.g. nuc ish 21q22.13(D21S259x3) or rsa (13,18)x2, (21)x3,(X)x2. Includes:
  - Trisomy 21 NOS

B. Patau syndrome, Trisomy 13 (Q91.4, Q91.5, Q91.6, Q91.7): Trisomy 13, also called Patau syndrome, is a chromosomal condition associated with severe intellectual disability and physical abnormalities in many parts of the body. Individuals with trisomy 13 often have heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), extra fingers or toes, an opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (a cleft palate), and weak muscle tone (hypotonia). Due to the presence of several life-threatening medical problems, many infants with trisomy 13 die within their first days or weeks of life. Only five percent to 10 percent of children with this condition live past their first year.

- ***Prenatal diagnoses not confirmed postnatally***: Trisomy 13 may be included when only diagnosed through direct analysis of fetal chromosomes or molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization) of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth since the placenta may contain mosaic cell lines not present in the fetus. Mosaic trisomy 13 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples (NBDPN, 2017).

- ***ICD-10-CA coding:***

  - ***Q91.4*** Trisomy 13, meiotic nondisjunction - When the two copies of chromosome 13 from one parent do not separate during egg or sperm

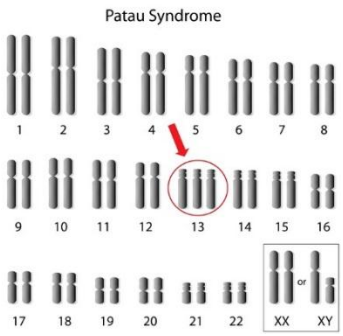
formation, three copies of the entire chromosome 13 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+13 or 47,XY,+13. This is the most common type of trisomy 13 and is associated with advanced maternal age, particularly of 35 years or greater (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).

- ***Q91.5*** Trisomy 13, mosaicism (mitotic nondisjunction) - Mosaic trisomy 13 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 13. In this instance, the karyotype is written as 46,XY/47,XY,+13, for example (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).

- ***Q91.6*** Trisomy 13, translocation - Translocation trisomy 13 occurs when two separate copies of chromosome 13 are present, but a third copy of part of chromosome 13 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 13, e.g. 46,XX,+13,der(13;14)(q10;q10) (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).

- ***Q91.7*** Patau's syndrome, unspecified – This code can be used when unable to determine if nondisjunction or translocation. E.g. nuc ish 13q14(RB1x3) or rsa (13)x3,(18,21,X)x2.

- ***Illustrations:***



NIH U.S. National Library of Medicine: Genetics Home Reference - Trisomy 13

3. **Edwards syndrome, Trisomy 18 (Q91.0, Q91.1, Q91.2,Q91.3)**: Trisomy 18, also called Edwards syndrome, is a chromosomal condition associated with abnormalities in many parts of the body. Individuals with trisomy 18 often have slow growth before birth (intrauterine growth retardation) and a low birth weight. Affected individuals may have heart defects and abnormalities of other organs that develop before birth. Other features of trisomy 18 include a small, abnormally shaped head; a small jaw and mouth; and clenched fists with overlapping

fingers. Due to the presence of several life-threatening medical problems, many individuals with trisomy 18 die before birth or within their first month. Five to 10 percent of children with this condition live past their first year, and these children often have severe intellectual disability (NIH U.S. National Library of Medicine: Genetics Home Reference - Trisomy 18).

Prenatal diagnoses not confirmed postnatally: Trisomy 18 may be included when only diagnosed through direct analysis of fetal chromosomes or molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization) of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 13 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth since the placenta may contain mosaic cell lines not present in the fetus. Mosaic trisomy 18 diagnosed through chorionic villus sampling should always be confirmed by direct examination of fetal chromosomes from amniocentesis, PUBS, or preferably postnatal blood or tissue samples (NBDPN, 2017).

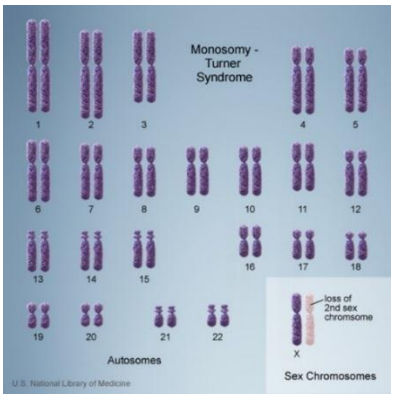
- ***ICD-10-CA coding:***
    - ***Q91.0*** Trisomy 18, meiotic nondisjunction - When the two copies of chromosome 18 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 18 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+18 or 47,XY,+18. This is the most common type of trisomy 18 and is associated with advanced maternal age, particularly of 35 years or greater (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).
    - ***Q91.1*** Trisomy 18, mosaicism (mitotic nondisjunction) - Mosaic trisomy 18 occurs when some, but not all, of the cells in the body contain three copies of all or a large part of chromosome 18. In this instance, the karyotype is written as 46,XY/47,XY,+18, for example (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).
    - ***Q91.2*** Trisomy 18, translocation - Translocation trisomy 18 occurs when two separate copies of chromosome 18 are present, but a third copy of part of chromosome 18 is attached to another chromosome. In this instance, there are 46 total chromosomes present, but 3 copies of part of chromosome 18 (NBDPN Guidelines for Conducting Birth Defects Surveillance, Appendix 3.1).
    - ***Q91.3*** Edwards' syndrome, unspecified – This code can be used when unable to determine if nondisjunction or translocation. E.g. nuc ish 18cen(D18Z1x3) or rsa (13)x2, (18)x3,(21,X)x2.

4. **Turner syndrome (Q96):** Presence of an absent or structurally abnormal second X chromosome in a phenotypic female (NBDPN, 2017).

    - ***Prenatal diagnoses not confirmed postnatally***: Turner syndrome can be included only when diagnosed through direct analysis of fetal chromosomes (karyotype) or molecular cytogenetic analysis of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic Turner syndrome is noted, the abnormality should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (NBDPN, 2017).

    - ***ICD-10-CA coding:***
        i. ***Q96.0*** Karyotype 45,X
        ii. ***Q96.1*** Karyotype 46,X iso (Xq)
        iii. ***Q96.2*** Karyotype 46,X with abnormal sex chromosome, except iso (Xq)
        iv. ***Q96.3*** Mosaicism, 45,X/46,XX or XY
        v. ***Q96.4*** Mosaicism, 45,X/other cell line(s) with abnormal sex chromosome
        vi. ***Q96.8*** Other variants of Turner's syndrome
        vii. ***Q96.9*** Turner's syndrome, unspecified

    - ***Illustrations:***



Appendix 2. Literature for Evaluation of Sources

Some suggested literature for review when evaluation sources include the following:

1. Boutlet SL, Shin M, Kirby RS, Goodman D, Correa A. Sensitivity of birth certificate reports of birth defects in Atlanta, 1995-2005: Effects of Maternal, Infant, and Hospital Characteristics. Public Health Rep. 2011;126(2):186-194.
2. Frohnert BK, Lussky RC, Alms MA, Mendelsohn NJ, Symonik DM, Falken MC. Validity of discharge data for identifying infants with cardiac defects. J Perinatol. 2005;25(11):737-742.
3. Metcalfe A, Sibbald B, Lowry RB, Tough S, Bernier FP. Validation of congenital anomaly coding in Canada's administrative databases compared with a congenital anomaly registry. Birth Defects Res A: Clin Mol Teratol. 2014;100(2): 59-66.
4. Salemi JL, Tanner JP, Block S, Bailey M, Correia JA, Watkins SM, Kirby RS. The relative contribution of data sources to a birth defects registry utilizing passive multisource ascertainment methods: does a smaller birth defects case ascertainment net lead to overall or disproportionate loss? J Registry Manag. 2011;38(1):30-38.
5. Wang Y, Cross PK, Druschel CM. Hospital discharge data: can it serve as the sole source of case ascertainment for population-based birth defects surveillance programs? J Public Health Manag Pract. 2010;16(3):245-251.