Chapter 1

The Whys and Hows of Birth Defects Surveillance – Using Data

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1.1 Introduction

The ultimate value of any public health surveillance program lies in the ways in which the data collected are used to improve the health of the public. State birth defects surveillance programs are no exception; they exist to improve public health. Every program must have clear goals and objectives that drive how their surveillance data are used toward improving public health. Public input through partnerships with state agencies and organizations and the effective utilization of advisory committees are essential to establishing and revising program objectives and ensuring that the resources exist to meet them.

The purposes and objectives established by state birth defects surveillance programs are constantly evolving. Some objectives are traditional, such as those having to do with the epidemiologic purposes of surveillance; others have emerged more recently, serving to broaden the scope of surveillance programs. Birth defects surveillance programs increasingly use data for services planning and evaluation, for development and evaluation of prevention strategies, to inform parents of children with birth defects about available services, for studies of the societal impact of birth defects, for referral of families to needed services and resources, and for clinical research studies. The consistent theme among these emerging data uses is how birth defects surveillance may benefit other programs in the quest to improve the public's health. In the face of fluctuating resources for public health and obstacles resulting from concerns about confidentiality of health records, the need to incorporate public input in planning and priority-setting has never been greater. This chapter will attempt to address some of the issues in the forefront as we plan for the future of birth defects surveillance.

In the remainder of this chapter we present the rationale for conducting birth defects surveillance (Section 1.2), key steps in establishing a state-based birth defects surveillance program (Section 1.3), and some important uses for birth defects surveillance data (Section 1.4). References cited in this chapter may be found in Section 1.5.

1.2 Rationale

When contemplating initiating or enhancing a birth defects surveillance program, a number of questions come to mind:

- What is the rationale for conducting birth defects surveillance?
- Why is birth defects surveillance important?
- How do birth defects surveillance data benefit other programs?
- What are the barriers to collection and full utilization of birth defects surveillance data?

In this chapter, we provide answers to these questions, which may help you advocate for and prepare to launch or expand a birth defects surveillance program in your area.

1.2.1 What is the rationale for conducting birth defects surveillance?

CDC defines public health surveillance as:

> *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. The final link of the surveillance chain is the application of these data to prevention and control. A surveillance system includes a functional capacity for data collection, analysis, and dissemination linked to public health programs (Centers for Disease Control, 1988).*

It is clear from this definition that a birth defects surveillance program must establish goals and objectives for how data are to be collected, analyzed, disseminated, and used. It is through the latter (i.e., data use) that the efforts from the former are translated into public health action and health improvement. Thus, using data to meet a program's objectives is the most important aspect of any public health surveillance program; merely collecting data is not enough. How data are being used is also what programs tout when they need to showcase their activities to agency officials and legislators.

Because of the essential relationship of the ultimate uses of data to the design and conduct of birth defects surveillance, we begin these guidelines with a consideration of fundamental data-related issues, considering not only the rationale for birth defects surveillance but the key steps for establishing state-based birth defects surveillance programs, followed by a discussion of the use of surveillance data for improvement of the public's health. Every surveillance program should have a plan for data utilization that incorporates public input on all phases of the process – from data development, through data collection, to data dissemination to the public. Suggestions for developing a data utilization plan are presented in Section 1.2 below.

1.2.2 Why is birth defects surveillance important?

States have many reasons for conducting birth defects surveillance. The value of birth defects surveillance lies in how the data are collected and how they are used, with respect to the goals of the program. All programs should establish goals and objectives, which make it clear that the ultimate rationale for conducting public health surveillance is to have data that can be used to improve the health of the public. Reporting the data certainly qualifies as "using the data," yet this should never be considered sufficient as it fails to meet the definition of public health surveillance cited above.

The objectives of state birth defects surveillance programs have evolved over the past 40 years. Lynberg and Edmonds (1992) assessed the objectives that had been established by surveillance programs by the early 1990s. Table 1.1 organizes these objectives under broad purposes originally suggested by Reed and Meaney (1988) with some slight modifications. A review of the table highlights the potentially broad mission of birth defects surveillance, providing state programs with a way of assessing how they are utilizing data currently and possible new uses.

Table 1.1 Purposes and Objectives of Birth Defects Surveillance

Purposes	Objectives
Epidemiologic	Develop timely baseline birth defects rates
	Monitor trends and relationships to environmental factors
	Perform cluster investigations
	Provide basis for ecologic and etiologic studies
Planning/Prevention	Provide data for services planning
	Provide basis for prevention strategies
	Evaluate efficacy of preventive services
Educational/Social	Inform public about public health importance
	Inform parents about resources and care facilities
	Provide data for studies of economic impact
	Provide data for follow-up studies of long-term effects
Healthcare and human services	Refer children to services and resources
	Evaluate services utilization
Clinical	Provide basis for clinical research

Adapted from Lynberg and Edmonds (1992) and Reed and Meaney (1988) with modifications.

1.2.3 How do birth defects surveillance data benefit other programs?

The benefits of birth defects surveillance data to human service programs include: identifying children in need of services to ensure that they and their families are referred appropriately; evaluating service utilization by children with birth defects and their families; and planning the location of services for particular conditions in areas of highest frequency. An important use of surveillance data is monitoring birth defects trends following the initiation of prevention programs in order to evaluate their effectiveness.

One of the public health benefits of the computer age is enhanced capacity for record linkage. Record linkage using public health data has a longer history than most people realize, beginning in the 1950s with the availability of computers in university settings. Pioneering investigators like Harold B. Newcombe (1962) recognized the utility of linking vital records data in studying human populations. The potential now exists for extensive computerized record linkage in birth defects surveillance programs, allowing for the tracking of children with a health-related condition from the point of identification through access to services. Many computer-based systems already exist for documenting health care delivery, including diagnostic and procedure codes. Birth defects surveillance records have been linked to many other public health program databases. These include, for example, newborn screening to conduct epidemiologic studies, special education data to predict the need for services for children with mental retardation, and early intervention program data to assess the overlap and utility of a birth defects surveillance program as a "child find" resource.

In the final section of this chapter we describe a number of applications of these approaches that can serve as models for states developing birth defects surveillance programs, as well as for programs considering expansion of the current uses of their data. To date, the potential for applications of these types exceeds available resources to support them and to overcome some of the obstacles discussed immediately below.

1.2.4 What are the barriers to collection and full utilization of birth defects surveillance data?

While improved methods and technological advances have increased our ability to collect data, there have been intensified efforts to protect the confidentiality of records and the information they contain. Many birth defects surveillance programs – based both in health departments and in other institutions such as universities – have encountered increasing concerns and pressures as a result of Health Insurance Portability and Accountability Act (HIPAA) regulations and issues surrounding their interpretation and implementation. A variety of HIPAA-related issues are discussed in Chapter 2 of these guidelines. Even though the HIPAA regulations include public health exclusions regarding access to records without a patient's consent, programs have seen increased awareness and concerns on the part of hospitals and clinics reporting cases and data. These concerns are magnified when a surveillance program attempts to expand data usage through linkage to other databases covered under HIPAA regulations.

Prior to HIPAA, concerns often surfaced about data sharing among officials in different programs within the same state agency or among programs located in different agencies. Such concerns were usually due to program-specific regulations about data use. Program regulations frequently impede attempts to link records between case-finding databases and service-delivery databases. As a result, attempts to meet the very reasonable public health goal of ensuring access to services by those in need may be thwarted. Thus, programs are strongly urged to consider strategies for surmounting these problems well in advance of undertaking data collection and record linkage.

1.3 Synopsis of Key Steps in Establishing State-Based Birth Defects Surveillance Programs

In this section we outline some of the key steps in establishing a birth defects surveillance program. These include:

- Defining the objectives and purposes of the program
- Considering legal issues
- Engaging external support
- Leveraging resources
- Considering record linkage

Time devoted upfront to serious consideration of these issues will be well spent and will ensure that the resultant program is established on a firm footing.

1.3.1 Defining the Objectives and Purposes of the Program

The success of a birth defects surveillance program is likely to be highly dependent on the host agency's commitment and support. Without programmatic commitment and resource support at the agency level, programs are apt to languish in circumstances that do not allow much beyond the collection and reporting of data. In these situations, using data in ways other than the calculation of rates and their dissemination in reports is usually not possible. Programs committed to expanding how birth defects surveillance data are used must establish programmatic objectives and demonstrate to agency officials how the data could be used. This involves prioritizing what uses would be of greatest utility in terms of meeting agency goals and objectives, demonstrating (or "marketing") to the agency how beneficial these data uses could be, and working to achieve commitment of additional agency resources.

Another strategy for increasing support from the agency in which the surveillance program resides is to gather support from other intra-agency programs and from external agencies that could benefit from the use of birth defects surveillance data to meet their own programmatic goals. Often other programs and agencies, given enough information about birth defects surveillance and the objectives of the program, will see potential uses of the data that are beyond the current scope of the surveillance program.

There has been an increase in intra-agency collaboration during the last ten years through the availability of federal support for data linkage and integration. A prime example of data collaboration would be linking birth defects surveillance databases with Children with Special Health Care Needs (CSHCN) program databases that collect data on program enrollment and services. These linked data sets could then be used to evaluate the rates at which this long-term maternal and child health program is utilized. Such applications have been accomplished in some states through grant support from the Maternal and Child Health Bureau (MCHB) of the Health Resources and Services Administration (HRSA) and through cooperative agreements with the Centers for Disease Control and Prevention (CDC). Interagency collaboration in linking birth defects surveillance program databases with services databases (such as those for early intervention programs or developmental disabilities) have begun in a few states. The benefits to be gained in this way – i.e., by utilizing birth defects surveillance data as a means of identifying children eligible for special programs, such as early intervention – is clearly a “selling point” that can lead to additional resource allocation, either from within the host agency for the birth defects program or from an external agency in need of the data.

Most birth defects surveillance programs experience cyclical problems with availability of state resources, leading them to define precisely what they can and cannot do given the resources available to them. While it is certainly necessary for programs to realistically budget their resources to ensure continued viability, programs also need to engage both intra-agency and interagency support for their goals and objectives as a means to maintain and expand a surveillance program. At a minimum, programs should allocate personnel time to educate officials of their own agency and other agencies about birth defects surveillance and its importance and potential uses in the public health field.

1.3.2 Considering Legal Issues

To the extent possible, programs should consider the inclusion of references to data use in the legislation that authorizes birth defects surveillance. Given the relative ease with which rules – as compared to laws – can be changed, it is generally desirable to make references to potential data uses for surveillance data more general in the statute and more specific in the rules. Rules and regulations that refer to the authorizing statutes are the obvious choice as to where best to specify detailed uses to which surveillance data will be put. Relevant issues and legal considerations are discussed extensively in Chapter 2 of these guidelines.

1.3.3 Engaging External Support

Beyond seeking intra-agency and interagency support for a new surveillance program or for expansion of an existing surveillance program, program staff should also seriously consider means to attract the support of both non-governmental partnering organizations and the public.

Partnering organizations. The importance of building partnerships with organizations such as the local March of Dimes can never be sufficiently stressed. In recent years, the success story of the birth defects surveillance program in North Carolina is arguably without peer. The program has consistently credited the partnership it built with the March of Dimes as a major contributor to its success in garnering additional resources for the program. In Texas, the March of Dimes was also instrumental recently in restoring funds to maintain the Texas Birth Defects Monitoring Division, funds that had not been requested in the budget put forward by the Texas Department of Health. These kinds of partnerships should be entered into with clear and consistent agreement among the players regarding the objectives of the program relative to data usage, prioritization of data uses, and planning toward future applications of the data. In other words, the contribution of organizations such as the March of Dimes can be beneficial from the design of data utilization plans through to the reporting of actual outcomes.

Advisory committees with agency, organizational, and public representation, including political officials, are another means of obtaining input regarding uses of birth defects surveillance data. The available computer technologies such as listservs and webpages decrease the need for face-to-face meetings among interested parties, while increasing the frequency with which information about a program can be communicated and feedback solicited. New ideas about potential uses to which a program’s data can be put and the resources needed to accomplish programmatic activities can be shared with advisory committee members for immediate feedback as to the feasibility of the idea and its potential for success.

Programs should create opportunities for formal input from advisors on a regular basis to ensure the availability of support in times of fiscal crises. Advisory group members’ knowledge of surveillance data collection activities and uses for surveillance data can be critical to securing resources for a program in times when limited resources require justification for program continuation.

Public involvement. Birth defects surveillance programs generally have not engaged consumer and parent participation other than through advisory group representation. Members of the public, including parents of children identified through these programs, are often not well informed about public health surveillance activities. If not already doing so, birth defects surveillance programs should engage both consumers – here defined as adults with birth defects, and parents and caretakers of children with birth defects – in the planning and implementation of any and all programmatic changes. There are a number of advocacy and parent support groups, such as the Spina Bifida Association of America, Family Voices, and the Alliance of Genetic Support Groups, that can play important roles in planning and conducting birth defects surveillance programs.

Programs should embrace the concept of *participatory action research* (PAR) (Whyte, 1991). PAR is a way to obtain public input into programmatic activities from design though dissemination of results. PAR ensures input from the community members most affected by potential data uses. Again, as discussed with respect to advisory group input, computer technology can be immensely beneficial in obtaining feedback on new initiatives and more importantly in soliciting input about programmatic activities from community members.

1.3.4 Leveraging Resources

For birth defects surveillance, as for other public health surveillance programs, the ways in which data are used will influence continued availability of program resources. In the age of evidence-based medicine and increased emphasis on demonstrating program efficacy for continued support, birth defects surveillance programs should work toward expanding data use. Fiscal trends in states suggest that the likely survivors in times of increasingly fewer tax-based resources will be programs that adapt by reinventing themselves in terms of data utilization. While emphasizing the application of surveillance data to improving human services and then evaluating their impact will not ensure the survival of a program, it should increase its chances.

Surveillance programs (particularly those housed in health departments) may be given adequate resources for data collection and management, but often do not have adequate personnel or resources for data analysis beyond simple descriptive reporting. Program managers and staff often use lack of adequate resources as an excuse to minimize the number of new initiatives they undertake, but this may well be a short-sighted approach. We have already discussed the importance of partnerships, advisory groups, and public involvement in increasing the probability of acquiring additional resources. While programs must, realistically, work within the limits of available resources, partnerships with agencies and institutions can represent a means to extend and enhance programmatic achievements. Universities, particularly those with public health training programs or medical schools, will have faculty and trainees potentially interested in birth defects. What a birth defects surveillance program lacks in resources for data analysis and research often can be compensated for through partnerships with interested faculty members willing to direct student theses and dissertations that focus on birth defects. New programs and programs that do not currently have such partnerships should give serious consideration to forming these types of collaborations, which can lead to additional resources through contracts and grants.

1.3.5 Considering Record Linkage

As touched upon in Section 1.3.1, the potential to link records and consolidate information from different databases contributes to the public health applications of surveillance data. For example, data from birth defects surveillance programs can be used to determine whether reported cases of birth defects represent existing cases in other databases, such as records in interdisciplinary clinics and schools with programs to assist children with disabilities. The ability to link records on individuals in more than one database can streamline the treatment and referral processes and help maintain a certain level of fidelity and trust in prevalence data. Record linkage can streamline the research process by consolidating several different databases. Another utility of record linkage is the ability to supply crucial data required for various research efforts. Specifically, the data located in one database can be used to elicit information from a second.

1.4 Uses of Surveillance-based Birth Defects Data

Most US states have implemented birth defects surveillance programs that monitor and disseminate information regarding birth defects. Public health staff and researchers nationwide have used these data in a variety of ways. The actual and potential uses of birth defects data are discussed and exemplified in the following sections. Data from birth defects surveillance programs can be employed to define the magnitude of a problem, to support research, as well as to assess the efficacy of prevention and treatment, playing a key role in the core public health function of *assessment* (Institute of Medicine, 1988).

For convenience, the uses of birth defects surveillance data can be grouped into the following categories:

- Prevalence studies
- Epidemiologic studies
- Mortality assessment
- Needs assessment for services
- Referral to clinics and services
- Program evaluation
- Clinical research

Each of these categories of use will be discussed in further detail below. While comprehensive coverage of works in each of these categories is beyond the scope of this chapter, we have selected published studies that exemplify the kinds of research that can be conducted in each category. Naturally, what an individual program is able to do depends ultimately on its goals and objectives. When programs are faced with limited resources to conduct data analysis and research, collaborations with universities or contractors with epidemiologic expertise can often yield mutually satisfactory results.

1.4.1 Prevalence Studies

A common use of data produced by birth defects surveillance programs is to describe the occurrence (***prevalence at birth***) of the monitored conditions. Such uses of surveillance data include identification of trends in birth defects occurrence, definition and evaluation of clusters of congenital defects, and assessment of the need for resources and interdisciplinary services.

Khoury et al. (1986) is an example of an early study by a state surveillance program that used data in this way. This study was the outcome of a partnership between the state health department-based surveillance program and university-based researchers. Khoury and co-workers used 1984 data collected from the Maryland Birth Defects Reporting and Information System (BDRIS) to determine rates of occurrence and to identify potential trends. The prevalence at birth of "sentinel" defects, as determined from the Maryland BDRIS data, was 52.7 per 10,000 qualified births. Furthermore, trends in the occurrence of several specific birth defects were identified. The study revealed an association of low birth weight and prematurity with birth defects, an association between twinning and the rate of birth defects, racial differences in the prevalence of neural tube defects, and a relationship between Down syndrome and advanced maternal age. The importance of determining prevalence at birth is that the data can be compared with similar data collected from other birth defects monitoring systems to assess differences in rates that may exist among surveillance areas and to direct further research efforts in an attempt to identify the reasons behind the differences.

An example of a more recent prevalence study is one reported by Ethen and Canfield (2002), who investigated the effects of including elective pregnancy terminations, prior to 20-weeks gestational age, on birth defects prevalence. In many surveillance programs, pregnancies ending prior to 20 weeks gestational age, including elective terminations, are not ascertained to be included among reported cases. The researchers concluded that when elective terminations at less than 20 weeks were considered, the prevalence of some congenital defects increased, while others remain unchanged. Specifically, anencephaly, spina bifida, and encephalocele increased substantially, while cleft palate did not change. The underlying assumption is that pregnancies resulting in debilitating or potentially terminal conditions are more likely to be terminated electively than those resulting in less severe or treatable malformations.

These two studies show the potential usefulness of prevalence data to reveal important trends and associations. These types of data often provide the impetus to initiate subsequent research. A consequence of producing birth defects prevalence data is that it frequently opens other avenues of exploration. Quite simply, without basic prevalence data to lead inquiry, many research investigations never would be conceptualized, much less carried out.

1.4.2 Epidemiologic Studies

Cases from birth defects surveillance programs have played key roles in conducting etiologic research in the United States and internationally. Cases from the Metropolitan Atlanta Congenital Defects Program (MACDP) have provided the basis for numerous research studies that have shed light on both the causes (Khoury et al., 1982; Oakley, 1984; Erickson, 1991; Dott et al., 2003) and prevention (Roberts et al., 1995; Olney et al., 2002) of birth defects. Similarly, the California Birth Defects Monitoring Program (CBDMP) has been the source of cases and etiologic research that has resulted in dozens of seminal papers on a variety of specific congenital malformations and their risk factors (Croen et al., 1991; Shaw et al., 1996; Ritz et al., 2002). Other state programs have contributed cases for epidemiologic studies leading to a growing number of multi-state investigations of specific risk factors (for example, Olney et al., 1995). Reference to the annual report of the International Clearinghouse for Birth Defects Monitoring Systems (International Centre for Birth Defects, 2002) demonstrates the large number of studies based on individual surveillance systems and collaborative projects among programs.

An example of an early methodological study, based on surveillance data, is a study by Khoury et al. (1988) that assessed the patterns of maternal residential mobility between conception and delivery. The authors' rationale was that most epidemiologic studies of environmental risk factors are based on maternal residence at the time of delivery. Such an assessment would be invalid, however, in instances where the mother had moved prior to delivery. The researchers examined demographic data for infants born with congenital defects. Both the demographic data as well as the birth defect data were taken from the Maryland BDRIS in 1984. The researchers concluded that, on average, 21% of all mothers whose pregnancies resulted in a child affected by one of the birth defects included in the Maryland BDRIS had moved between conception and delivery. This is important for several reasons. First, it is well understood that the effects of environmental teratogens occur early in embryogenesis; so assessing the influence of environmental exposures must be related temporally to conception. In addition, potential exposures to teratogenic environmental factors could possibly be misrepresented if examined at delivery rather than around the time of conception. Maternal mobility could also skew data regarding geographic clusters of birth defects. This study was made possible because the Maryland BDRIS determines the residence of the mother not only at the time of delivery, but also at the time of conception. This is an important aspect of the Maryland BDRIS that is not common to all birth defects surveillance programs.

Examples of surveillance-based etiologic research of associations between maternal exposures and congenital defects include studies of cigarette smoking and orofacial clefts. Among the earliest research efforts investigating this association was a study by Khoury et al. (1987) using data collected in 1984 from the Maryland BDRIS. A case-control study examined the history of cigarette smoking among mothers of infants with orofacial clefts and a group of control mothers. The researchers concluded that odds ratios for cleft palate (2.39, CI 1.04-5.45) and cleft lip with and without cleft palate (2.56, CI 1.13-5.78) were increased for women who smoked. Furthermore, the researchers identified a dose-response effect. Khoury and his co-workers also took into account possible confounding factors, including race, gender, residence, maternal age, parity, and several pregnancy exposures or complications. None of these affected the results significantly. This is a classic example of how surveillance-based birth defects data can be used to examine etiologic factors through the use of simple epidemiologic techniques. Sometimes the importance of earlier epidemiologic studies is not appreciated when comparing them to more recent research. It is worth noting that the association between maternal cigarette smoking and orofacial clefts has been corroborated through more recent studies using several surveillance-based investigations. The paper by Khoury et al. (1987) has been cited in many contemporary research publications (Shaw et al., 1996; Lieff et al., 1999).

Some states have used surveillance data to look for associations between environmental factors that are known to cause specific birth defect syndromes and other birth defects. For example, maternal alcohol use during pregnancy is a known cause of the fetal alcohol syndrome, but its role in more common, isolated, craniofacial defects is not well understood. A population-based, case-control study of orofacial clefts was conducted in Iowa based on births from 1987-1991 (Munger et al., 1996). Cases were identified by the Iowa Birth Defects Registry and classified as having a cleft lip with or without cleft palate (CLP) or cleft palate only (CP) and as to whether the cleft was isolated or occurred with other birth defects. Controls were selected from normal Iowa births. Maternal alcohol use during pregnancy was classified according to self-reported drinks consumed per month. Compared to women who did not drink alcohol during pregnancy, the relative odds of isolated CLP rose with increasing level of maternal drinking as follows: 1-3 drinks per months, 1.5; 4-10 drinks per month, 3.1; more than 10 drinks per month, 4.7 (chi-square test for trend, $P = 0.003$). Adjustment for maternal smoking, vitamin use, education, and household income did not substantially alter the results. No association was found between alcohol use and isolated cleft palate or clefts in children with multiple birth defects. Based on these data, alcohol use during pregnancy may be a cause of isolated cleft lip with or without cleft palate.

As described, epidemiologic investigation is an important area of research supported by birth defects surveillance data. In the past, this research effort primarily focused on environmental exposures as possible etiologic factors. However, with the recent explosion of molecular genetics and a more thorough understanding of molecular biology, the avenues of epidemiologic investigation have widened significantly. Investigators now have an enhanced ability to examine the contributions of both maternal and fetal genotypes to disease risk. Examination of the interplay between genetic predispositions/susceptibilities and environmental exposures is a growing area of study, with potential major implications with respect to understanding birth defects etiology. This is illustrated by the genetic component of the National Birth Defects Prevention Study, a multicenter case-control study being conducted by CDC and participating state surveillance programs (Yoon et al., 2001; Rasmussen et al., 2002).

Continuing with the study of the association between smoking and clefts, epidemiologic studies have focused on the relationship between certain alleles of a transforming growth factor and maternal cigarette smoking with regard to risk of orofacial clefts. The most promising associations are seen in polymorphisms of the transforming growth factor alpha (TGFα) gene taq1 and maternal cigarette smoke exposure. An example is a study by Hwang et al. (1995), supported by surveillance data, that examined this association. The data on infants born with orofacial clefts were taken from the Maryland BDRIS. The Maryland BDRIS was not only able to supply cases of orofacial clefts, but also information about maternal prenatal behaviors, including maternal smoking during pregnancy. Cases were genotyped and screened for the rare C2 taq1 polymorphism.

The researchers concluded that the C2 genotype, combined with maternal smoking, significantly increased the risk of orofacial clefts. Using data collected through a birth defects surveillance program, they were able to identify a possible interaction between an environmental exposure and a genetic predisposition with respect to risk for orfacial clefts.

Studies like this represent another generation of epidemiologic research. The power of these molecular epidemiologic studies lies in their ability to elicit possible etiologies of birth defects beginning with prevalence data, demographic information, and biologic samples. While the epidemiologic research methods have evolved significantly, the ultimate goal of these studies has remained constant: namely, to identify, define, and associate birth defects with possible etiologic factors. The development and application of molecular genetic methods serve as stepping stones to future research based on surveillance-derived cases.

1.4.3 Assessing Mortality Associated with Birth Defects

A 1995 Texas study assessed survival rates for selected birth defects among babies born between January 1, 1995 and December 31, 1997, by linking two databases: the state's active birth defects registry and the infant death registry (Nembhard et al., 2001). The goal of the study was to determine mortality among cases with various birth defects identified through the birth defects surveillance system by matching those cases against infant death files. Specifically, the researchers found the birth defects with the lowest survival were anencephaly (0%) and trisomy 13 (7.4%), while the birth defects with the highest survival were gastroschisis (92.9%) and trisomy 21 (92.3%). These survival data were only for the first year of life.

Another example of a mortality study is that carried out by Druschel et al. (1996), who examined infant mortality among children with orofacial clefts, comparing their mortality rates to those of children with no congenital malformations. In the absence of malformations in other organ systems (isolated clefts), mortality was not increased among children with orofacial clefts. The study revealed, however, that many children with orofacial clefts have other malformations that increase their risk of death. These findings suggest the need for careful evaluation of possible additional malformations among children with orofacial clefts as these children may be at higher risk of death.

1.4.4 Estimating the Need for Services

Estimating service needs based on birth defects prevalence has significant direct social consequences. Accurately predicting the demand for various interdisciplinary clinics and social and educational services is critical for children born with birth defects. Estimating future service needs allows for capacity building to ensure that necessary resources will be accessible and that appropriate professionals will be available to provide the services.

Brewster et al. (1992) linked demographic and diagnostic data from1980 – 1982 in a birth defects surveillance program database (the Arkansas Reproductive Health Monitoring System) with education databases. The data were first used to estimate the percentages of infants with specific birth defects who were at risk for developmental disabilities and mental retardation. Once prevalence rates were determined, two clinicians estimated the various services that would be needed by children with the various birth defects most likely to contribute to developmental disabilities. This included academic and other services these infants would require as they matured. The researchers estimated that between 32% and 56% of all children in schools who were classified as mentally retarded were also identified by the Arkansas Reproductive Health Monitoring System.

This study showed that recognition of children with mental retardation, who were also identified years earlier as having congenital defects, allowed researchers to refine their estimates of the birth defects that will contribute most significantly to mental retardation in school-aged children. This is useful in improving the ability of health care professionals to predict accurately future needs of the current cohort of newborns with birth defects.

1.4.5 Referral to Services

Information collected as part of birth defects surveillance can be used to refer specific children and their families to appropriate services. Established referral networks serve as a resource for children and their families to learn about available medical services, community programs, and social support. Affected children and their families can be connected with appropriate services in a timely fashion.

Many papers have been written detailing the process of identification and ultimate service referral. One of the first papers on this topic comes from the Maryland BDRIS, where investigators examined the referral of children identified with orofacial clefts through the surveillance program to the Maryland Crippled Children's Service Program in the 1960s (White, 1981). This study examined referral rates to services. A more recent paper on referral and treatment patterns for orofacial clefts comes from Florida, where referral and treatment patterns of live-born Florida infants diagnosed with orofacial clefts identified through the Florida Birth Defects Registry were determined (Williams et al., 2003).

Another example is a paper describing service referrals in Colorado that use birth defects data taken from their birth defects surveillance program (Montgomery and Miller, 2001). The Community Notification and Referral Program (CNRP), operating from within the state's health department, uses birth defects data to link affected infants with an organization that can refer them and their families to agencies and interdisciplinary clinics. In 1998, 259 families were referred for services as a result of being identified through the birth defects registry. There are a number of services to which patients are commonly referred, including developmental screening and evaluation, public health programs, early intervention programs, financial assistance, parenting classes, medical services, recreational programs, and family support groups. Additionally, the effectiveness of this program has been assessed through the use of surveys and questionnaires.

A review of the use of surveillance data relative to provision of early intervention services can be found in a recent paper on identification and referral programs by Farel and colleagues (2003). Having agencies use birth defects data to link patients with appropriate services is a critical data use that has immediate and direct impact on the lives of those affected. Although epidemiologic and laboratory efforts may illuminate etiologies and possible preventive measures for future use, the fact remains that effective therapeutic efforts in the present can significantly improve the lives of persons with birth defects; scientific studies take years to complete and primarily aid future patients. Meanwhile, there are people who require immediate assistance, and service referral is an important mechanism through which they can receive that help.

1.4.6 Program Evaluation

Another use of birth defects surveillance data is program evaluation. Typically, this use is employed subsequent to research efforts, many of which were also based on surveillance data and may represent a baseline from which post-intervention improvement can be measured. Program evaluation is a valuable and desired area of activity with important scientific, academic, social, and policy applications. Program evaluation can focus on different aspects of surveillance program activities, such as case referrals and clinical interventions. First, evaluating a program for service referral can give investigators information on the efficacy of their referral agencies or the appropriateness of the services offered. Second, evaluating clinical intervention studies allows researchers to assess both the effectiveness of the intervention and the validity of their clinical assumptions.

One study involving program evaluation of a clinical intervention using birth defects surveillance data was performed in Nuevo León, México (Martinez de Villarreal et al., 2002). The investigators assessed the effectiveness of a folic acid campaign in reducing the occurrence of neural tube defects. Investigators first developed a base rate for neural tube defects prior to administration of the folic acid and counseling services. An intervention was then initiated that included five mg of folic acid supplementation per week, as well as counseling and social services. After 28 months, the rates of neural tube defects were ascertained again. From the baseline in 1999 (95 cases of neural tube defects), neural tube defects declined by 50% in the next two years (59 cases in 2000, 55 cases in 2001).

This study illustrates the wide range of uses for birth defects surveillance data in evaluation. First, data were used to assess an initial rate of neural tube defects and at the conclusion of the intervention to assess its appropriateness and efficacy. In addition, the study demonstrated the efficacy of folic acid supplementation in reducing the occurrence of neural tube defects and the fact that the methods of administration were clinically appropriate and effective.

In another example of the use of surveillance data in program evaluation, Meyer and Oakley (2000) used data from the North Carolina Birth Defects Monitoring Program to assess the folic acid fortification mandates of the federal government. The results suggested that the decline in the occurrence of neural tube defects was marginal and not the predicted 50% decrease. The authors' recommendation was to increase the folic acid fortification standards on a national level.

1.4.7 Clinical Research

Recently a group of researchers in the United Kingdom carried out surveillance in one Health Region using multiple sources to identify all individuals with specific conditions (Holland et al., 1998; Whittington et al., 2001). The condition the researchers captured that is of greatest relevance to birth defects is Prader-Willi syndrome (PWS). The first step was to conduct population-based surveillance in the Cambridge Health District (eight English counties with a base population of 280,000 individuals) (Whittington et al., 2001). The birth prevalence of PWS was estimated to be 1:22,000 and the mortality rate more than 3% per year. The next step was to carry out population-based clinical research about phenotypic features, including the prevalence of behavioral and health problems in PWS. Clarke et al. (2002) reported the prevalence of compulsive and similar behaviors among individuals with PWS in this population. Butler et al. (2002) presented data on the prevalence of comorbidities in PWS that could contribute to reduced life expectancy for persons with this condition. Most recently Holland et al. (2003) reported on the specific behaviors that comprise the proposed behavioral phenotype in PWS.

Although this work represents a non-traditional method of surveillance compared to state surveillance programs in the United States, it is important in terms of clinical research that has been conducted and the potential for conducting similar work using state-based surveillance data. A major advantage of these clinical studies is that they are population based. Even though all individuals identified through the surveillance work did not participate in the collection of behavioral and health data, the sample of individuals with PWS who participated in the clinical research can be compared to the total population of ascertained individuals to evaluate how representative the sample is of individuals in the Health Region who have PWS. Usually this is not possible using common methods of clinical research.

1.4.8 Using Birth Defects Data in the Future

For the data collection process itself, abstracting methods continue to be refined. Quality assurance procedures and ongoing training, aimed at increasing data accuracy and validity, are being implemented in order to assure a certain level of fidelity and trust in the data collected. Improving and standardizing these procedures are among the objectives of these guidelines.

The future uses of birth defects surveillance data are related to scientific advances in other areas of research. Several developing scientific fields will utilize birth defects data in novel ways. For example, our understanding of molecular biology has developed exponentially. With the successful sequencing of the human genome, the resulting information will provide significant information on genetic factors influencing disease risk. Consequently, these discoveries will be investigated for certain genetic regulatory mechanisms and environmental triggers. Using birth defects surveillance data, investigators will be able to examine possible environmental exposures that are etiologically associated with birth defects in the presence of a particular genetic background. Discoveries of gene-environment interaction will allow researchers to understand etiologic associations. Additionally, the way in which these environmental conditions regulate gene expression will further illuminate these associations.

Future advancements in research supported by birth defects data will benefit from the integration of electronic medical records. Current methods for obtaining birth defects data are laborious. They frequently involve extensive abstraction procedures, reporting cases to the respective health department, entering the abstracts into the database, and categorizing the data. These methods will be streamlined, as medical records and birth defects surveillance systems are maintained electronically. This will have two general effects: first, it will help facilitate the abstraction process by eliminating bulky charts containing information not necessarily applicable to the birth defects surveillance program and, second, it will allow researchers to access these information-rich databases more quickly and efficiently. Furthermore, database search functions will allow researchers to identify cases of interest instantaneously without physically sifting through thousands of reported cases. Ultimately, researchers will be given access to the electronic surveillance database. Using surveillance systems researchers will able to search for cases of interest and refine their cohort by filtering cases by demographics, location, or maternal prenatal behaviors. A study that currently takes weeks to conclude would be completed in the course of several hours.

Researchers continuously find new and exciting uses of the data from birth defects surveillance programs. Given the breakthroughs achieved through earlier studies using surveillance data, the possibilities of future revelations are staggering. In their relatively short existence, birth defects surveillance programs have changed the ways in which professionals view birth defects both clinically and socially. The importance of the impact of birth defects surveillance programs on clinical and public health research cannot be overstated, as such research is revolutionizing the way scientists, clinicians, and health care professionals approach, treat, and manage infants affected by birth defects, while also advancing our understanding of preventive measures.

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