

Chapter 2: Case Definitions

Contents

2.0 Introduction.....	1
2.1 Programmatic Case Inclusion Criteria	1
2.1.1 Geographic Catchment Area.....	1
2.1.2 Residency Status.....	2
2.1.3 Pregnancy Outcome.....	4
2.1.4 Gestational Age	4
2.1.5 Age-at-Diagnosis	5
2.2 Birth Defect Case Definition Criteria and Certainty.....	6
2.2.1 Selecting Birth Defects for Surveillance	6
2.2.2 Certainty of the Birth Defect Descriptions.....	7
2.3 References.....	8

Appendix

Appendix 2A: NBDPN Birth Defect Case Definition Criteria Descriptions

Chapter 2

Case Definitions

2.0 Introduction

Standardized surveillance case definitions are important to meet national and jurisdictional goals and objectives. These include pooling data from multiple birth defects surveillance programs to produce national prevalence estimates, comparing data across jurisdictions to identify meaningful differences, and identifying changes over time and space.

For the purpose of these guidelines, a **case** refers to an infant or fetus with at least one birth defect which may be included in a program's surveillance database if it meets criteria. In this chapter, we define two components of birth defects surveillance case definitions: **programmatic case inclusion criteria** and **birth defect case definition criteria**. Programmatic case inclusion criteria and birth defect case definition criteria are influenced by external factors including the birth defects surveillance program's authorizing language (see **Chapter 1**), agency or organization policy and priorities, and available program resources.

Important parameters discussed include the geographic catchment area, residence, pregnancy outcome, gestational age, and age-at-diagnosis, along with the diagnosis itself. NBDPN encourages birth defects surveillance programs to apply the case definition guidance in this chapter to other defects collected and ascertained by their program.

2.1 Programmatic Case Inclusion Criteria

In birth defects surveillance, **programmatic case inclusion criteria** generally refer to:

- Geographic area that defines the surveillance **catchment area**
- Residency status of the mother
- Pregnancy outcome
- Age at which the defect was diagnosed

These criteria are typically consistent across all birth defects to ensure that each surveillance program collects data on cases within its scope and jurisdiction. In this section, we provide additional guidance about each of the programmatic case inclusion criteria to aid program implementation.

2.1.1 Geographic Catchment Area

Recommendation 2.1.1: Birth defects surveillance programs must define the geographic boundaries (i.e., catchment area) of the source population for case identification.

For the purposes of generating and reporting birth defects surveillance data across multiple states, NBDPN recommends that birth defects surveillance programs engage in **population-**

based surveillance. This provides an assurance that every case that occurs within a population in the defined area has the potential to be identified. A defined catchment area (or jurisdiction) may be an entire state, a region, or selected county/counties. Programmatic case inclusion criteria that are jurisdiction-wide and population-based typically enable the most comprehensive ascertainment of birth defects cases. Some specific surveillance projects may have a different geographical catchment area depending on the goals, objectives, and resources of the project. When the geographic area is less than statewide, the following factors should be considered:

- Define the specific geographic boundaries, and determine how the boundaries are calculated (e.g., city, county, or region boundaries).
- Identify health care services that are *outside* of the geographic boundaries but regularly utilized by the population *within* the geographic area. This information is useful for determining effective boundaries for the surveillance area to align the population's location and health services as closely as possible with the geographic boundaries of the inclusion criteria, and planning which data sources to collaborate with for the surveillance functions of case identification, investigation, and verification (see **Chapter 5**).
- Ensure that for the area captured, the birth defects surveillance program can access population-based birth information to serve as the denominator and enable accurate prevalence calculations and monitoring (see **Chapter 8** [June 2004 Edition]).

2.1.2 Residency Status

Recommendation 2.1.2a: Birth defects surveillance programs must define the programmatic case inclusion criteria for residency.

Defining criteria for residency enables a birth defects surveillance program to calculate rates within the population, evaluate changes in these rates over time, plan for prevention and intervention services, and assess program goals and effectiveness. Population-based birth defects surveillance programs must strive to ascertain birth defects that occur among the entire population residing within a defined geographic catchment area at the time of pregnancy outcome.

Criteria for residency in a birth defects surveillance program usually refers to the mother's place of residence at the time of the pregnancy outcome, which appears straightforward. However, there are some special considerations:

- **Residency at different points in time:** In cases when a child or parent/family has moved, the residence at the time of delivery may differ from the residence at the time of diagnosis, or at the time the birth defects surveillance program received the documents for case identification, and/or the current residence.

Recommendation 2.1.2b: For surveillance purposes and when available, birth defects surveillance programs should use the address on the vital record to determine whether the residence meets the programmatic case inclusion criteria for residency.

This helps avoid double-counting a case when the residency has changed to include addresses in different catchment areas over time. For birth defects surveillance programs with referrals to services, many use the current address to ensure the case can be connected to services in the area where they currently live, regardless of whether the case was born elsewhere.

- **Referrals during pregnancy:** Women who reside in one state or community may travel outside that area – such as to an adjacent state, specialty care center, or military facility – for obstetric care, delivery, and/or postnatal care. In these instances, the mother’s place of residence at the time of delivery (rather than the actual location of the delivery) should be used to determine whether to include her pregnancy in the birth defects surveillance database.
- **Multiple addresses:** More than one residential address may be identified for an individual woman. For example, the address of the health insurance policyholder listed in a hospital delivery record may differ from the mother’s address listed on a birth certificate. In cases without a vital record, the birth mother’s address at the prenatal visit closest in time to the pregnancy outcome or the address recorded in the delivery record are appropriate alternatives.
- **Whether a residence is located within the surveillance area:** Particularly for birth defects surveillance programs with a catchment area that does not include the entire state, it may be challenging to determine whether a particular address lies within the surveillance area. Birth defects surveillance programs should develop standard procedures for distinguishing addresses within their area. Potential references include street maps, US Postal Service listings, tax assessor records, census tracts, and the latitudes and longitudes defining the surveillance area (*geocodes*). The accuracy of geocoding will depend on the accuracy of the addresses to which the latitude and longitude are assigned.

Standard 2.1.2

Residency Status of Cases Included in Birth Defects Surveillance Program’s Activities	
If your program is not state-level, then take “in-state” to mean “in your program’s catchment area.”	
Core	In-state deliveries to in-state residents.
Enhanced	Core tier criteria plus the following: Out-of-state deliveries to in-state residents (e.g., a resident may get care or deliver at a major medical center on the border of a neighboring state).

Including in-area residents whose pregnancy outcome occurs outside the surveillance area, and *excluding out-of-area residents* whose pregnancy outcome occurs within the catchment area, are essential to conduct comprehensive surveillance. Whether an individual birth defects surveillance program attains this level of comprehensiveness will depend on how frequently residents travel outside the surveillance area to receive care, the magnitude of the potential impact such out-of-area care may have on birth defects rates, the staff and resources available for tracking services received, and the existence of data-sharing relationships with facilities and birth defects surveillance programs outside the catchment area. Primary care physicians and electronic case

reporting are often helpful resources for following up on continuing care.

2.1.3 Pregnancy Outcome

Recommendation 2.1.3: Birth defects surveillance programs must define the set of pregnancy outcomes to be included as a component of programmatic case inclusion criteria.

Ideally, for birth defects surveillance to be comprehensive, all birth defects occurring in a population should be ascertained regardless of the pregnancy outcome (i.e., live birth, fetal death, other, unknown).

Standard 2.1.3

Pregnancy Outcomes Included in Birth Defects Surveillance Program's Activities	
Core	Live births
Enhanced	<i>Core tier criteria plus some/all of the following:</i> Fetal deaths delivered at a gestational age of ≥ 20 completed weeks (i.e., stillbirths) >350 grams if gestational age is unknown
Extended	<i>Core tier criteria, plus some/all items from Enhanced, plus some/all of the following:</i> Elective terminations of pregnancy at any gestational age Fetal deaths delivered at a gestational age of < 20 completed weeks (i.e., miscarriages)

Reference: MODEL Law: <https://www.cdc.gov/nchs/data/misc/mvsact92b.pdf>

Each birth defects surveillance program should consider the advantages and limitations when determining whether to expand its inclusion criteria to include non-live births. Ascertainment of reported outcomes other than live birth often depends on the birth defects surveillance program's authorizing language. Other considerations include program goals, objectives, availability of staff and resources, access to case ascertainment sources (e.g., prenatal diagnostic and clinic sites, fetal death certificates; see **Appendix 5A**), accessibility of information about specific outcomes, and the magnitude of the potential impact that excluding these outcomes would have on individual defect rates. For additional guidance on data analysis and interpretation, see **Chapter 8** [June 2004 Edition].

2.1.4 Gestational Age

Gestational age at the time of delivery is a key consideration for defining pregnancy outcomes (see section 2.1.3 above). Furthermore, the frequency of some birth defects may vary by gestational age, leading to variations in rates depending on the length of gestation. For example, some defects are identified more frequently among preterm infants. Others, such as persistence of the patent ductus arteriosus and undescended testes, may be abnormal in term infants but considered physiological (i.e., not abnormal) in preterm infants considering their developmental timeline. Some small ventricular septal defects that are present at birth in preterm infants might have closed during the last weeks of gestation if the pregnancy had continued to term.

Gestational age may be derived in various ways based on measurement of the fetus by prenatal ultrasound, the date of the last menstrual period, or the newborn clinical exam. These methods may not be equally accurate and may yield conflicting results.

Recommendation 2.1.4: When multiple estimates of gestational age are ascertained for an individual case, the NBDPN suggests using the most accurate method available in the records to determine the case status.

The methods below are listed according to their generally accepted accuracy for calculating gestational age:

Accuracy	Method of Estimating Gestational Age
Highest	Prenatal ultrasound with a reported gestational age of < 14 weeks
Moderate	Date of the last menstrual period Prenatal ultrasound with a reported gestational age of $\geq$ 14 weeks
Lowest	Clinical exam after delivery

American College of Obstetricians and Gynecologists (2017); Deputy NP, et al. (2017)

2.1.5 Age-at-Diagnosis

Recommendation 2.1.5: Birth defects surveillance programs must define the maximum age by which a birth defect must have been diagnosed for it to meet the programmatic case inclusion criteria for age-at-diagnosis.

Age-at-diagnosis refers to the age of the child when the defect was diagnosed, or at which the signs or symptoms were first recognized as potentially representing the birth defect. Of note, the date the case is first reported to the birth defects surveillance program is not synonymous with the date the birth defect was diagnosed. Often, but not always, the diagnosis date is close in time to a date of hospital admission or discharge, the date of a specialty outpatient appointment, or the date the data source processed the case data for reporting, but dating is affected by other parameters which may vary widely. Birth defects surveillance programs should develop standard procedures for documenting when birth defects were first diagnosed.

Certain birth defect types tend to take longer for the diagnosis to manifest, or for the level of severity to become fully understood. When appropriate, birth defects surveillance programs may utilize a different maximum age-at-diagnosis for such birth defect types, in order to allow a longer case identification period. For example, a birth defects surveillance program might collect cardiovascular conditions diagnosed until 10 years of age, but only collect other birth defects diagnosed up to 2 years of age.

Standard 2.1.5

Up to What Age-at-Diagnosis the Surveillance Program Ascertains Birth Defects Age at which the signs and symptoms brought the case to medical attention and the diagnosis was suspected. Although definitive test results for final diagnosis may have taken longer, the date of clinical findings that initiated the diagnostic process can be considered the “age-at-diagnosis.”	
Core	Birth defects surveillance program ascertains cases diagnosed on or before the first birthday.
Enhanced	<i>Core tier criteria plus the following:</i> Birth defects surveillance program ascertains cases diagnosed after the first birthday. Birth defects surveillance program utilizes a later maximum age-at-diagnosis in conditions for which the diagnosis or level of severity are better understood over a longer timeline.

2.2 Birth Defect Case Definition Criteria and Certainty

Consistent birth defect case definition criteria are critical for the accuracy of birth defects surveillance data. Inclusion of cases with uncertain or incorrect diagnoses can inflate estimated birth defect prevalence rates and impair the accuracy of surveillance data. For more information on calculating positive and negative predictive values, refer to **Chapter 8** [June 2004 Edition].

In birth defects surveillance, the *birth defect case definition criteria* refer to criteria that are specific to the defect or diagnosis. Case definitions are unique to each birth defect and inform surveillance program decisions on whether a case should be included in the surveillance system or not. The use of consistent birth defect case definition criteria ensures that data are comparable and can be aggregated across programs. **Appendix 2A** provides detailed case definitions to serve as inclusion and exclusion criteria for each individual birth defect, along with guidance on determining the certainty of the diagnosis from a surveillance standpoint. In this section, we discuss the approach to selecting which birth defects to include in a program’s scope.

2.2.1 Selecting Birth Defects for Surveillance

The birth defects surveillance program’s authorizing language, its agency or organization’s policies and priorities, and available resources all influence the set of birth defect diagnoses included in the program’s scope. At a minimum, programs should strive to ensure their scope includes all of the birth defects on the NBDPN Core list (see **Appendix 2A**).

Standard 2.2.1

Set of Defects Included in Birth Defects Surveillance Program's Activities (see Appendix 2A)	
Core	Defects on the NBDPN Core list of birth defects.
Enhanced	<i>Core tier criteria plus the following:</i> Defects on the NBDPN Enhanced list of birth defects.
Extended	<i>Core and Enhanced tier criteria, plus some or all of the following:</i> Diagnoses not included on the NBDPN list but included in birth defects surveillance program activities as needed to meet jurisdictional goals.

The NBDPN Core birth defects in **Appendix 2A** serve important purposes for monitoring and estimating prevalence, informing public health activities, and supporting public health research. Twenty-two birth defects were selected as Core using the following rationale:

- High importance to public health and/or public health research priorities
- > 90% of cases diagnosed before 1 year of age
- > 90% of cases are live births
- Clinical presentation that lends itself to ease of surveillance (i.e., consideration for factors such as the need for specialized testing, or whether the condition has a spectrum of severity that may impact the consistency and completeness of diagnosis and reporting)

Outside of the NBDPN Core list, birth defects from the NBDPN Enhanced list or beyond may be added to the scope of a birth defects surveillance program's activities to support broader goals and objectives. While programs can create their own defect-specific inclusion criteria for internal use, only the cases that meet NBDPN birth defect case definitions should be reported to NBDPN.

Recommendation 2.2.1a: Birth defects surveillance programs should meet data quality standards for Core defects before expanding to include Enhanced or Extended conditions.

Recommendation 2.2.1b: For any defect under surveillance, birth defects surveillance programs should use the NBDPN birth defects case definition criteria in **Appendix 2A** when determining whether to report the case to NBDPN.

2.2.2. Certainty of the Birth Defect Descriptions

The certainty of a birth defect diagnosis can be affected by several factors, including the type of healthcare facility reporting the case; diagnostic procedures performed; level of detail with which the defect is described; expertise of the person describing the defect (e.g., nurse, therapist, general physician, subspecialist); and the timing of the diagnosis. **Appendix 2A** includes descriptions of certainty for diagnosis (accepted versus confirmed) and NBDPN guidance for inclusion of less certain diagnoses. Additional information can be found in the **NBDPN Certainty of Diagnosis Fact Sheet**.

2.3 References

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