Chapter 6

Case Ascertainment Methods

Table of Contents

Appendices

6.1 Introduction

The National Birth Defects Prevention Network (NBDPN) is committed to improving the quality, accuracy, completeness, comparability, and timeliness of birth defects surveillance data. Information on the prevalence of birth defects reported by surveillance systems can vary considerably due to differences in case definition, method of case ascertainment, and the types of data sources used.

This chapter describes two major approaches to birth defects surveillance: active case ascertainment and passive case ascertainment. The ***active case ascertainment*** approach is the intensive level of case identification that involves staff finding cases at strategic data sources. Ascertainment is usually very complete, and each diagnosis in the database is confirmed. In the ***passive case ascertainment*** approach the surveillance program receives case reports of birth defects from data sources. The completeness of ascertainment is highly dependent on the number and types of data sources used by the surveillance program and on the consistency of case reporting from the data sources. Since case reports usually are not confirmed by staff in a passive case ascertainment program, it is particularly important for these programs to implement quality assurance procedures aggressively.

Although the two surveillance approaches are operationally different, it is possible to achieve comparable levels of data quality. Programs should evaluate their surveillance approaches regularly for accuracy, completeness, and timeliness and should be creative in identifying strategic means of quality improvement.

In this chapter we first introduce some relevant terminology (Section 6.2). We then discuss general surveillance development (Section 6.3) and introduce approaches to case identification (Section 6.4). In Sections 6.5 and 6.6 we present in some detail information on the two main approaches to case identification (active and passive case ascertainment, respectively). The remaining sections cover additional topics in case ascertainment, including data sources (Section 6.7), sources of information that may be available at a given data source (Section 6.8), and issues relating to infant risk factors and case identification (Section 6.9). References cited in this chapter may be found in Section 6.10.

Appendices to this chapter provide additional detail on the following important data sources for birth defects surveillance: vital records (Appendix 6.1), hospital data sets (Appendix 6.2), hospital and patient services logs (Appendix 6.3), and genetic services (Appendix 6.4).

6.2 Terminology

Surveillance (public health)	The ongoing, systematic collection, analysis, and interpretation of health data essential to the planning, implementation and evaluation of public health practice, closely integrated with the timely dissemination of these data to those who need to know (Centers for Disease Control and Prevention, 1988).
Monitoring	The performance and analysis of routine measurements using statistical methods aimed at detecting changes in the environment or health status of populations (Last, 1995).
Registry	A system of ongoing registrations, such that cases of a disease or other health-relevant condition are defined in a population and can be related to a population base. Birth and death registration systems are examples. Some disease registries, like the cancer registry, closely resemble public health surveillance systems and have epidemiologic value (Last, 1995).
Case ascertainment or identification	The process of identifying – from existing sources and using defined case definitions – embryos, fetuses, neonates, infants, and children who have a birth defect.
Active case ascertainment	A surveillance approach to case identification that is based on surveillance staff being engaged intensively in all activities related to finding and confirming potential birth defects cases. Surveillance staff seek out data sources and conduct systematic investigations of pertinent sources of information to identify potential cases of birth defects. Data collection sites include hospital medical records, diagnostic indices, unit logs, pathology departments, and specialty sites.
Passive case ascertainment	A surveillance approach to case identification whereby birth defects programs receive case reports from data sources. Passive case ascertainment programs receive case reports from one or many different data sources and may accommodate multiple reporting formats including hard copy, electronic, and web-based reports, as well as administrative data sets. There may be variability in the completeness and accuracy of case ascertainment in programs that use this type of case ascertainment.
Population-based surveillance	Surveillance that identifies a population under study, usually defined by geopolitical boundaries, and establishes the denominator from which cases come. A data source that is population based covers an entire population within a defined area. Some examples of population-based data sources are: vital records (birth, death, and fetal death), statewide newborn genetic screening programs, and statewide newborn hearing screening programs.

Data source	Any facility, site, or entity that has cases or potential cases of birth defects or other pertinent medical information. This includes a hospital, clinic, physician's office, laboratory, prenatal diagnosis center, as well as administrative databases.
Reportable disease	A disease, laboratory result, or health condition of public health significance that requires notification of its occurrence to a public health agency. Authorizing legislation or regulations usually define which conditions are reportable, which data sources are required to report, timelines for reporting, and what demographic information is required, at a minimum, in a case report.
Reporting source	A data source that is required, by law, to report or allow access to cases of birth defects and other pertinent medical conditions to the birth defects program.
Administrative data set	A data set or database that is created to fulfill operational or managerial objectives. Many are developed as information management systems with multiple functions. Examples include hospital discharge data, Medicaid data, vital records master index, clinical management information systems, health care billing and insurance claims systems.
Unit	A component, section, or department within a data source that serves a specific function or performs a specific activity. Examples include health information management department, labor and delivery unit, neonatal intensive care unit, and pathology department
Data collection	The process of gathering information, which includes receiving, retrieving, accessing, abstracting, and extracting information from information sources.
Medical records review (information review)	The process of reading, identifying, interpreting, and translating documentation per specific program objectives. Medical records review precedes abstracting.
Abstracting	The process of recording information, identified when reviewing documentation in a medical record or other information source, and entering the information into data fields in a specified format. Information may be recorded on hard copy forms or through formatted data entry computer screens.
Disease coding	The process of assigning a standardized disease code (e.g., ICD-9-CM or 6-digit CDC code) to medical information.
Case abstract or case record	The documentation file(s) containing complete information about the birth defects case.

6.3 General Surveillance Development

Birth defects surveillance systems should be developed to facilitate the essential activities of data collection, data analysis, data evaluation, and information dissemination consistent with a program's established goals and objectives. The general guidelines below can be applied to developing a new system or improving an existing system. We are indebted to Mausner and Bahn (1974) and Teutsch and Churchill (2000) for much of the information in this section.

In the following sections we discuss planning and documenting the system (Section 6.3.1), identifying data sources (Section 6.3.2), obtaining knowledge about individual data sources (Section 6.3.3), implementing data quality procedures (Section 6.4.3), and evaluating surveillance method and analytical capability (Section 6.3.5).

6.3.1 Plan and Document

A birth defects surveillance program must be organized and have supporting documentation before beginning operations. The program can begin to process case reports once the logistics of case identification and data collection are established with data sources. Therefore, it is important to engage surveillance staff, data sources, stakeholders, advisors, and others affected by program operations early in the planning process.

The program should:

- **Understand** the legal authority and restrictions that shape surveillance operations, including processes for changing or amending legislation (see Chapter 2 on Legislation).
- **Develop** a mission statement and define the surveillance program's goals and objectives. Determine what outcome measurements are desired by the program. For example, the program may want to describe the distribution of birth defects in their population, calculate rates and perform statistical analyses, and identify children who require services. Ideally, the development of goals, objectives, and outcome measurements will be done in collaboration with stakeholders and with internal and external advisory groups.
- **Define** the parameters of case definition for the surveillance program, including residency, pregnancy outcomes, eligible diagnoses, and age range. Define the minimum criteria for an eligible case report (see Chapter 3 on Case Definition).
- **Define** the method of case identification that will be used. Usually, a program will develop an infrastructure to support functions of active or passive case ascertainment. It is essential to document procedures, protocols, decision items, and methods of data collection (the program's surveillance approach). Records review and data collection procedures should be defined precisely.
- **Determine** the data variables needed to fulfill program goals and objectives. Define the minimum information that must be collected and address other information that would be beneficial to the surveillance program (see Chapter 4 on Data Variables).
- **Document** protocol and procedures regarding the privacy of the individual and the confidentiality of health information.

- **Design** forms for reporting, data collection, and abstracting that are adaptable to computer technology. This could include web-based reporting and forms that provide for easy data entry or scanning and that support abstracting medical records in the field (see Chapter 9 on Data Management and Security).
- **Develop** a database that has record linkage capability and that also functions as an information management system. The database should be flexible, adaptable, and able to accept electronic transfer of data files, web-based case reports, and case record abstracts from multiple sources. The database should support identification of all sources of information through which a diagnosis is identified or reported. It is also useful to be able to track and monitor medical records requests and perform other information management functions (see Chapter 9 on Data Management and Security).

6.3.2 Identify Data Sources

A key component in surveillance is identifying data sources for case ascertainment. A program needs to understand and evaluate the traits, characteristics, and operating procedures of all data sources. This is particularly important if there are potential sources of bias or underreporting associated with the way cases may be identified at a source.

The program should:

- **Identify** all potential data sources able to provide information that will help to fulfill the program's mission (e.g., hospitals, genetics and specialty clinics, cytogenetics laboratories, administrative data sets, vital records).

- **Determine** which data sources are included in any legislation mandating reporting and any additional sources for voluntary reporting. Consideration should be given to recommending legislative changes if program objectives change or are expanded, or if important data sources are omitted from mandatory reporting. For example, when adding prenatal diagnosis surveillance to program operations, it may be necessary to amend legislative language to include new data sources or facilities.

6.3.3 Obtain Knowledge about Individual Data Sources

For each reporting data source the program should:

- **Know** the data source's mission or goals.
- **Identify** professional or legal mandates governing operations of the data source that may affect access to, or quality of, data from that source.
- **Describe** the population served by the data source.
- **Chart** the flow of information that is relevant to the surveillance program through the unit and/or data source. This is a good way to determine how the information is collected originally and whether or not the information is forwarded to a central repository (e.g., centralized computer file, medical records department, administrative database).
- **Maintain** an up-to-date directory of names and contact information for relevant people at the data source (e.g., medical records personnel).

- **Utilize** multiple data sources. Surveillance programs should use multiple data sources, both for case identification and data collection. It is important for the surveillance program to realize that one source rarely fills all of a surveillance system's needs for case record accuracy, completeness, and timeliness.
- **Develop** record linkage procedures to facilitate matching all reports to the correct case record. This is especially important when programs utilize multiple data sources (see Chapter 9 on Data Management and Security).

6.3.4 Implement Data Quality Procedures

Surveillance programs should evaluate data for completeness, accuracy, timeliness, and comparability to other birth defects programs. At a minimum, programs should develop quality assurance procedures (manual and/or computerized) to identify potential issues in data quality. This includes accuracy, completeness, and timeliness. Additionally, programs should maintain documentation on data collection, data abstraction, and medical records review procedures. This will reduce the risk of multiple interpretations that lead to an inconsistent application of procedures and interpretation of medical information. For further details, see Chapter 7 on Data Quality Management.

6.3.5 Evaluate Surveillance Method and Analytical Capability

Surveillance programs should evaluate the surveillance approach and determine whether the surveillance system is meeting program objectives. Additionally, outcome measurements should be evaluated. NBDPN recommends the guidelines offered in the document *Updated Guidelines for Evaluating Public Health Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

6.4 Birth Defects Surveillance – Approaches to Case Identification

Cases of birth defects are generally identified in one of two ways: through 'active case ascertainment' (i.e., staff conduct case finding) or through 'passive case ascertainment' (i.e., case reports are received by the program). While some surveillance systems use both kinds of ascertainment approaches for case identification, program activities are generally structured around one or the other approach.

Birth defects rates are directly related to the method of case identification and type of surveillance approach. Table 6.1 presents birth defects rates based on various surveillance approaches (Edmonds, 1997).

Table 6.1 Birth Defects Rates by Surveillance Approach

Data Source	% of Babies Reported with Birth Defects
Birth Certificates in 1996	1.5
Newborn Hospital Discharge Data (Florida)	4.3-7.1
Mandatory Hospital Reporting (New York)	3.4
Linked Data Sources (North Carolina)	4.7
Active Hospital Surveillance (Atlanta 1992-1996)	2.6
Physical Exam of Infants (Collaborative Perinatal Project)	8.3

Although a physical examination of each infant provides the most complete assessment of birth defects among births, and therefore the highest prevalence, few programs can sustain this type of intensive case ascertainment. At the other extreme, the prevalence at birth of defects is clearly underreported when only birth certificates are used in case ascertainment. The NBDPN promotes case ascertainment approaches that provide a more complete description of birth defects prevalence in the US.

Whereas the previous section on general surveillance development (Section 6.3) provides a foundation for surveillance systems, the following two sections (Sections 6.5 and 6.6) discuss unique issues that arise in using either active or passive case ascertainment approaches in the identification of birth defect cases. We are indebted to Lynberg and Edmonds (1992) for much of the information in Sections 6.5 and 6.6.

6.5 Active Case Ascertainment

With active case ascertainment, cases of birth defects are identified at data sources by surveillance staff. The case-finding process includes identifying potential birth defects cases, reviewing and abstracting information from medical records, and conducting follow-up in order to complete abstracts or verify information. Programs take measures to ensure complete case ascertainment by using multiple data sources and multiple units within data sources. Case-finding activities may vary depending on the program's resources and objectives. A program's resources, as well as program goals and objectives, should be used to determine the intensity of case finding. Regardless of the case-finding methods used, active surveillance programs should provide detailed instructions on the case-finding process, document procedures for collecting information and completing case abstracts, nurture relationships between the program and its data sources, evaluate the quality and effectiveness of all steps in the case-finding process, and implement quality improvement methods.

In the sections below we discuss characteristics of active case ascertainment (Section 6.5.1), a recommended approach for active case ascertainment (Section 6.5.2), data quality issues in active case ascertainment (Section 6.5.3), evaluation (Section 6.5.4), and tips and hints for active case ascertainment (Section 6.5.5).

6.5.1 Characteristics of Active Case Ascertainment

- *Surveillance staff identify birth defects cases by visiting data sources.* Staff should follow a thorough and systematic set of investigative methods so that all potential birth defects cases are identified.
- *Surveillance staff are trained to find birth defects cases.* Staff learn how to find (or ***cull***) cases in hospitals, medical facilities, clinics, laboratories (e.g., cytogenetics laboratories, genetics clinics, prenatal diagnostic centers), and in medical records that relate to each potential case (e.g., prenatal, maternal delivery, newborn, infant, pediatric).
- *Staff are trained to gather information from information sources and medical records.* This includes following abstracting procedures and documentation guidelines. Staff are trained in birth defects coding and learn how to conduct follow-up.
- *Multiple information sources are used to obtain data.* All potential data sources should be part of the case-finding investigative process, and some are essential (e.g., birth hospitals, unit logs in birth hospitals). Surveillance systems should evaluate the effectiveness of case finding at each data source.
- *Case abstract forms are detailed and comprehensive and usually include a number of variables pertaining to the pregnancy, delivery, and outcome.* Information on the mother and infant is often collected in detail, including medical and prenatal care history, complications of pregnancy or delivery, reproductive history, physical examinations, postnatal procedures, and birth defects diagnosis.
- *Clinical reviewers, usually physicians, are trained to confirm, qualify, and evaluate the diagnostic information collected* by the surveillance abstractors.
- *Active case-finding surveillance should result in accurate and complete identification of birth defects cases.* The data are of high quality due to extensive staff training. The data collected are comprehensive and result in a detailed case abstract.

6.5.2 Approach to Active Case Ascertainment

Active surveillance is based on surveillance staff investigating data sources and finding potential birth defects cases. Although other activities may be part of the active surveillance approach, case finding is the primary task. There are various approaches to the case-finding process. Some programs have staff review all pertinent data sources and information reports, while others limit case finding to the most important information sources. Some use existing databases or lists of potential cases that are generated by the data source. Because case finding is labor intensive, most programs evaluate case-finding activities and determine ways to identify cases effectively and efficiently, yet still be relatively sure that case ascertainment is complete. It is necessary to take into account the legal or legislative issues that govern program activities.

Essential program activities for active case ascertainment include those listed below.

- **Identify** program objectives. It is important to develop or enhance the case-finding approach based on the purpose and objectives of the surveillance system. For example, if information is used to refer children to services, then the case-finding process should be designed to collect identifying and contact information early enough in the process to make the referral in a timely manner.
- **Develop** a flow chart of the case-finding process. Identify the data sources that are consistently used for case finding. At a minimum the program must conduct extensive case finding at birth and major pediatric hospitals. Within the data sources, the program should identify which units and departments will always be used. Important units and departments to consider are labor and delivery, nursery, surgery, and pathology (see list of data sources in Section 6.7). Some programs use the medical records department to generate a list of diagnoses (i.e., disease codes) from the disease index.
- **Define** the type of information to look for and collect during the case-finding process. Information gathered may be sketchy, incomplete, and general. This is especially true when gathering information from unit logs. The case-finding process may also include gathering information for the conditions of low birth weight, prematurity, and other conditions that may potentially lead to a case.
- **Define** the frequency of case-finding activities (i.e., visiting sources of information and completing abstracts). Frequency and consistency of case-finding activities affect the timeliness of the surveillance database. For example, if the program identifies a child who needs to be referred for services, it is usually important for the referral to be made in a timely manner. Timeliness can be measured by setting goals for the maximal length of time between birth and referral.
- **Conduct** case finding (culling). This is the systematic and ongoing process of identifying birth defects cases. Potential cases at the data source are found by surveillance staff through one or more procedures: (1) reviewing information at unit logs within a data source and creating a list of medical records to be pulled by the health information department within the data source; and/or (2) requesting a line listing of potential cases from the data source or unit, usually by identifying the cases by ICD codes (e.g., hospital index); or (3) reviewing the medical records for every delivery, termination, miscarriage, etc. occurring at the data source.

- **Conduct** medical records reviews. Potential cases of birth defects identified by the case-finding process are further investigated through medical records reviews. Requests for medical records are provided to staff of medical records departments at hospitals (or other sites), who pull the charts and make them available to surveillance system staff. Surveillance staff, who determine if the child or fetus meets the eligibility criteria for inclusion as a case, review the medical records. Multiple medical records may be reviewed during this process. These may include: maternal medical records during prenatal care, hospital admits during the pregnancy, and the delivery record. Medical records for a child include the newborn delivery record and any medical (hospital) records generated after the birth.
- **Abstract** information. As medical records are reviewed, surveillance staff abstract (record) the required information and record it on the case abstract form. Trained surveillance staff follow program guidelines and procedures for completing the data elements on the case abstract, confirming a diagnosis, and conducting follow-up to find cases at data sources and within units at data sources. Although a surveillance program develops its own set of abstracting guidelines and procedures, these should be based on established guidelines when available. In some programs, the abstractor assigns the disease code. In others, assigning the disease code occurs separately from abstracting.
- **Perform** a clinical review. Some programs have an expert in medical diagnosis issues review the case abstract after it is complete. The abstract is evaluated for incomplete data variables (i.e., data fields), accuracy of the medical information, and accuracy of the disease code assigned. Some clinical reviews result in the further classification of the case with a summary diagnosis, as an isolated or syndromic case, or other classification.

6.5.3 Data Quality Issues in Active Case Ascertainment

Active case finding requires surveillance staff to review and collect information from medical records. Staff are involved directly in verifying and confirming medical information and determining whether further follow-up or investigation is needed. In these programs, the burden of maintaining the quality of the database rests with the surveillance staff. It is essential to understand the challenges to data quality that occur in active surveillance and to implement strategies to identify and to correct them (see also Chapter 7 on Data Quality Management).

- *Field work (case finding, record review, abstracting) should be evaluated for accuracy, incomplete data variables, and consistency.* Desired outcome benchmarks in each of these areas should be identified and improvements implemented and tracked.
- *Data sources and individual units should be evaluated with respect to the staff resources expended and the results obtained.* Since case finding is labor intensive, programs should streamline and improve operations whenever possible. The value of the output of each unit or department utilized should be evaluated against the staff resources used. The program should determine whether unnecessary medical records are being reviewed and identify which non-anomaly ICD codes are most effective in identifying potential cases.
- *The surveillance database should be evaluated for timeliness.* This includes measuring how current the database is in relation to calculating disease rates. Although programs may collect information on individual birth defects cases over many months or years, they should set benchmarks for finalizing an individual case record or meeting a level of productivity by specified times.

6.5.4 Active Case Ascertainment Surveillance Evaluation

Evaluation of active ascertainment surveillance methods should occur at two levels. Both levels directly impact data quality and the program's ability to meet goals and objectives. One level targets case identification and data collection. Examples of areas that should be evaluated are:

- Completeness
- Accuracy
- Timeliness
- Measurability

Programs should develop outcome measurements that will improve data quality and are important to meet program needs and surveillance objectives. See Chapter 7 on Data Quality Management for a more detailed discussion of this topic.

The other level focuses on the surveillance system itself. For a comprehensive approach to evaluating surveillance systems refer to CDC's *Updated Guidelines for Evaluating Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

6.5.5 Tips and Hints in Active Case Ascertainment

- *Establish precise guidelines and criteria for data requests to data sources.* The process of active case ascertainment includes requesting information from data sources. Specific criteria or data variable parameters should be provided (e.g., ad hoc reports generated from an existing database, extracted information from databases).
- *Visit tertiary care (e.g., major pediatric site) hospitals first.* These sites usually have the most complete diagnostic information on a birth defects case. Surveillance staff can follow the case back to the birth hospital for the remaining information. Often a delivery that occurs in a rural hospital is transferred to the tertiary care facility.
- *Coordinate the schedule of site visits with the data sources* to minimize inconvenience for them.
- *Form relationships with staff in medical record departments (directors, coders, those coordinating release of information, record retrievers), birth registrar at the hospital, and hospital unit staff.* Discuss the purposes of the surveillance program with them and describe the work that surveillance staff perform at their sites.
- *Know key information technology or data processing staff at the data source.* These individuals often can access and retrieve specific pieces of information collected at the data source or within a component unit.
- *Use caution with ICD lists or an ICD disease index generated by medical records (or information technology) staff.* Hospital medical records coders are required to adhere to a set of federal guidelines when assigning a disease code to the medical record. Surveillance staff use a set of abstracting guidelines developed by the program (or NBDPN). Measure the benefit of using a disease code from an index against the output gained and resources used. For example, evaluate the results of a medical records review after using disease codes from an index.
- *Use laptops.* Design computer screens to assist in the case-finding process. Direct data entry during information gathering is more efficient, and likely more accurate, than recording information on paper forms and then entering it into the database.

- *Be conscious of HIPAA*, especially as this relates to the privacy and security rules that covered entities (i.e., health care facilities) are required to follow. Be knowledgeable about public health exemptions in HIPAA. Provide reassuring documentation to sites as appropriate (see Chapter 2 on Legislation).

6.6 Passive Case Ascertainment

With passive case ascertainment, case reports are submitted by data sources to the surveillance program. The level of interaction between programs and reporting sources varies, as do the methods of reporting. Some programs create birth defects case reporting forms and instruct reporting sources on how to complete them. Other programs merge or extract pertinent information into the surveillance program's database from a data source's existing database. Many use a combination of reporting methods to develop as complete an identification of birth defects cases as possible within the resources available. Regardless of the methods used, operating a surveillance system that receives case reports from data sources requires the program to identify and use multiple data reporting sources, provide detailed instructions to case reporting sources, nurture the relationship between the program and the reporting data source, and evaluate the quality of the case reports received.

In the sections below we discuss characteristics of passive case ascertainment (Section 6.6.1), a recommended approach for passive case ascertainment (Section 6.6.2), data quality issues in passive case ascertainment (Section 6.6.3), evaluation (Section 6.6.4), and tips and hints for passive case ascertainment (Section 6.6.5).

6.6.1 Characteristics of Passive Case Ascertainment

- *Birth defects cases are reported by data sources* to the surveillance program.
- *Medical information is received by the program as a case report* and is generally accepted as reported (i.e., the program does not confirm every case report for accuracy or comprehensiveness of diagnostic information).
- *The operational procedures used by each data source influence the accuracy, totality, definition, and timeliness of reported diagnoses.* This, in turn, influences the quality of the data in the surveillance program's database.
- *Information is usually reported from multiple reporting sources.* Data sources often serve different purposes for a program. Many can be used as major sources of clinical information (e.g., hospital reports, hospital discharge index, cytogenetics laboratories). Some are used as a source of demographic and statistical information (e.g., vital records). Others are used primarily for tracking or follow-up (e.g., genetics clinics, pathology and autopsy reports, specialty treatment clinics, and developmental centers). See the list of data sources in Section 6.7.
- *The database is developed to accommodate various reporting formats.* Information may be submitted in many ways and formats, including web-based reporting, electronic transfer and digital format, computerized reports, and hard copy reporting forms. Medical information may be reported in text format or in ICD code.
- *Record-matching procedures are used since data are collected from multiple sources and existing databases.* Case report information is extracted from administrative databases (e.g., hospital discharge data set, Medicaid data, vital records) and from existing databases within a facility (e.g., laboratories, specialty clinics, prenatal diagnostic centers).

6.6.2 Approach to Passive Case Ascertainment

Passive case ascertainment is based on case reports submitted to the program by data reporting sources. Reporting sources may include mandatory hospital reporting and physician reporting and/or administrative databases (e.g., hospital discharge data set, Medicaid data, vital records). Completeness in the surveillance system is improved by using multiple data sources, especially when data sources are selected to fill a gap in case identification (e.g., fetal death certificates, pathology and autopsy reports). Customized reporting forms may be used, or a program may elect to use other methods for receiving case reports. All legal or legislative issues that govern program operations must be taken into account.

Essential program activities for passive case ascertainment include those listed below.

- **Establish** the type and scope of passive case ascertainment that defines program operations, including whether surveillance includes fetal deaths. Some programs have limited disease reporting guidelines and a smaller set of data sources that are required to report. Some programs may have more liberal disease reporting guidelines but, due to limited resources, have to limit the scope of program operations. Generally, programs that use multiple data sources will have more complete case ascertainment than those that use only one or two data sources. If programs use the birth certificate as a data source for case reports, they should use another data source for case identification.
- **Identify** the case identification data sources. These include birth and major pediatric hospitals. If fetal death is an outcome that is ascertained, it is important to use the fetal death certificate, and possibly cytogenetics laboratories, as a source of case identification. See the list of data sources in Section 6.7.
- **Define** case reporting requirements precisely for each data source. This includes identifying the required or minimum data variables that should be reported. Some data sources will only report the required minimum data variables, while others, like an administrative database, may be able to furnish the program with additional pieces of information. Refer to Chapter 4 on Data Variables.
- **Develop** data reporting methods and procedures for each data source, including data format, timeliness, or reporting schedules. When possible, encourage electronic or web-based reporting. Data sources are usually more consistent in reporting when the burden of submitting the case report is minimized.
- **Develop** record linkage capability. It is important not only to accommodate multiple case reporting formats, but also to use the efficiencies of technology in processing case reports from administrative and existing databases and linking them to case records in the program's database.
- **Develop** procedures for abstracting information from medical records. This includes using the NBDPN Abstractor' Instructions (see Chapter 3 on Case Definition, Appendix 3.1), assigning disease codes, recording other pertinent information, and entering data into the database. Passive case ascertainment programs should review medical records as part of data quality evaluations. Additionally, medical records reviews are often conducted for other focused surveillance functions. For example, some programs that perform statistical monitoring regularly review medical records to confirm a diagnosis. Other times it is important for surveillance staff to review medical records to confirm a diagnosis during a community investigation or when investigating a suspected cluster.

6.6.3 Data Quality Issues in Passive Case Ascertainment

In passive case ascertainment, reporting sources submit case reports to the surveillance system. The reports are accepted without prior confirmation or verification of the information. Therefore, evaluations for quality must be conducted, especially regarding key program outcomes such as completeness, accuracy, and timeliness. Evaluations are often done by reviewing medical records and comparing results between the review and the reported diagnosis. A result of the evaluation process should include quality assurance procedures to identify future problems and methods to track improvement (see also Chapter 7 on Data Quality Management).

- *The quality of a reported diagnosis should be evaluated for accuracy and comprehensiveness.* Errors and differences in reporting will occur, resulting in underreporting, overreporting, and inaccurate reporting. By "rating" the quality of a reported diagnosis, data sources can also be evaluated. Results can be used to adjust quality control and assurance procedures and direct strategic programmatic decisions.
- *The surveillance database should be evaluated for timeliness.* This includes measuring how 'current' the database is in relation to the program's ability to calculate disease rates. Track timeliness of reporting per data source and identify reporting time lags. For example, watch reporting trends to identify whether some calendar months or quarters are problematic for some data sources. Evaluate the surveillance program's data processing procedures for time lags.
- *The disease coding classification system should be evaluated to identify weaknesses, limitations, and problematic codes.* This is especially important for data sources that report cases in ICD code format, which can happen with a data source such as an administrative or existing database. Additionally, although federal coding guidelines are used to direct a hospital or clinic medical records coder in assigning a disease code, the interpretation of medical documentation in the chart is often the reason for a particular code assignment. A good way for a surveillance program to identify potential code problems is to understand some of the conditions that may surface during the newborn time period. For example, a problematic code could be 748.0, choanal atresia or stenosis, since some newborns do experience difficulty in breathing in the first few hours of life. Additionally, situations that might cause a misuse of codes are low birth weight and prematurity (see Chapter 3 on Case Definition, Appendix 3.3). To gain experience in understanding these issues, medical records should be reviewed and results evaluated.
- *The surveillance database should be evaluated for fluctuations in counts and rates of specific diagnoses.* It is possible that an increase in a rate may be due to a change in procedure at a data source. Passive case ascertainment systems must understand that procedures and processes at the data source affect the quality of information in the surveillance database.
- *The surveillance program should develop benchmarks for desired outcome measurements and develop strategies for how to improve the outcome results.* For example, a critical data source that is consistently lagging in reporting might be the focus of a strategic plan to improve timeliness.

6.6.4 Passive Case Ascertainment Surveillance Evaluation

Evaluation of passive case ascertainment surveillance methods should occur at two levels. Both levels directly impact data quality and the program's ability to meet goals and objectives.

One level targets data reporting sources, case identification, and data collection. Examples of areas that should be evaluated are:

- Completeness
- Accuracy
- Timeliness
- Measurability

Programs should develop outcome measurements that will improve data quality and are important to meet program needs and surveillance objectives. See Chapter 7 on Data Quality Management for a more detailed discussion of this topic.

The other level focuses on the surveillance system itself. For a comprehensive approach to evaluating surveillance systems refer to CDC's *Updated Guidelines for Evaluating Surveillance Systems* (Centers for Disease Control and Prevention, 2001).

6.6.5 Tips and Hints in Passive Case Ascertainment

- *Use record linkage to link to the vital record early in the data collection process.* The vital records data source is excellent for establishing a unique case in the database and one that readily identifies the residency of the pregnancy outcome. Additionally, the birth and fetal death certificates fulfill many data collection variables for pregnancy outcome, maternal, and pregnancy information, as well as other statistical information (see Chapter 4 on Data Variables). See the detailed description of the vital records data source in Appendix 6.1.
- *Identify high-quality data sources that report a confirmed diagnosis.* A diagnosis from a high-quality source is an efficient way to improve the accuracy of the database. It also offsets the need to conduct a medical records review for quality evaluations for the specific diagnosis.
- *Ensure cooperation and compliance of data sources as critical factors in passive case ascertainment.* Ease the burden on data sources by encouraging electronic, computerized, and web-based reporting formats for submitting case reports. Offer technical assistance to sites. Many data sources already have the information the surveillance system needs in a database. It is usually easier to sustain consistent, timely, and compliant reporting using a computer program to extract information, rather than expecting staff at the data source to complete a case report.
- *Be flexible when discussing reporting methods and reporting requirements with a data source*. All data sources may not be able to provide all of the desired 'minimum' data fields easily. Evaluate the contribution, including efficiencies, the data source can make to the surveillance system and adjust reporting requirements accordingly. Identify which sources can usually be depended upon to report the majority of demographic information.
- *Be knowledgeable about the information flow through respective hospitals and sites.* Understand medical records content and documentation practices, including how the ICD code classification is used. Passive case ascertainment systems should be proactive in understanding where to go and who to contact to clarify issues when problems arise.

- *Consider conducting 'case finding' at a data source as an alternative to receiving the case report.* Although 'case finding' is not part of the passive surveillance approach, this method should be considered for data sources that may not have an efficient or reliable method of reporting (e.g., outpatient specialty clinics), that may not be able to report in a thorough manner (e.g., autopsy/pathology), or that are not required to report (i.e., voluntary reporting).
- *Communicate with data sources on how birth defects data are used.* Identify the users of the data (the customers) and some of the products produced using surveillance information. Reporting sources like to be recognized for the contributions they make (i.e., reporting cases) and appreciate knowing that the data they provide are used and serve important and valuable purposes.
- *Be active and creative in managing the quality of the database.* It is possible to develop program strategies that not only promote the efficiencies of passive case ascertainment but also improve the important outcome measurements of accuracy, completeness, and timeliness.
- *Be conscious of HIPAA,* especially as this relates to the privacy and security rules that covered entities (i.e., health care facilities) are required to follow. Be knowledgeable about the public health exemptions in HIPAA. Provide reassuring documentation to sites as appropriate (see Chapter 2 on Legislation).

6.7 Data and Case Identification Sources

Information on birth defects cases can be obtained from many sources, each of which has strengths and limitations. Rarely is one source able to provide all of the information necessary to complete a case record. Some, like birth and pediatric hospitals, are ideal for identifying a large number of cases. However, it is important not to overlook data sources like cytogenetics laboratories and specialty outpatient clinics, since they may identify cases previously unknown to a birth defects program. The challenge for birth defects surveillance programs is to evaluate and select data sources that meet the objectives of the program and that can be accessed using available resources. Most data sources can be useful for both active and passive case identification. Differences arise between the two case ascertainment approaches in how the information is gathered and collected. Some data sources are more conducive to active case ascertainment since the only way to access the information is to physically gather it. Some of the major data sources – including vital records, hospital discharge data, hospital unit logs, and genetics clinics – are described in further detail in Appendices 6.1, 6.2, 6.3, and 6.4.

Vital Records (see Appendix 6.1 for detailed description)

- Birth certificates
- Fetal death certificates
- Elective termination reports
- Death certificates

Hospital Information (see Appendix 6.2 for detailed description)

- Hospital discharge data set
- Hospital disease index

Hospital Unit Logs, including (see Appendix 6.3 for detailed description):

- Labor and delivery
- Surgery
- Nursery
- Neonatal Intensive Care Unit (NICU)

Hospital Departments, including:

- Pathology
  Forensic (autopsy) pathology
  Surgical pathology
- Surgery
  Inpatient and outpatient/ambulatory

- Specialty and outpatient clinics
  Obstetrics
  Prenatal
  Perinatology
  Laboratory
  Pediatric medicine

Prenatal and Obstetrics Centers

- Birthing centers
- Obstetrics services
- Planned Parenthood, and other women's care clinics
- Prenatal diagnosis and high-level ultrasound referral sites
- Prenatal genetics counseling services

Specialty Clinics

- Genetics (see Appendix 6.4 for detailed description)
- Oral-facial, craniofacial
- Meningomyelocele
- Cardiology
- Pulmonary/respiratory
- Musculo-skeletal
- Developmental and growth
- Audiology and speech
- Early intervention
- Neuro-developmental
- Ophthalmology

Laboratories

- Cytogenetics
- Prenatal diagnosis
- Metabolic

Physicians

- Pediatricians
- Obstetricians
- Specialists

Health Care Professionals

- Audiologists
- Developmental therapists

Administrative Databases

- Statewide hospital discharge data set (see Appendix 6.2 for detailed description)
- Medicaid data
- HMO data sets

Other Sources of Information

- University-based medical clinics
- Newborn hearing screening program
- Newborn genetic screening program
- Coroners and medical examiners
- Child fatality/mortality review programs
- Public health maternal and child health programs

  Public health clinics, including developmental clinics
- School records

6.8 Sources of Information in a Data Source

In this section we discuss the various sources of information that may be available at a given data source. In Section 6.8.1 we provide a general introduction to the medical record, followed by a more detailed discussion of the various types of documentation within a medical record in Section 6.8.2. Other sources of information discussed include maternal delivery medical records (Section 6.8.3); prenatal medical records (Section 6.8.4); cytogenetic laboratory reports (Section 6.8.5); and autopsy, pathology, and laboratory reports (Section 6.8.6).

6.8.1 Medical Records

By law, all health care facilities are required to maintain some form of medical record on every patient for every service encounter that occurs in the facility. A medical record provides documentation on the course of treatment and progress of the patient at the facility for each admission or service encounter. The medical record may also include information from other health care facilities that may be pertinent to the treatment at that facility. For additional information on the professional practices and standards for medical records and other issues related to health information management, please consult the American Health Information Management Association (http://www.ahima.org).

Medical records differ according to type of health care facility. Medical records maintained by a private health care provider, genetic counseling facility, hospital, or cytogenetics laboratory are likely to differ in the documentation included in the record and how the records are organized. The medical records that birth defects program staff are most likely to work with are those maintained by hospitals, particularly birth and tertiary care pediatric hospitals, and specialty clinics.

The documentation required in a hospital medical record is usually defined by state legislation. Additionally, accreditation organizations maintain standards regarding required documentation (e.g., the Joint Commission on the Accreditation of Healthcare Organizations). Therefore, although medical records from different hospitals in a given state may be compiled and stored differently, the required content is the same. This is useful to know, especially if documentation appears to be deficient.

Since the early 1990s, the 'traditional' medical record has been undergoing change. Today, it is not unusual for the content of medical records to be a combination of hard copy, electronic, and computerized formats. Therefore, surveillance staff should be aware that the hard copy medical record that is traditionally stored and managed by hospital medical records departments may not appear to be 'complete' with respect to documentation. Some documentation may be in computer files or on electronic storage files (e.g., CD-ROM, microfiche, microfilm).

A hospital medical record is generated for every admission and service encounter, and each record follows the guidelines for standard documentation. Some exceptions to this rule may apply in certain pregnancy outcomes. Programs should consult with hospitals and delivery sites for their procedures for outcomes other than live birth. The following are offered as possible scenarios:

- *Live birth.* The infant and mother will each have individual medical records.
- *Live birth with neonatal demise shortly after birth*. The infant *may* have a newborn medical record. However, most useful information will be in the mother's delivery medical record (e.g., if autopsy or cytogenetics laboratory work is done, the results may be placed in the mother's chart).

- *Fetal death.* The fetus *may* have a medical record. However, most of the useful information will be in the mother's delivery medical record (e.g, if autopsy on cytogenics laboratory work is done, the results may be placed in the mother's chart).
- *Elective termination.* A medical record will be created only for the mother. Sometimes the admission at the hospital (or other site) will be as an outpatient.

There are other locations and places where births and other pregnancy outcomes can occur (e.g., in transit, in clinics, at home). Most, but not all, of these sites will generate a delivery medical record at least to fulfill federal and state requirements to complete a vital record. The depth of the information may be incomplete or inconclusive; therefore, additional investigative effort is usually required.

6.8.2 Type of Documentation in the Medical Record

Surveillance staff should be aware of the typical documents found in a medical record. This is true for staff conducting active case finding, as well as for staff conducting a medical records data quality audit for passive case ascertainment. Surveillance staff should consult with individual sites regarding records content requirements and how the documents are stored at the site (i.e., hard copy or computer file). The following are offered as examples:

- *Face sheet.* Contains demographic information, facility-specific information (e.g., medical record number, attending physicians, primary care provider, insurance).
- *History and physical.* Information is gathered and an exam is conducted at admission, at birth, and at various periods during the hospital stay (depending on the length of stay).
- *Discharge summary.* A document that is completed by a physician after a patient leaves the hospital. The summary pertains to a specific hospital stay and includes: admission diagnoses; pertinent medical history prior to the admission and problems, progress, and treatment during the hospital stay; a list of discharge diagnoses; and recommendations for follow-up, such as future visits to specialists and medications to be taken. At some hospitals a discharge summary may not be required for a very brief length of stay (e.g., less than 48 hours). Sometimes discharge summary information is recorded in the progress notes.
- *Consultations.* Specialists such as neurologists, geneticists, or cardiologists also see the patient and provide diagnostic clarification.
- *Progress notes.* Health care providers (e.g., physicians and nurses) document treatment and plans.
- *Diagnostic reports.* Any procedure, whether invasive or non-invasive, requires documentation. This includes: diagnostic tests, laboratory analysis, surgery, cytogenetics, pathology, and autopsy. Sometimes, the final report will not be in the medical record (e.g., it may be in an electronic file or on file in a department of the respective site). Some results will be referred to in the discharge summary, progress notes, or consultation, while others may not be completed for several weeks (e.g., autopsy cytogenetics).

6.8.3 Maternal Delivery Medical Record

In addition to standard documentation required in hospital medical records, the mother's delivery medical record contains unique pieces of information that are important for case ascertainment.

- *Labor and delivery summary.* Many hospitals use a standardized form to record important aspects of the outcome (e.g., time, weight, pregnancy risk factors).
- *Prenatal medical records.* Although the private obstetrician maintains these, some documents may be inserted in the mother's delivery record (or located in other places in the mother's hospital medical record). These include copies of the course of pregnancy management and results of prenatal diagnostic procedures, such as ultrasounds, amniocentesis, and cytogenetics analyses, particularly if a birth defect is detected prenatally.
- *Pathology and laboratory reports.* Pathological analysis is important in the case of fetal demise. Laboratory reports are important when there are suspected infectious disease or toxicology concerns in the mother. For example, there may be concerns about an exposure that could be passed along to the infant through breast milk.
- *Autopsy.* If an autopsy is performed on a fetal demise or neonatal death, the report is often inserted in the mother's medical record or may need to be tracked to the appropriate department.

6.8.4 Prenatal Medical Record

Currently, prenatal care may result in a woman having multiple medical records generated over the course of the pregnancy.

- *Obstetrician's prenatal care medical record.* This record contains documentation of how the pregnancy is managed. The content of this medical record is very similar to a hospital-based medical record; thus, it is important for birth defects surveillance. Sometimes the prenatal care medical record is inserted into the maternal delivery medical record.
  - *Prenatal care forms.* These are often in a standardized format and facilitate complete recording of information (e.g., laboratory work, family history, risk factors, genetic screens, and tests).
  - *Flow charts of care.* Prenatal visits, care and treatment, and patient discussions are documented, although often written by hand.
  - *Diagnostic tests.* The record may contain diagnostic tests, laboratory results, genetic counseling reports, consultations, and referrals to diagnostic centers.

Prenatal diagnosis is growing in importance for birth defects surveillance. There is a long history of chromosomal diagnoses that are detected prenatally through the procedures of amniocentesis and chorionic villus sampling. Many more diagnoses can now be detected through the use of high-level ultrasound. Technology and diagnostic methods will continue to advance in the area of prenatal diagnosis.

- *Referral prenatal diagnostics and diagnosticians.* Referral centers specialize in high-risk pregnancy and have high-level diagnostic capabilities. Depending on the course of a high-risk pregnancy, the referral physician (diagnostician) may assume primary management of the pregnancy and may attend the delivery. However, usually, the referral diagnostic site and diagnostician do not follow the patient throughout the pregnancy. Medical records generated at the referral diagnostic sites may contain pertinent information from the primary obstetrician's

office, including demographic information, index prenatal care history, medical history, risk factors, and reasons for referral. They also contain unique information for case ascertainment. Sometimes the referral prenatal diagnostics are inserted into the obstetrician's prenatal care medical record.

- *Diagnostic and laboratory results.* The medical record includes the results and discussion of the results.
- *Genetic counseling.* Documentation in this report includes significant family history, discussion of prenatal diagnosis, and discussion of prognosis.

6.8.5 Cytogenetic Laboratory Reports

Cytogenetic analysis may be performed at the hospital (in-house) or at freestanding laboratories. Programs are encouraged to use cytogenetic laboratories as data sources that consistently report cases. It is important for birth defects program staff to have some knowledge of basic genetics and the chromosomal terminology they are likely to encounter in medical records. For additional information on cytogenetics terminology (and corresponding abbreviations and symbols) refer to the reference manual, *International System for Human Cytogenetic Nomenclature (ISCN)* (Mitelman, 1995).

The report on cytogenetic findings is created by the lab that did the analysis. The report usually identifies:

- Name of patient
- Date of birth
- Referring facility and/or physician
- Reason for referral (or suspected diagnosis)
- Result/karyotype
- Narrative regarding the analysis

Rarely does the report provide an address for the patient. This presents a challenge for a surveillance program that regularly receives case reports directly from the cytogenetic laboratory, since the laboratory may also perform analyses for patients from several states. Surveillance programs should develop quality control procedures that address this and other challenging issues when working with cytogenetic laboratories. One possible approach is to develop a list of the locations of the referring facilities and/or physicians.

The original report of the result of a cytogenetic analysis (or other test) is the property of the laboratory that performed the analysis. A copy of the report may or may not be sent to the referring facility or physician (or included in the referring facility's medical record). The results may be communicated orally or referenced in a medical record. The surveillance program should develop abstracting procedures for accepting a referenced cytogenetics analysis and for determining when it is necessary to locate the initial source of medical information.

There is a growing trend for hospitals to use out-of-state laboratories. Surveillance programs should investigate the feasibility, including legal authority, of using and contacting out-of-state laboratories.

6.8.6 Autopsy, Pathology, and Laboratory Reports

Pathology laboratories are usually associated with hospitals, while autopsies may be performed in selected hospitals or through coroner's offices. Autopsy and pathology reports are usually placed in the patient's medical record, but the autopsy report may be completed long after death (some states have 45- to 60-day time frames for completion of autopsies). Therefore, the autopsy report may not be filed with the admission medical record; it may be in the outpatient or 'other' section of the record. It is important to note that there are two completion status categories for autopsy findings or reports: provisional and final. Surveillance staff should place the highest level of diagnostic certainty on the final report.

Anatomical pathology laboratories usually produce high-quality case reports due to the exacting nature of the procedures performed during autopsy. An important exception to this is when the specimen is destroyed, macerated, or otherwise compromised, as is the case with many fetal deaths. When this happens, the autopsy and tissue analysis may be of limited value for birth defects case identification. Still, the autopsy report or tissue analysis will often provide the most definitive information on structural defects. Additionally, the type of tissue sample can provide useful information regarding the time frame of the pregnancy. Therefore, it is important to track and examine these reports.

Autopsy and pathology laboratories may have information management systems, manual or computerized, specific to the laboratory. Diagnostic information is usually accessible since these laboratories catalog their findings for forensic investigations, historical and legal archives, case studies, and medical board reviews.

Surveillance programs should understand that there might be varying degrees of quality in autopsy reports. Much depends on the expertise of a given pathologist or coroner, the majority of whom are not fetal and pediatric anatomical pathologists, the experts in this area. In some states these pathologists, and the hospitals or sites where they work, act as referral centers for specialized autopsies. Programs should consult with the respective pathologists and sites to better understand referral patterns in a given state and to evaluate the level of expertise available in this specialized area.

6.9 Infant Risk Factors in Case Identification

A condition that affects an individual's chance of having a particular outcome is called a ***risk factor***. Various maternal and pregnancy exposures and conditions have been associated with an increased risk for birth defects. Birth defects programs can use these risk factors to identify potential cases, either through including their ICD-9 codes on the discharge lists obtained from medical records departments, through reviewing logs for any entries citing these risk factors in addition to birth defects, or through identifying vital records with particular birth weights, etc.

However, even though certain factors are associated with increased birth defects risk, the majority of infants and fetuses with these risk factors will not have a birth defect. Thus, a large number of records will be reviewed that do not turn out to be cases.

Moreover, the list of risk factors that may be used as case-finding sources can become very large. It is possible that a large portion of the potential inclusion population will have at least one of the risk factors used as a case-finding source. Most risk factors only result in a small to moderate increase in birth defect risk, so the majority of records reviewed on this basis will not yield eligible cases. Such risk factor lists are developed from experience, logic, and research. Programs that use risk factors should evaluate the yield in their case identification approach and determine whether using risk factors as case-finding sources is useful to the program over time.

In the short term, the use of risk factors as screens for identifying potential cases of birth defects may be a valuable effort when the program is involved in a concentrated focus on a specific outcome, exposure, medical condition, or cluster investigation.

Surveillance staff may encounter various postnatal complications during the review of data sources and units. This information is most likely found in the infant's medical record, and often in progress summaries. In the situation of a fetal demise or stillbirth, the information is usually found in the maternal delivery chart.

The list below provides some examples of risk factors that may be useful as case-finding sources. Surveillance staff should use pediatric references to become familiar with newborn conditions and evaluate which conditions are appropriate to use for case finding. Passive case ascertainment programs should also evaluate the effectiveness of using risk factors. The majority of the items listed below are identified in data fields on the vital record (birth and fetal death, death certificates) and easily accessible to both active and passive case ascertainment surveillance systems.

Examples of infant risk factors include:

- Infants who weigh less than 2,500 grams (5 lbs, 8 oz) or are < 36 weeks gestational age
- Fetal and neonatal deaths
- Infants with a history of asphyxia at birth (Apgar score at 5 minutes less than 7)
- Infants admitted to neonatal intensive care or special care nurseries
- Multiple births
- Infants with respiratory distress
- Infants with heart murmurs

6.10 References

Centers for Disease Control and Prevention. *CDC Surveillance Update, January 1988.* Atlanta, GA: Centers for Disease Control and Prevention; 1988

Centers for Disease Control and Prevention. Updated guidelines for evaluating surveillance systems: recommendations from the Guidelines Working Group. *MMWR*. 2001;50(No. RR-13).

Edmonds LD. Birth defect surveillance at the state and local level. *Teratology*. 1997;56:5-7.

Joint Commission on Accreditation of Healthcare Organizations (JCAHO). http:/www.jcaho.org

Last JM, ed. *A Dictionary of Epidemiology*. 3rd ed. New York, NY: Oxford University Press; 1995.

Lynberg MC, Edmonds LD. Surveillance of birth defects. In: Halperin W, Baker EL Jr., eds. *Public Health Surveillance*. New York, NY: Van Nostrand Reinhold; 1992:157-171.

Mausner JS, Bahn AK. *Epidemiology: An Introductory Text*. New York, NY: W.B. Saunders Company; 1974.

Mitelman FS, ed. *International System for Human Cytogenetic Nomenclature*. Farmington, CT: Karger Publishers, Inc.; 1995.

Teutsch SM, Churchill RE, eds. *Principles and Practice of Public Health Surveillance*. 2nd ed. New York, NY: Oxford University Press; 2000.

Appendix 6.1

Data Source Described in Detail – Vital Records

Appendix 6.1
Data Source Described in Detail – Vital Records

Source or Site

- Birth certificates
- Fetal death certificates
- Elective termination reports
- Death certificates

Birth, death, and fetal death certificates provide a standardized way of reporting vital events that occur in a politically defined unit, a state. Vital records include facts about an individual and the specific circumstances regarding the reported event. Vital records are particularly important in that they fulfill two significant functions: they provide a mechanism for registering the occurrence of vital events, and they provide a mechanism for collecting demographic, social, and health information regarding the person in a standardized way. Integral to these functions is the fact that they are population based.

Legal or Professional Mandates

Federal law mandates birth and death registration. The lead federal agency is the National Center for Health Statistics (NCHS), Centers for Disease Control and Prevention (CDC). NCHS maintains the national birth and death registration system and is the recipient of vital records data from the states and territories. Recording births and deaths is the responsibility of the individual states and territories. The procedures and regulations regarding the reporting of these vital events are established by the individual states and territories. NCHS provides guidelines and recommendations for standardization of the information collected by birth and death certificates by promulgating standard certificates. Although federal law does not mandate the reporting of fetal deaths, there is an NCHS-recommended standard fetal death certificate. See http://www.cdc.gov/nchs for further information.

Mission or Objective

Provides a population-based statistical database of all births and deaths that occur in the United States.

Scope or Breadth

The birth, death, and fetal death certificates provide for registration of a defined vital event at a point in time. There are established criteria for what constitutes a live birth, but there is evidence to suggest that those criteria are not always followed. Registration of fetal deaths is usually defined on the basis of gestational age, with $\geq$ 20 weeks as the cut-off used by most states. Some states require the reporting of all fetal deaths, regardless of gestational age, and there is recognized underreporting of early fetal deaths.

Operational Structure

- *Data.* NCHS recommends standard data elements on birth and death (and fetal death) certificates. States are required to complete a minimum data set for national reporting and may add other data elements to their certificates. The birth certificate is usually revised and updated every decade. In 2003, the final drafts of a new version of the certificate are being reviewed. Please refer to http://www.cdc.gov/nchs for further information.

- *Certification*. State statutes, regulations, and procedures stipulate who is responsible for certifying a live birth, death, or fetal death. The designated person is required to certify date, time, and place of birth/death as well as other priority areas on the certificate. The completion of death certificates has additional protocols, procedures, and instructions because of the many circumstances that may surround a death.

- *Filing the certificate*. State statutes, regulations, and procedures stipulate time requirements for filing. Although the timing varies among states, the certificate is usually filed with the state registrar's office within 5 to 10 days of the event. Many states now have methods for entering and filing birth certificates electronically. The timing for filing a fetal death certificate depends on state guidelines. Although filing a death certificate is required within a specified time period, it may not be complete at filing, as some data elements may be missing due to autopsy, coroner investigation, or other legal proceeding. These data may or may not be added subsequently and the certificate revised.

- *Unique identification of an individual event.* Each state has a numbering system that uniquely identifies the respective event.

- *Storing the information*. Most states have a centralized database specifically designed to collect, amend, transmit, retrieve, sort, print, and analyze vital records information.

- *Reciprocity.* Agreements with bordering states ensure reporting of life events occurring in neighboring states to the state of residence.

Types of Information Collected

- NCHS and other interested parties have developed a set of standardized data elements or ***minimum data variables*** that are required to be reported, as well as a set of ***recommended data variables*** and recommended standard certificates. Of importance is the unique identifying information per person, per event.
- The birth certificate and fetal death certificate are each divided into two sections: legal and statistical. The ***legal section*** contains the unique identifying information about the person, date, time, place, and type of life event. It is this portion of the certificate that registers the vital event. The information in the legal section is certified, and this is the part of the certificate that is issued to individuals when proof of the life event is required. The ***statistical section*** – labeled "Information for medical and health purposes only" – contains demographic, prenatal care, pregnancy risk factors, and medical conditions of the mother and of the newborn, including congenital anomalies. The statistical part is not released to the public, and many states do not keep the statistical part attached to the legal certificate. The statistical information is usually data entered and maintained in a database.
- The death certificate is a certified legal document, and it is available to authorized individuals in its entirety.

Accessibility and Retrievability

States transmit vital records information to NCHS electronically. State laws and regulations stipulate how the information is made available for other users at the state level. Due to the confidentiality of the information, states protect the medical and health information on vital records from unwarranted or indiscriminate disclosure. Most states have legal safeguards in place to further protect the information.

- The information contained in the birth, death, and fetal death master index computer file is usually available to authorized public health programs. Sometimes confidentiality or security agreements are required.
- Many states copy the legal sections of the hard copy certificate into a permanent electronic storage format (e.g., microfiche, film, CD-ROM). The storage format is cataloged for easy information retrieval.

Strengths as a Data Source

- *Timeliness.* Electronic filing allows information to be available to users as soon as the reports are filed in the state database. This may be as early as 30 days after the event.
- *Population base.* Provides statistical and denominator data.
- *Unique identification.* States assign a unique ID to each person, per vital event.
- *Legality of case report.* State laws require that some information must be certified for all births and deaths. Additional attention to legal procedures is required for death registration.
- *Comprehensiveness as a data source.* Over 97 percent of all births occur in a hospital or birthing facility. Out-of-hospital births are also registered because of the necessity for a child to have a birth certificate. There may be some underreporting of early infant deaths, and there is marked underreporting of fetal deaths at early gestational ages.
- *Existing data set and one that is accessible over time.* There is historical depth to vital records, but there have been major changes in format, content, and coding over time.
- *Record linkage.* Useful in combination with other data for building the case record. The use of unique identifying information permits matching and linking with other data sources. Many states routinely link vital records to each other, for example a death certificate with the birth certificate, providing a linked birth-infant death file.
- *Risk factor screening tool.* Some data elements can be used to identify potential birth defects cases. Examples include: low birth weight, prematurity, low Apgar scores, neonatal death, multiple births.
- *Intervention.* The availability of information in a timely manner is conducive to rapid intervention or investigation.

Weaknesses as a Data Source

- *Data quality.* Much of the medical information on the certificate has been shown not to be reliable.
- *Case ascertainment*. The birth certificate has been shown to underreport birth defects. As shown in Section 6.4, rates from this source are 1.5 percent, compared to 3 to 4 percent for hospital reporting and from using linked data sources.

Liaisons and Partnerships

- *Vital records/registrar's office.* These are staff that are involved in managing the activities involved in filing the certificate. These staff often go to hospitals to train personnel in the procedures and methods of filling out the certificate. Other activities include amending a certificate, maintaining the centralized database, and cross-referencing other vital record certificates.
- *Hospital.* These are staff that are involved in providing information for completing the certificate. Includes medical records services, neonatal nursing, labor and delivery unit staff.

Hints and Tips

- *Neonatal and infant death.* A death certificate is issued upon death for any infant who was live born, regardless of duration of the pregnancy. These individuals will have a birth and a death certificate. There is no distinction in death certificates for 'neonatal' or 'infant' deaths. Many vital records divisions cross reference the birth and death certificate numbers to make sure that a birth certificate is issued if a neonatal or infant death is reported. Sometimes, the facility will overlook filing a birth certificate for an early neonatal death. Sometimes a fetal death certificate is filed as well as a birth certificate and/or a death certificate. In these situations further investigation should occur to determine the actual vital status at birth.
- *The timing for filing birth and death certificates is similar*. However, often the birth certificate is processed by vital records more quickly since many hospitals use the electronic birth certificate. It is important for birth defects programs to be aware of these timing issues if they refer children to services, especially if they refer children based on low birth weight, prematurity, and other severe conditions. Regardless of how quickly a case report is sent to the surveillance program, it is a wise practice to allow a period of time to elapse before referring a child with severe conditions. A time period to consider before referring a child to services is 60 to 90 days past the date of birth.

- *Fetal death certificate*. This certificate is usually issued for any pregnancy that results in a non-live outcome at the end of a pregnancy that is $\geq$ 20 weeks gestational age. What constitutes 'live' is subject to legal definition, and most states have clear guidelines in state statutes for what is considered a 'live birth'. Some states accept any sign of life (e.g., a pulse), regardless of the intent for the delivery (e.g., elective termination). Surveillance systems need to understand the definition of 'live birth' in their state. There may be instances when an Apgar score is a very low number (e.g., 1) at the first minute, and 0 for the fifth minute. Some states might count this as a live birth or a termination, depending on the age of the fetus and intent of the delivery. Some states have guidelines that exclude filing a fetal death certificate if the intent of the pregnancy delivery is for a termination, regardless of the gestational age.
- *Termination reports.* Some states collect statistical information on terminations. Often there is no identifying information; however, a birth defect may be listed as a reason for the termination. In most instances these reports do not have sufficient identifying information to link to an individual. Additionally, although some states require the filing of these reports, compliance is notably poor, such that there is an underreporting of these events and conditions.

References

Mitelman FS, ed. *International System for Human Cytogenetic Nomenclature.* Farmington, CT: Karger Publishers, Inc.; 1995.

National Center for Health Statistics. *Hospitals' and Physicians' Handbook on Birth Registration and Fetal Death Reporting*. DHHS Publication No. (PHS) 87-1107. Hyattsville, MD: US Department of Health and Human Services, Public Health Service, National Center for Health Statistics; Oct 1987.

National Center for Health Statistics (NCHS). http://www.cdc.gov/nchs

Appendix 6.2

Data Source Described in Detail – Hospital Data Sets

Appendix 6.2
Data Source Described in Detail – Hospital Data Sets

Source or Site

- Hospital discharge data set
- Hospital admissions reporting system
- Hospital disease index

Discharge information is collected by the data source in a standardized format on individuals admitted for hospital-based services. This usually includes inpatient stays and outpatient surgery but may also include services performed in outpatient hospital clinics and emergency rooms.

Legal or Professional Mandates

- *Federal law*. The Health Information Portability and Accountability Act of 1996 (HIPAA) legislation defines electronic health care transactions, health information privacy and security standards, electronic signature codes, transaction standards and code sets, and unique health identifiers.

- *Other professional mandates* dovetail with federal requirements (e.g., Joint Commission on the Accreditation of Healthcare Organizations, American Hospital Association).

Mission or Objectives

Discharge data are collected for a wide range of possible uses. These include population-mix studies, market share analysis, hospital charges comparisons, length-of-stay studies, disease-specific and clinical information-specific case volumes, health care delivery access analysis, and crude and severity-adjusted death rate analysis. Discharge data are also used indirectly for financial analysis and billing.

Scope or Breadth

These data result from ongoing data collection and include all inpatient encounters. Some hospital data sets may also include outpatient encounters. The age of population served is defined by the mission of the site (e.g., a children's hospital may serve patients up to age 20 years). A discharge data set may consist of information from one hospital or may be a large statewide discharge data set of all hospitals. A record is created for each defined admission for hospital service. Discharge data sets are defined by a period of time (e.g., year) and are maintained so that they can be accessed over time.

Operational Structure

Information for the data set is collected from many places in the hospital, incorporated into the individual's medical record, and compiled in a standardized format. Health information management or medical records departments are responsible for processing the information that results in the data record for each patient encounter and in ensuring that the medical record contains the required documentation (content).

Type of Information Collected

Information included in this type of data set usually does not include patient names or Social Security numbers. The data elements collected, however, can lead one to a specific medical record. These data sets usually include: hospital identifier, patient medical record number, admission and discharge dates, patient type, patient date of birth, patient gender, patient's residential location (e.g., zip code, county), insurance source, charges, physician type, diagnosis and procedure codes in ICD format, and length of stay. Other information may be collected depending on the objectives of the data set.

Accessibility and Retrievability

Hospital discharge data sets are computerized and are used to generate routine reports and to respond to ad hoc queries. Some hospitals submit their discharge data to a larger organization that collects data from each hospital and compiles the information into a single statewide hospital discharge data set.

Strengths as a Data Source/Site

- *Existing database.* Data are easily accessible, retrievable, and available in a computerized format.
- *Specific information.* Specific data fields can be identified and extracted from the data base.
- *Cross-referencing.* Available data fields provide information that can be used to locate the medical record.
- *Disease classification system.* Information on discharge diagnoses and procedures is collected in a coded and standardized format, currently ICD-9-CM.
- *Timeliness.* Data are usually available rapidly, within 6 months of discharge. Internet technology has increased accessibility and improved timeliness of data from this source for some states.
- *Consistency of the data set.* Data fields are filled in as required for billing and for federal reimbursements.
- *Follow-up.* Hospitals have unique medical record numbers for patients, facilitating tracking and monitoring of cases.
- *Screening tool.* Specific data fields, especially ICD-9-CM disease and procedure codes, can be selected for further investigation.

Weakness as a Data Source/Site

- *Discharge set bias.* The discharge data set is an administrative database. Information is collected and compiled using procedures that suit a particular health facility or meet other legal requirements. It is a services-, planning-, and financial-based data set.
- *Population base.* The service area and patient population for most hospitals are not well defined. Therefore, the relationship of the hospital's patients to a larger group of persons is difficult to quantify.
- *Disease classification system.* Some disease categories and codes for birth defects are not specific and are limited in scope.
- *Accuracy and clarity of diagnosis.* Federal and professional standards are used to govern interpreting medical record documentation, which includes identifying a diagnosis and assigning a representative disease code. Suspected and rule-out conditions may be coded as a final

diagnosis at discharge, leading to overreporting. A diagnosis may not be recorded for many reasons. Underreporting may occur if not all of the diagnoses documented in the patient's medical record are coded.

- *Personal identifiers.* Externally recognizable personal identifiers usually are not available. Data elements can be used to locate medical records. Some states have adopted legislation to permit the reporting of identifying information directly in the discharge data set for specific reportable conditions (e.g., Colorado adopted regulations to permit named reporting from hospital discharge data).
- *Maternal information.* Information on the mother is not recorded on the discharge data record for a newborn infant or child.
- *No medical record is generated.* In some circumstances a medical record is not created. For stillbirths and even some neonatal deaths, a medical record may not be created for the infant. Information pertaining to the delivery outcome, including autopsy and laboratory reports, will be in the mother's delivery medical record. However, the mother's chart cannot be coded to reflect an infant's medical conditions. Therefore, in these circumstances a birth defect diagnosis will be missed. Surveillance staff should use other data sources, such as the vital record, to identify a case where a medical record might not be created.

Liaisons and Partnerships

- *Data processing unit.* Hospital staff in a data processing unit manage the computerized information that is collected from various departments in the hospital. These persons can assist surveillance staff by accessing birth defects information that is stored in computer format.
- *State hospital associations.* Some state hospital associations may serve the function of producing the statewide hospital discharge data set. They have a vested interest in providing customer service to a hospital by compiling aggregate statewide hospital data. Often these associations are also actively involved with the major users of the discharge data set (e.g., health departments, epidemiology programs, health planners).
- *Health information management and medical records departments.* The hospital's medical records staff are responsible for managing the information contained within a medical record. In addition to assembling the medical record and ensuring that it contains the required documentation, skilled personnel – coders – assign the disease classification codes and abstract pertinent information for administrative purposes (e.g., billing and the discharge data set). Since surveillance staff often use the disease classification codes to identify cases, it is helpful to maintain open communication with medical records departments regarding questions about hospital coding rules and other issues that might affect data quality.

Additional Comments

The hospital discharge data set is facing significant changes due to evolving federal regulations, including HIPAA and the conversion of the disease classification from the ICD-9-CM system to ICD-10-CM. HIPAA requirements address electronic transmission of data, standard data elements, and privacy and security issues. ICD-10 is a larger and more complex disease classification system, one that will affect the general taxonomy used for coding purposes.

References

American Hospital Association (AHA). http://www.hospitalconnect.com/

Health Information Portability and Accountability Act of 1996.

Joint Commission on Accreditation of Healthcare Organizations (JCAHO). http://www.jcaho.org/

Appendix 6.3

Data Source Described in Detail – Hospital and Patient Services Logs

Appendix 6.3
Data Source Described in Detail – Hospital and Patient Services Logs

Source or Site

- Hospital unit logs
- Patient services logs (in non-hospital settings)

Hospital units operate within a hospital or clinic and serve specific operational functions. Traditional units relevant to birth defects case ascertainment include Neonatal Intensive Care, Critical Cardiac Care, Labor and Delivery, and the Newborn Nursery. In some hospitals, units are their own departments, like Pathology and Surgery. A ***unit log*** is the documentation that provides information in general terms on the patients who used (or were admitted to) the unit.

Legal or Professional Mandates

- *Legal – state statute*. Hospital-based unit logs are operated in accordance with hospital licensing and accreditation.
- *Legal – state statute*. Non-hospital-based unit logs (e.g., birthing centers, prenatal diagnosis referral centers, genetics clinics), are usually operated in accordance with licensing guidelines.

Mission or Objective

Determined by site. Logs are used to record specific events or health system encounters in a particular hospital department or facility setting. Logs may also account for equipment use. The log represents an inventory of events or activities.

Scope or Breadth

Logs are point-in-time accounts of events. The unit log accounts for each entry or use of services into the specific area. Most logs identify an entrance time, and an exit time, as well as other information specific to unit requirements.

Operational Structure

Determined by site. Logs are designed to be read easily and to provide sufficient information to establish why the patient was in the unit or department.

Type of Information Collected

Determined by the site. Generally, logs are used by surveillance programs as a case identification screening tool. Most logs provide enough cross-referencing information to support follow-through or tracking. This includes name, date of birth, medical record or other identification number, and current date and time. Additionally, information is collected specific to the purpose of the encounter. Examples include:

- *Labor and delivery log.* Prenatal information, maternal risk issues, prenatal diagnosis, and event or outcome measurements.
- *Neonatal Intensive Care Unit (NICU) log*. Event/outcome measurements, perinatal medical issues, diagnosis, other risk factors.
- *Surgery log.* Preoperative diagnosis, possible risk factors.
- *Prenatal diagnostic center log.* Prenatal information, referring physician, referring diagnosis, procedure, medical risk factors.

Accessibility and Retrievability

Logs are used as management tools within individual facility units. Therefore, information is gathered for and used by the unit and, possibly, by the facility. While some information may be collected and entered into a database, most logs consist of paper copy record books or reports.

Strengths as a Data Source

- *Timeliness.* Information is recorded in real time, as events occur. Rapid identification of potential cases is possible.
- *Consistency in recording information.* The population base is well defined for each particular unit since each service encounter is recorded. For example, if a surgical procedure was performed at the site, a surgical log will record the episode.
- *Case identification screening tool.* Generally, enough information is recorded so that surveillance staff can identify potential cases for further investigation.

Weaknesses as a Data Source

- *Effort to retrieve the information.* Generally, logs are kept in hard copy format and are based on a handwritten recording of events. Review of the information can be effort intensive.
- *Accuracy and clarity of clinical information.* Information recorded may be inaccurate or incomplete with respect to diagnoses or medical conditions. For example, a prenatal ultrasound log may state 'referred for cardiac irregularity'.
- *Documentation in the log.* Information recorded on a log may be of limited use for case identification. Sites establish criteria for log documentation to meet internal or ward management objectives, not for disease coding. As such, the information is most relevant for immediate patient management rather than as a tool in medical diagnosis and treatment.
- *Different logs within the data source* may provide conflicting information on the same patient. Surveillance staff should develop management tools to keep track of information recorded from different logs.

Liaisons and Partnerships

- *Unit staff.* These persons are usually front-line staff who work in the unit and have a use for the information that is recorded.

- *Office staff*. These are the persons at the unit who are usually responsible for compiling statistics for the unit and who monitor occupancy. They may be able to assist the surveillance staff in identifying efficient ways to access log information. For example, they may be able to generate a computer listing of the log or provide a photocopy of the log sheet.

Issues to Consider

Surveillance program time and efficiency issues. Unit logs usually require surveillance staff to spend time identifying potential cases on the log and following up by reviewing medical records. Case identification screening criteria and the quality of information included in a log are significant factors to consider when evaluating the amount of time spent on finding cases using this source. Inefficiencies result when follow-up medical records reviews result in too many non-cases. Time and effort evaluations should be conducted for the case identification processes involved in using unit logs.

Unit logs serve as a management tool for individual components of a facility. Therefore, a potential birth defects case may show up on multiple logs. It is useful to compare the information recorded at each unit within the data source and to develop a surveillance management tool that tracks case-finding activity. Such a tool will minimize staff time spent requesting and reviewing a medical record multiple times.

References

None.

Appendix 6.4

Data Source Described in Detail – Genetic Services

Appendix 6.4
Data Source Described in Detail – Genetic Services

Source or Site

- Regional/state genetics networks
- Hospital-based genetics clinics
- University-based genetics clinics
- Provider-based genetics clinics

Geneticists and dysmorphologists are skilled at evaluating a constellation of findings, providing differential diagnoses, and determining the definite medical condition. They use diagnostic procedures such as chromosomal analysis and genetic testing, as well as drawing from their personal experiences and extensive literature in evaluating a patient.

The information from this data source is of high quality.

Legal or Professional Mandates

- *Legal.* State statutes for hospital-based clinics. These are operated in accordance with hospital licensing and accreditation.
- *Legal contract.* Specified in individual contracts or collaborative agreements.
- *Professional.* Certification and professional credential as required.

Mission or Objective

Genetic diagnostic and counseling services, therapeutic management of genetic diseases.

Scope or Breadth

Clinics may include prenatal, pediatric, and/or general population. Some may be specialized by disease category (e.g., Down syndrome, cystic fibrosis). Some providers include diagnostic and research laboratories, clinical research centers, and off-site clinics.

Operational Structure

Genetics clinics may be set up as a referral site (i.e., to provide a diagnosis back to the referring physician), for services (i.e., for ongoing treatment and consultation), or for research or study (i.e., database).

Type of Information Collected

Depends on the focus of the encounter (i.e., prenatal, pediatric, and counseling). As a rule, genetics clinics collect a core set of information for each patient, including demographic data and family medical history. A detailed physical exam and diagnosis, if known, as well as a case summary, is also usually available.

Copies of outpatient diagnostic tests and procedures may also be found. Clinics may use multiple disease classification systems depending on the diagnosis (e.g., ICD-9-CM, ISCN or *International System for Human Cytogenetic Nomenclature*, [Mitelman, 1995]) and/or use proprietary coding systems (e.g., POSSUM, Mendelian Inheritance in Man). Clinic charts may also include letters and notes from other physicians, results of research studies, or diagnostic testing that borders on research.

Accessibility and Retrievability

Usually the medical charts for clients/patients are available at the clinic site for review and abstraction. Many clinics collect information in database format for insurance purposes, clinic needs, and network-wide data collection. Due to the nature of the information gathered, the data often are retained permanently. However, state statutes should be consulted for statute of limitations for health information.

Strengths as a Data Source

- *Accuracy*. High quality. The status of a diagnosis is qualified (i.e., the definite, rule out, possible). Although some patients never get a definitive diagnosis, the differential diagnosis is usually provided.
- *Level of detail*. High quality. Specific information on syndromes (identification and description of dysmorphic features) and chromosomal anomalies is often provided.
- *Case identification.* Specialty clinics, like those for genetics, are important outpatient data sources. Previously unknown cases may be identified for the surveillance program.
- *Case identification or screening.* This is a useful source for prenatal diagnosis cases. Clinics may provide diagnosis and/or genetic counseling services.
- *Retrievability*. Most pertinent information is entered into an electronic file (i.e., a database). This facilitates requesting specific pieces of information that can be extracted in electronic format.

Weaknesses as a Data Source

- *Population base.* May not be well defined.
- *Incomplete information.* Nature of the clinic business or the clinic encounter determines whether the complete diagnostic picture is available (i.e., the case may be referred for cytogenetics laboratory confirmation only).
- *Timeliness of diagnosis.* Some diagnoses are not confirmed until multiple diagnostic procedures have been conducted. Some syndromes take a long time to be diagnosed definitively.
- *Follow-up*. Often a case is referred for consultation and is lost to future tracking. This is important if the diagnosis is reported to the surveillance program as possible or rule out and is in the continuing or discovery phase.

Liaisons and Partnerships

- *Genetic counselors.* Clinics are often staffed by genetic counselors who contribute documentation concerning a patient's evaluation. They are often accessible to surveillance staff if a medical records review or other follow-up is needed.
- *Database managers and other office administrators at clinic sites.* Clinical information is often abstracted from documentation in the medical record for billing, research, or other clinic use. These persons can assist the surveillance staff in identifying efficient reporting and case identification methods.
- *Network system managers.* Regional genetics information may be collected and compiled in a database. Like hospital discharge data, regional genetics information is collected from participating clinics in a standardized format and compiled in a centralized format. Surveillance staff can utilize the efficiency of accessing a centralized database and bypass having to collect the case reports from individual clinics. Of importance is the fact that data from these sources are unlikely to include personal identifiers

Issues to Consider

- *Scope of information collected*. Genetics clinics may collect information and provide a diagnosis that extends beyond the types of defects included in a birth defects surveillance system. Passive case ascertainment systems should be precise in specifying the diagnoses that are included in the program's case definition and which are reportable. Active case ascertainment programs could improve efficiencies by developing a more precise list of diagnoses and medical conditions that can be used to screen for potential birth defects cases in the database or log of the clinic.
- *Confidentiality issues.* Genetics information may be protected by additional federal or state statutes. The surveillance system should research applicable legislation, and if necessary, strengthen security procedures and processes in the surveillance system.

References

McKusick VA. *Mendelian Inheritance in Man (MIM) Catalogs of Human Genes and Genetic Disorders*. 12th ed. Baltimore, MD: Johns Hopkins University Press; 1998.

Mitelman FS, ed. *International System for Human Cytogenetic Nomenclature (ISCN)*. Farmington, CT: S. Karger Publishers, Inc.; 1995.

Possum, Murdoch Children's Research Institute at the Royal Children's Hospital. CD-ROM. http://www.possum.net.au/about.htm