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Assessment and Framework for Population-based Surveillance of Birth Defects in the United States

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Dedication

This dissertation is dedicated to my husband, Alex, and our children, Maia and Dylan.

I also dedicate my work to my dad, Loc Mai (1941-2012), for his sacrifices and steadfast belief that the impossible becomes possible through hard work and persistence.
May he rest in peace.

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ACRONYMS

CDC	Centers for Disease Control and Prevention
CMS	Centers for Medicaid and Medicare Services
EUROCAT	European Surveillance of Congenital Anomalies
ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
NBDPN	National Birth Defects Prevention Network

SUMMARY

Birth defects are a leading cause of infant mortality in the United States (U.S.) and contribute substantially to health care costs and life-long disabilities. They are conditions that: 1) result from a malformation, deformation, or disruption in one or more parts of the body; 2) are present at birth; and 3) have a serious, adverse effect on health, development, or functional ability. The World Health Assembly adopted a resolution calling on member countries to develop and strengthen surveillance systems for birth defects given their impact on infant and child morbidity and mortality.

The objectives of this study are to examine the infrastructure, data collection and utilization of U.S. population-based birth defects surveillance and to assess how these programs are meeting current and emergent needs by using the Centers for Disease Control and Prevention's (CDC) Strategic Framework for Public Health Surveillance in the 21st century. Areas covered in this framework include standards and lexicon, legal authority, technological advances, workforce, and analytic capacity. Three data sources are used to examine the study questions: 1) National Birth Defects Prevention Network (NBDPN) Program Survey, 2) NBDPN Annual Report Directory, and 3) semi-structured interviews with nine birth defects surveillance program directors.

Forty-three states perform population-based surveillance for birth defects, covering approximately 80% of live births in the U.S. Seventeen employ active case-finding approaches, whereas 26 rely primarily on passive approaches. These programs all monitor major structural malformations; however, passive case-finding programs generally monitor a broader list of conditions, including developmental and newborn screening conditions. More active case-

finding programs use clinical reviewers, cover more pregnancy outcomes, and access more extensive prenatal and postnatal health data sources.

Population-based birth defects surveillance programs focus their activities on generating prevalence data and understanding the impact of these conditions on the community. They also focus on epidemiological studies, service planning, and referral of affected infants to medical and social services. However, as life expectancy for children born with birth defects increases, these surveillance programs should increase their efforts to better understand health outcomes and the types and level of service utilization of these children across their lifespan.

The semi-structured interviews on the application of the CDC Strategic Framework for birth defects surveillance programs suggest that programs should: 1) concretely communicate the utility of their surveillance data in order to increase the transparency of their activities to stakeholders; 2) ensure their surveillance activities are politically acceptable; 3) prepare for a new reality of data flux as medical information transition from paper to electronic formats and increase its reliance on bi-directional system communication for data transactions; 4) seize on remote access opportunities for increased efficiency of record review and verification; 5) strengthen workforce knowledge on information technology (IT) given reliance on IT systems; and 6) build a community of peer learning through collaborative work using pooled data to address public health impact and epidemiological issues.

The breadth and depth of information collected at a population level by birth defects surveillance programs in the U.S. serves as an important data source to guide public health action. Collaborative efforts at state and national levels can help harmonize data collection and increase the utility of birth defects programs.

I. Background and Problem Statement

Ia. Background and Context

Public health surveillance is "the ongoing, systematic collection, analysis, interpretation, and dissemination of data regarding a health-related event for use in public health action to reduce morbidity and mortality and to improve health" (CDC, 2001). This activity enables public health professionals to carry out the core functions of assessment, policy development, and assurance. Surveillance systems designed to monitor health conditions vary greatly in methodology for data collection and use. The changing landscape of electronic health record adoption, consolidated data sources, and diminished resources is altering the practice of public health surveillance for disease-specific conditions such as birth defects.

Birth defects are conditions that: 1) result from a malformation, deformation, or disruption in one or more parts of the body; 2) are present at birth; and 3) have a serious, adverse effect on health, development, or functional ability (CDC, 2006). They are common, affecting one in every 33 babies; costly, over $2.6 billion spent on just hospital costs in a given year; and deadly, contributing to one in every 5 infant deaths (CDC, 2011). Although the U.S. lacks a unified national population-based surveillance system to collect data on major birth defects, 40 states and Puerto Rico currently have a system to monitor these conditions (NBDPN, 2012). The first state statute that established a program to capture birth defects from reporting sources (passive case ascertainment) was enacted in New Jersey in 1926; approximately 40 years later, the U.S. Centers for Disease Control and Prevention (CDC) began the first active case ascertainment system within the Metropolitan Atlanta area (Mokdad, Annest, Ikeda, & Mai, 2010). Although these systems highlight two basic approaches to population-based birth defects

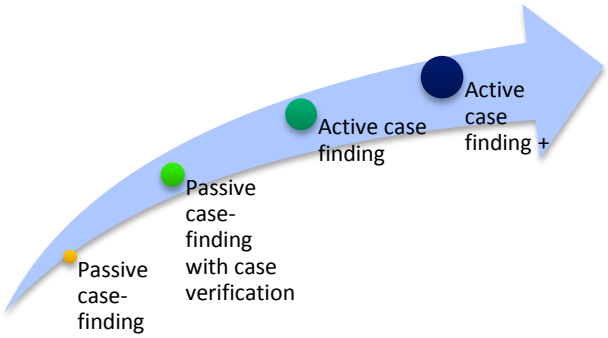


Figure 1. Case Ascertainment Methodologies for Birth Defects Surveillance Program

surveillance, adaptations and varying degrees of intensity in case ascertainment are seen among state programs (Figure 1). This continuum ranges from the simplest system (purely passive case ascertainment with limited data sources for live births only) to an active case ascertainment system with extensive data sources for all affected pregnancy outcomes. A number of factors affect the methodology, including the purposes and functions of the programs (TABLE I). This table was adapted from the National Birth Defects Prevention Network's "Guidelines for Conducting Birth Defects Surveillance" (NBDPN, 2004).

The practice of birth defects surveillance has remained relatively unchanged for the past few decades, but several emerging forces, such as electronic health records and health information exchanges, will affect the way birth defects surveillance is conducted. The collection and transmission of clinical and laboratory data are going through dramatic changes, which will influence data collection for public health surveillance systems. In addition, birth

TABLE I. PURPOSES / OBJECTIVES OF BIRTH DEFECTS SURVEILLANCE PROGRAMS

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

defects surveillance systems have generally focused on ascertaining cases at birth, but wider recognition of the need to monitor health outcomes of these children as they age could shift how the programs are designed. Therefore, a thorough evaluation of birth defects surveillance programs is needed to assess the function and utility of these systems and how they are positioned to meet emerging challenges.

Ib. Problem Statement and Study Objectives

In July 2012, CDC released its "Vision for Public Health Surveillance in the 21st Century" report that highlights six major areas that must be addressed to advance public health surveillance, including: 1) lexicon and conceptual framework; 2) global health surveillance; 3) information sciences and technological advances; 4) surveillance work force; 5) access and use

of public health surveillance data covering legal, policy, ethnical, regulatory, and practice concerns related to data sharing; and 6) analytic challenges such as database management (Buehler, 2012). These areas form the *CDC Strategic Framework* for Public Health surveillance that can provide a structure to examine the readiness of birth defects surveillance programs to meet current and future needs. Given the diverse challenges and approaches to surveillance for domestic and global birth defects surveillance, the scope of this project will focus predominately on surveillance programs in the U.S., but the lessons learned could potentially have implications for global surveillance practice.

Population-based birth defects surveillance is an important function performed by state health departments, yet no comprehensive evaluation of the collective efforts in the U.S. has been conducted. Therefore, this research project will systematically assess U.S. population-based based birth defects surveillance programs to determine how well they meet both intended purposes and how poised these programs are to meet emergent needs.

Ic. Study Questions

This DrPH research focuses on the following study questions:

1) What are the characteristics, e.g., methods and infrastructure, of the population-based birth defects surveillance systems in the United States?
2) To what extent are population-based birth defects surveillance programs in the U.S. addressing the intended purposes of the systems?
3) How prepared are birth defects surveillance programs to address the major considerations identified in "CDC's Vision for Public Health Surveillance in the 21st Century"?

Id. Leadership Implications and Relevance

Surveillance is a core public health activity, and programs developed to monitor health conditions need to ensure that they are responding effectively and efficiently to public health problems. Many states have established surveillance systems for disease-specific conditions such as birth defects; therefore, an assessment of the design and utility of these systems is needed to better understand current and future practice. The infrastructure of these systems relies on on-going state and federal investments, and, given resource constraints, it is important to reflect and strategically determine approaches to collect the information needed to ensure maximum return on government investment.

Public health surveillance systems are dynamic, and the changing field of health information data sources and exchanges coupled with the problem of diminished resources requires a critical review of the practice of population-based surveillance for birth defects. Lessons learned for population-based birth defects surveillance systems in the U.S. could have implications for other systems around the world.

II. Relevant Literature and Conceptual Framework

IIa. Literature Review

A literature review was conducted using PubMed, Embase, and Web of Science databases using the following search expressions: ‘population-based birth defects surveillance’, ‘public health surveillance’, and ‘birth defects methodology’. Based on the research questions for this DrPH dissertation, the review centered on 3 areas: 1) intended purposes and approaches to birth defects surveillance, 2) demonstrated utility of birth defects surveillance systems for public health action, and 3) major surveillance considerations (lexicon and standards,

information science and technology, skilled workforce, data access and use, and strong data analytic foundation) as they relate to birth defects surveillance programs.

Existing Practice: Intended Purposes and Approaches

Why establish a surveillance system for birth defects?

The majority of surveillance systems for birth defects were established from a community's perceived need for data as a response to the requirement for evidence-based information to carry out a public health action. Many of the systems around the world, especially those in the Europe and United States, were established in the late 1960s, 1970s, and 1980s due to concerns about environmental triggers of birth defects, such as Thalidomide (Botto et al., 2006; Boyd et al., 2011; Edmonds, 1997). Concern over environmental triggers is still used today as one justification to improve and sustain programs. Wedgwood (2012) recently called for national funding to improve birth defects monitoring in England and Wales to ensure that potential clusters of defects could be investigated.

In addition to the traditional use of surveillance data for investigations into environmental exposures and clusters, Correa and Kirby (2010) outlined other traditional uses of this data, such as descriptive epidemiology involving the quantification of the burden of diseases, identifying populations at risk and/or health disparities, monitoring trends, evaluating outcomes among children with birth defects, evaluating prevention efforts, and participating in etiologic research. They also advocated for developing enhanced ways to use the information collected with the interest in "chronic conditions that impact individuals across the lifespan including the prevalence of adolescents and adults living with birth defects in defined communities, and the development of evidence-based guidelines and policies for secondary prevention."

Hanson (1995) noted that "despite a quarter century of efforts, adequate research and effective prevention and intervention strategies remain largely unrealized for fetal alcohol syndrome and for most other birth defects categories." He pointed to the issue that the public health community itself needs to be better informed about birth defects and to view it as part of a broader public health agenda; yet he also highlighted some positive trends, including the feasibility of birth defects surveillance systems to provide valuable information to understand causes, risk factors, and impact of these conditions; availability of knowledgeable birth defects epidemiologists; and congressional actions to fund these programs. During this time, interests in establishing birth defects surveillance systems also focused on primary prevention given the emerging evidence that pointed to the role of folic acid in preventing neural tube defects, and the ability of these systems to help link families and affected individuals to medical and social services. The newly created surveillance systems in the 1990s and recent years therefore focused on addressing prevention and intervention activities (Mokdad et al., 2010).

The recent addition of newborn screening for critical congenital heart diseases or defects (CCHD) to the Recommended Uniform Screening Panel of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children has spotlighted the role of birth defects surveillance programs. Olney and Botto (2012) outlined several areas where birth defects programs can potentially help with the evaluation aspects of CCHD screening, including: "1) health outcomes after newborn screening among affected children; 2) missed primary targets of screening (i.e., affected children who were not screened or had false-negative screens); 3) burden and screening accuracy for secondary targets; 4) the role of altitude, sociodemographic characteristics, and other special circumstances; 5) the contribution of prenatal and clinical diagnoses before newborn screening; and 6) costs and service utilization."

What approach to take?

The defining characteristics of birth defects surveillance systems in the U.S. include population-based coverage and multiple data source methodology. Population-based refers to a well-defined geographic coverage that encompasses a catchment area of state or contiguous counties within a state. Multiple data source methodology allows a system to enhance its ability to capture and validate as many cases of birth defects as possible. Beyond these two characteristics, variability exists among the systems.

Geographic coverage

A unified, systematic surveillance system for collecting birth defects data covering the entire U.S. has never been developed; however, attempts to understand the national prevalence and trends in birth defects have used administrative datasets such as the Healthcare Cost and Utilization Project (HCUP) and birth certificates (Bird, Hobbs, Cleves, Tilford, & Robbins, 2006; Honein, Paulozzi, Mathews, Erickson, & Wong, 2001; Lary et al., 1997). Relying on nation-wide administrative datasets has had limited uses given data quality concerns. For example, the birth defect variable on the birth certificate has repeatedly been shown to have low sensitivity for most birth defects (Honein & Paulozzi, 1999; Northam & Knapp, 2006; Watkins et al., 1996). Additionally, Boulet et al. found that the sensitivity varies significantly by race/ethnic groups; therefore, assumption about uniform under-ascertainment across these groups might be problematic (Boulet, Shin, Kirby, Goodman, & Correa, 2011).

Bird et al. (2006) proposed using HCUP databases with national weights as a complement to state-based birth defects surveillance systems for estimating prevalence. This could have some use for conditions that are recognizable at birth, although he notes several

limitations, including under-ascertainment by "as much as 28% of infants ultimately diagnosed with a birth defect through other means and lack of ability to determine multiple hospitalizations by a given patient." Given the challenges presented with available administrative datasets, increasing use of pooled data from multiple state birth defects surveillance systems has grown in recent years (Canfield et al., 2006; Parker et al., 2009; Parker et al., 2010; Kirby et al., 2013).

A number of countries that have attempted a "national" system such as Canada and England have reported difficulties in ensuring on-going quality data for the covered geographic areas. Misra, Dattani, and Majeed (2006) and Boyd et al. (2005) found that the National Congenital Anomaly System (NCAS) established in England and Wales was weak given its lack of any legal requirement to collect data on babies born with congenital anomalies, lack of integration with national birth and death databases, and limitation that hamper bi-directional communication with the providers. These challenges, especially limited resources and lack of legal requirements, are not unique to NCAS but are felt as well in other countries attempting to establish or maintain a national system. The national Canadian surveillance of congenital anomalies has had similar challenges, and Lowry (2008) proposed the development of a collaborative network of surveillance programs in Canada that would be similar to the European Surveillance of Congenital Anomalies (EUROCAT, http://www.eurocat-network.eu) or the U.S. National Birth Defects Prevention Network (NBDPN, http://www.nbdpn.org).

Multiple Data Sources, Case ascertainment, and Case Verification

A second defining characteristic that birth defects surveillance systems in the U.S. share is multiple source case ascertainment methodology. The basic passive systems rely on record linkage of administrative data using diagnostic codes from birth certificate and hospital discharge

datasets without verifying the case information. The more advanced and comprehensive systems rely on trained abstractors to collect case information from medical records and other data sources for all pregnancy outcomes (births, stillbirths, and terminations). The CDC's surveillance system in the metropolitan Atlanta area (MACDP) first employed this methodology over 40 years ago (Correa-Villasenor et al., 2003; Correa et al., 2007). Given the intensity in case finding, this is considered the "gold standard" (best approach) since it can offer the most precise birth defects data. Also, the addition of clinical review for accurate diagnosis offers the system maximum utility "given continuous, systematic data collection with identifiers, range of uses from on-going monitoring (basic prevalence) to more complex data uses (risk factor studies, mortality and morbidity, health service utilization) and prevention (developing and evaluating NTD prevention strategies related to the periconceptional use of folic acid supplements)." Approximately one third of the systems in the U.S. use an intensive, active case finding methodology, while the remaining two thirds of the systems rely on a passive reporting system that may or may not include case verification(NBDPN, 2012). Salemi et al. (2012) compared the passive case ascertainment methodology used by the Florida Birth Defects Registry to an enhanced system with hospital medical record review, and concluded that for epidemiologic or clinical studies, the program should implement a more comprehensive case ascertainment strategy that includes case verification. Similarly, the Department of Defense (DOD) conducted a feasibility study for conducting birth defects surveillance among military personnel and concluded that the administrative datasets would be enhanced with case verification (Bush, Smith, Honner, & Gray, 2001).

Another important aspect to consider is the types of data sources and what they can contribute to the overall completeness of a system. Feldkamp and colleagues stressed the

importance of understanding what the data sources can offer; they reported in their study that almost all their sources captured true cases of congenital heart defects not identified by another source and demonstrated the importance of having "ascertainment sources that vary along dimensions such as timing of diagnosis (pre- vs. postnatal), timing of reporting, and pregnancy outcome" (Feldkamp, MacLeod, Young, Lecheminant, & Carey, 2005).

Ideally, birth defects surveillance systems should capture all pregnancy outcomes, but this can be challenging. While most systems capture both live births and fetal deaths, approximately 40% are able to capture terminations. For some conditions, the lack of other pregnancy outcomes can greatly affect data completeness. Cragan and Gilboa (2009) found that adding prenatal sources (perinatologists' offices) to their other data sources increased the total defect prevalence by approximately 7% (28 per 1000 to 30 per 1000).

Organization location

The organizational location of a program will partially drive the emphasis of the system. Over half of birth defects programs reside within the maternal and child health (MCH) department and the other half are located within environmental health, vital statistics/health, university, or other locations (NBDPN, 2012). Walker (2000) noted that "State MCH programs need population-based community-level data in order: to conduct local and state needs assessment; to monitor state- and local-mandated programs, services, and screenings; to direct program planning and management; to conduct program and service evaluations; and to ensure that people identified through screening receive appropriate follow-up services." For a birth defects surveillance system to be relevant within MCH, she suggested that the birth defects

program should be included in the Title V MCH block grant application and birth defects prevention should be included.

Conditions monitored

At the core of all birth defects surveillance systems is the ascertainment of major structural defects, but the variability lies in which of the major structural defects to monitor as well as the addition of potentially related conditions and risk factors. For example, all programs collect a few core structural conditions such as spina bifida and clefts, but many systems expand their case definition to include selected developmental disabilities, hearing and metabolic conditions, muscular dystrophy, risk factors such as medications and assisted reproductive technology (ART), as well as reproductive outcomes such as stillbirths and preterm births

Two approaches for these expanded conditions, e.g. developmental disabilities, exist either as an addition to the current birth defects surveillance system or through periodic linkage with other existing data sources. Kirby, Brewster, Canino, and Pavin (1995) extended "a birth defects registry to identify cases of developmental disorder by adding cases of developmental disorders in early childhood by adding all known sources of diagnosis and service to case-finding methods." They concluded that adding developmental disabilities to an existing birth defects surveillance infrastructure was possible. The "approach has potential as a source both for case-control studies of the etiology and risk factors for specific categories of developmental disorders and as a conduit for referrals into early intervention programs and specialized services." They estimated that adding the developmental conditions "added 30-35% to the total cost of medical records abstraction and data processing and decreased the volume of records per abstracter to 6000 to 7500 birth events per year." The data "indicate that many birth defects are associated

with developmental disorders, and more research is needed on reliability and validity of diagnoses obtained from different types of facilities and specialists of health practitioners and of the contribution of each source to the overall prevalence estimates." A few state programs, such as New Jersey, have included selected developmental disabilities in their list of case inclusion (NBDPN, 2012). Others have linked birth defects surveillance data with existing data systems to examine the co-occurrence. For example, CDC's birth defects surveillance system was linked with data from the developmental disabilities surveillance system (Decoufle, Boyle, Paulozzi, & Lary, 2001; Schendel, Autry, Wines, & Moore, 2009). They concluded that their study "highlights possible early prenatal origins of some developmental disabilities and suggest that both the number of birth defects present and the number of anatomic systems involved are strongly related to functional outcomes. Our data suggest that birth defects pose a greater burden on society than previously recognized." Schendel et al. (2009) examined the co-occurrence of autism and birth defects and concluded that "birth defects were found among 6% of children with autism and were associated with a near twofold increased risk for autism overall."

Programs have used similar approaches to examine other conditions, including hearing and metabolic conditions as well as muscular dystrophy. The New York birth defects program was able to use its existing system to conduct long-term follow-up of children with genetic or metabolic disorders identified through newborn screening (Wang, Caggana, Sango-Jordan, Sun, & Druschel, 2011). They were able to examine this issue by linking data sources from both newborn screening and birth defects programs with vital records, hospital discharge files, and early intervention program. Miller et al. (2006) explored ways to expand existing infrastructure for muscular dystrophy surveillance. Each participating state used a different approach to build upon its existing resources and collaborations, e.g., being able to use current data sources such as

neuromuscular clinics, hospitals, and hospital discharge databases, private physicians, service sites for children with special needs, and birth defects surveillance programs. For the birth defects surveillance program, the staff was able to build upon their experience with data linkage, but given that muscular dystrophy was not included in its birth defects surveillance system, modifications such as expanding age limit to cover a later age (4 or 5 years old) and adding new data sources were needed.

Beyond monitoring specific conditions, birth defects surveillance program infrastructure has been used to examine risk factors such as medications and ART, as well as reproductive outcomes such as stillbirths and preterm births. Given that many systems were established to monitor adverse events, being able to collect medication data has been of interest to programs. Lisi et al. (2010) determined that it was possible to use routine data collected from birth defects registries to examine association between medications and risk for birth defects using an "exposed case-only" design. Another factor of interest is ART and its potential effect on health outcomes. Hansen et al. (2007) showed that less than one third of ART children identified with a major birth defect on the Western Australian Birth Defects Registry reported to the national Australian Assisted Conception Data Collection. In the U.S., the National ART Surveillance System (NASS) enhances its data collection by linking with other surveillance registries, including state birth defects systems (http://www.cdc.gov/art).

Likewise, similar approaches have been used to examine for reproductive outcomes such as stillbirths and preterm births. Duke, Williams, and Correa (2008) reported on expanding existing birth defects surveillance programs to include data for all stillbirths to mutually enhance the information collected from fetal death reports and this practice could benefit such programs by improving the ascertainment of birth defects. Makelarski et al. (2011) points to the need to

use active case finding with multisource case ascertainment for stillbirths, since relying solely on fetal death certificate data can be problematic.

Preterm births have been examined through record linkage. Rasmussen, Moore, Paulozzi, and Rhodenhiser (2001) linked CDC's birth defect surveillance system with its active case finding methodology to birth certificates data (gestational age) and found that the risk for birth defects is increased in premature infants. Similarly, Honein et al. (2009) examined data from 13 state population-based birth defects surveillance systems (covered about 30% of all U.S. births) and concluded that birth defects were more than twice as common among preterm births (24-36 weeks) compared with term births.

Utility of birth defects surveillance systems

The focus of this literature review has been not only to highlight potential uses and applications of birth defects surveillance systems, but also to demonstrate what has been done to show the value or utility of the systems. The review focused on core uses of birth defects data for etiologic research, multi-program monitoring and epidemiologic studies, referral to services, primary prevention, mortality and morbidity, including health service utilization. The New York State Department of Health (DOH) Congenital Malformations Registry (CMR) was under political pressure to show utility and accountability. They evaluated the registry by the intended purpose of the registry: 1) detect birth defects, 2) investigate potential etiologic factors, 3) plan and evaluate interventions, and 4) ensure appropriate care for persons in need (Druschel, Sharpe-Stimac, & Cross, 2001). The New York State DOH Committee concluded that CMR provided an important public health service but could streamline cost. This evaluation caused the program to develop technological efficiency that improved timeliness.

Monitoring and cluster investigation of environmental factors

Core to all birth defects surveillance systems is the ability to generate prevalence data regarding the population of interest. Another important role is on-going monitoring of specific defects and the ability to respond to community concerns about potential clusters. Often times, finding a causal link is problematic given small numbers of cases, and most attempts result in inconclusive findings. Dolk (2004) pointed out that "the likelihood of finding a common causal factor is so low that it may often be better not to investigate but instead to clean up the mess of the suspected contaminant without demanding causal proof." She proposed using a multi-site investigation of all communities with the suspected contaminant instead of focusing on just one site. Kucik et al. (2008) proposed another approach in proactively collecting the data from a series of cluster investigations using the same study methodology (interview guide and biologic specimen) to enable pooled analysis of the findings. Both methods discussed so far are reacting to a potential cluster while Yang et al. (1997) examined this issue from a proactive monitoring approach, using limb deficiencies as a case study to determine its ability to provide early warning of fetal exposure to Thalidomide. Thalidomide was commonly used as a sedative and antiemetic drug for morning sickness in the 1950s, but by 1962, it was withdrawn from European and Canadian markets since thousands of children were born with severe limb deformities related to the use of the drug (Franks, Macpherson, & Figg, 2004); the drug was reintroduced to the U.S. market in 1998 as a treatment for leprosy and now for selected cancers. In order to examine the effects of drugs such as Thalidomide, Yang et al. concluded that a large birth population (beyond 50,000 per year) is needed to detect a pregnancy exposure rate of 3.5% or less as well as the need for accurate case classification of the defects.

Etiologic Research

Reefhuis, de Jong-van den Berg, and Cornel (2002) conducted a comprehensive review of the use of birth defect registries for etiological research; they define registry as "a standardized, on-going ascertainment of children with birth defects, and possibly healthy controls, without a predetermined end date of birth after which infants with birth defects are no longer included in the registry," and found more than 100 birth defects registries in existence. They focused etiologic research on risk factors for CVS and amniocentesis, surgery during pregnancy, fever or illness during the first trimester, epilepsy and anticonvulsants, maternal drug use, maternal age, paternal age, lifestyle factors, maternal smoking, socioeconomic status, environmental exposures – pollution, tap water, hazardous waste sites, radiation, maternal occupation, and paternal occupation. They concluded that birth defects registries have been used to study many different risk factors in order to detect new teratogens and to quickly confirm or refute alarming results of case reports or other studies. However, the data still has to be explored further as a sensitive instrument to identify new risk factors, such as gene-environment interactions.

Multi-state collaborations

A number of multi-state or multi-national collaborations have been established to pool data from multiple surveillance systems to increase the power of the study projects to better address public health impact and to explore risk factors for birth defects. In the U.S., the collaboration is managed through the National Birth Defects Prevention Network (NBDPN). Other collaborations of surveillance systems include the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) and EUROCAT. ICBDSR is a collaborative effort of over 40 birth defects registries worldwide. Botto et al. (2006) highlighted several examples,

including: a rapid and annual assessment of major birth defects that has evolved to include other outcomes and thereby to promote the use of data for public health action; basic epidemiologic assessment to promote etiologic studies in areas of the world where data are often lacking; increasing collaborations with other networks in Europe and United States to develop guidelines and tools. While the ICBDSR and NBDPN are structured as a network of programs that are independent with no common databases, EUROCAT has created a shared database to lessen the variability in data reporting from their member registries.

Referral to Services

The NBDPN Guidelines for Conducting Birth Defects Surveillance (NBDPN, 2004) outlined some uses of birth defects surveillance data for "human services programs [that] 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." Farel, Meyer, Hicken, and Edmonds (2003) surveyed state birth defects surveillance programs to determine how these systems support referral efforts. At the time of the study, 32 of 52 states said they have an operational birth defects surveillance program, and 29 of these programs said they were either planning or had implemented a referral system. The authors further noted that the "cost of this activity against other public health and early intervention programs is likely much less than the cost of services for individuals whose secondary disabilities might have been prevented." Sharpe-stimac, Wang, Druschel, and Cross (2004) and Montgomery and Miller (2001) found that contacting families with information and/or referral to services was helpful and useful.

Evaluation of Prevention Strategies

Birth defects surveillance programs have traditionally focused on primary and secondary prevention. The most-well known example of primary prevention is folic acid intake for the prevention of neural tube defects. Honein et al. (2001) and Williams et al. (2002) both reported decreases in the prevalence of neural tube defects in the U.S. following the implementation of mandated folic acid fortification in all cereal grains. Grosse, Waitzman, Romano, and Mulinare (2005) concluded that folic acid fortification provided an annual economic benefit of $312 million to $425 million; they further noted, "few public health interventions beyond immunization and injury prevention are cost saving and that folic acid fortification is exceptional in the relative magnitude of economic benefits." Part of the cost saving calculations required data from birth defects surveillance systems.

Programs continue to explore other primary preventions strategies. The emerging risk factors, e.g. diabetes, obesity, and smoking, for targeted prevention strategies are multi-factorial and complex to address. Figure 2 shows the estimates of attributable fraction given selected maternal risk factors. A strategy for addressing these risk factors is to promote preconception health. The Preconception Health and Health Care Initiative launched a national campaign, *Show Your Love*, in February 2013 to improve maternal and infant health (http://www.cdc.gov/preconception/index.html).

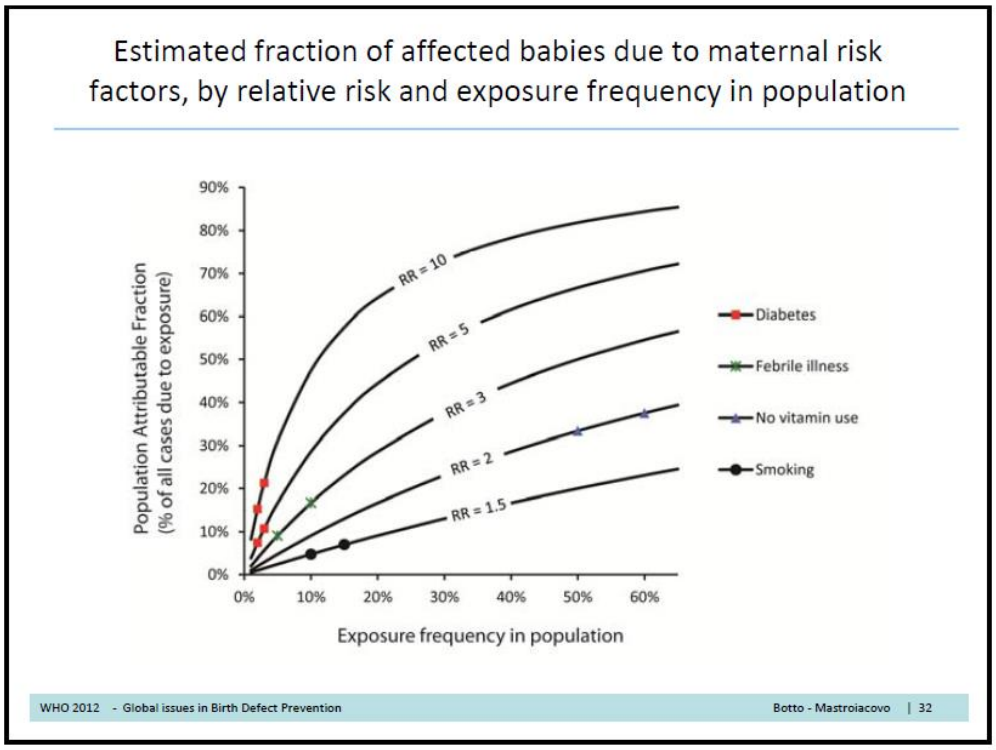
Survival and Health Service Utilization

Given the length of time that a number of birth defects surveillance programs have been established, they are able to examine long-term survival of children with birth defects. Shin et al. (2010) found that "the 1-year survival probability among infants with spina bifida showed improvements for whites (from 88% to 96%), blacks (from 79% to 88%), and Hispanics (from

88% to 93%).” Wang, Hu, Druschel, and Kirby (2012) reported an overall 25 year survival probability of 82.5%.

As survival increases for children with birth defects, being able to better understand long-term outcomes, including health service utilization, is important. For example, Yazdy, Autry, Honein, and Frias (2008) found that 26% of the children with oral facial clefts were in special

Figure 2. Estimates of Attributable Risk Factors



Source: Botto and Mastroiacovo www.searo.who.int/entity/child_adolescent/topics/child_health/birth_defects/en/index.html

education at least one year compared to 8% of children who had no major birth defects. They were able to study this by linking birth certificates data with CDC's birth defects surveillance program and special education files.

Newborn Screening for Critical Congenital Heart Defects

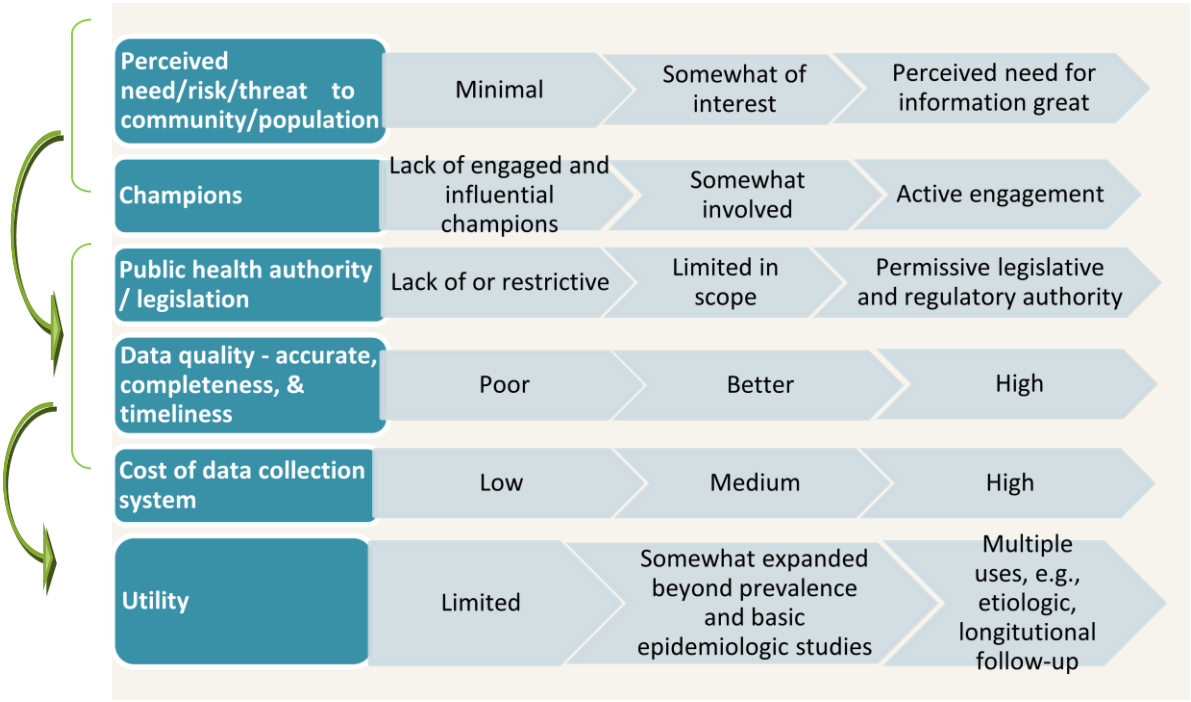
An enhanced use of data from birth defects surveillance systems has recently focused on understanding the impact of the critical congenital heart defects (CCHD) that are targeted for newborn screening using pulse oximetry. The National Birth Defects Prevention Network (NBDPN) focused its 2012 Annual Report data brief on the prevalence of seven primary targeted CCHD (Mai et al., 2012). This data highlighted the role that birth defects surveillance systems are able to play in providing data of national interest. In addition, attention has focused on evaluation of the CCHD newborn screening activities. The NBDPN also collaborated with CDC to assess state birth defects surveillance programs' roles in evaluation. They found that 28 states reported the ability to evaluate mortality associated with CCHD and 16 could examine related morbidities (Copeland et al., 2012).

Summary of review on public health practice of birth defects surveillance

Surveillance systems stem from a community's perceived need for data, resultant actions, and sustainability of these actions. Figure 3 provides a summary of key attributes that will determine the strength of a surveillance system. The perceived need along with champions can influence the authority granted, level of resource investment, and data quality, which will in turn influence the utility of the systems.

Attention to requestors of data and this entity's level of influence in directing funding for surveillance efforts is necessary. As stated in the NBDPN Guidelines for Conducting Birth

Figure 3. Attributes of a Surveillance System



Defects Surveillance (NBDPN, 2004), "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 about their own agency and other agencies about birth defects surveillance and its importance and potential uses in the public health field."

Birth defects systems need to evolve with needs of the community to ensure usefulness and sustainability of public health action. The 65th World Health Assembly adopted Resolution

WHA 63.17 in May 2010 that highlighted the importance of surveillance, research, prevention, and intervention actions to address birth defects given their impact on infant and child morbidity and mortality (WHO, 2010).

Preparation for 21st century for public health surveillance

The first two sections of the literature review focused on intended purposes and demonstrated utility of birth defects surveillance systems. This next section will focus on preparedness of birth defects surveillance systems to address challenges and issues raised in CDC's vision for public health surveillance in the 21st century, which calls for: 1) common lexicon and conceptual framework; 2) development of information sciences and technological advances; 3) enhancing surveillance work force; 4) access to and use of public health surveillance data covering legal, policy, ethnical, regulatory, and practice concerns related to data sharing; and 5) addressing analytic challenges such as database management (Buehler, 2012).

Lexicon, Conceptual Framework and Standards

A shared lexicon and framework is necessary to ensure consistency in public health practice. Hall et al. (2012) stressed that "public health surveillance is not defined by the system used to collect data but by the purpose of the data collection — the specific public health question that the data will be used to answer and the link to disease prevention and control." Currently, no national standards exist for birth defects surveillance in the U.S. In 2004, the NBDPN released its guidelines for conducting birth defects surveillance (NBDPN, 2004). These guidelines were established to provide guidance to state programs on what they should do to ensure quality data for public health action. However, wide variability exists, and states have

been looking for guidance on what must be done, especially given fiscal constraints. The NBDPN is in the process of developing standards focusing on data quality and utility.

Terminologies are not always used consistently and can be a source of confusion. For example, the terms "active," "passive," and "hybrid" are generally used to describe case finding approaches, yet confusion exists since these terms can be used loosely to describe the system's entire ascertainment process instead of just the case finding process. Therefore, if a system has passive data collection and uses an abstractor to verify cases, this system is still passive and is not considered "active" just because it uses staff to confirm cases. This same confusion also occurs for data from hospitals (data source) vs. hospital-based ascertainment (approach). Although all systems rely predominately on hospital data as a source, systems that capture all births within a geographic area are considered population-based. In the U.S., all birth defects surveillance systems within the NBDPN are considered population-based. In many parts of the world where the birth population cannot be reliably obtained, a hospital-based approach is often used. One of the most well-known example is ECLAMC, which is a "program for the clinical and epidemiological investigation of risk factors in the etiology of congenital malformations in Latin America hospitals, using a case-control methodological approach" (Castilla & Orioli, 2004). ECLAMC has shown that its hospital-based approach to collecting data on birth defects can be used for epidemiologic purposes, but they do note some limitations, including selection bias since hospitals that are likely to voluntarily participate will be more likely the "high complexity hospitals."

Programs designed to collect data on birth defects have used the term "registry" and "surveillance system" interchangeably. Generally, registry refers to "a file of data concerning all cases of a particular disease or other health-relevant condition in a defined population such that

the cases can be related to a population base" (Porta, 2008). However, registries are also used currently to refer to databases of affected individuals that are not population-based. For example, the National Institutes of Health (NIH) has established disease registries, such as the National Down Syndrome Patient Registry (http://downsyndrome.nih.gov/registry/Pages/default.aspx), where individuals or families consent to participate.

Although many of these terms are defined in the NBDPN guidelines, they are subjective to interpretation. As standards are developed, some of the key terms will need to be reinforced to ensure a common lexicon.

Information Science and Technology

Savel and Foldy (2012) discuss public health informatics as the "systematic application of information and computer science and technology to public health practice." They have outlined steps that public health agencies should take, such as 1) adopting specified standards from the Office of the National Coordinator for Health Information Technology (ONC) for accepting surveillance information from health care providers; 2) working with partners, such as academic centers or other agencies, to facilitate the transition to the use of standardized electronic data; 3) using or modifying existing systems, instead of stand-alone systems, that can be leveraged for multiple purposes; and 4) considering incremental steps rather than "immediate wholesale changes." Also, system communication is important. The Health Information Technology Standards Panel (HITSP) has created interoperability specifications that describe standards-based approaches for exchanging electronic healthcare data (Grannis & Vreeman, 2010).

Birth defects programs have attempted to leverage lessons learned from cancer surveillance (http://www.cdc.gov/cancer/npcr/meaningful_use.htm). An implementation guide for cancer surveillance has been developed that includes "business rules and specifications for EHR systems that include reportable cases, specific data elements, Health Level 7 Clinical Document Architecture (HL7 CDA) event report, and secure electronic transmission mechanism," which can potentially be used as a template for birth defects surveillance. Steps needed for birth defects surveillance include: 1) review of all relevant standards affecting the practice of birth defects surveillance, 2) inclusion of birth defects surveillance activities as part of Centers for Medicare and Medicaid Services (CMS) Meaningful Use efforts (stage 2 "special registry" category), and 3) developing tools to assist state programs in adopting practices for EHR.

Skilled Workforce

Drehobl et al. (2012) stress the importance of a workforce analysis, which should "include enumeration of the workforce and existing gaps, forecasting and identifying future needs, and monitoring how a workforce analysis is applied to addressing programmatic needs." There has not been any analysis on the workforce for birth defects surveillance systems. CDC has worked with the NBDPN to conduct periodic webinars on topics such as evaluation, methodology, coding, and media training. In addition, the NBDPN annual meeting provides a forum for trainings on current surveillance practice.

Additional actions suggested by Drehobl et al. (2012) included: "conducting job task analysis with representatives of different disciplines; identifying administrative inefficiencies (e.g., conduct cost analyses) and needed technologic tools; acquiring resources to ensure access to those tools; providing opportunities for career advancement; and monitoring workforce

retention." They also discussed partnerships among stakeholders to increase "visibility of workforce needs and influence supportive policies within organizations and at federal, state, and local levels."

Data Access and Use

For population-based birth defects surveillance systems, access to data sources with identifiers is essential. Without this information, linkage and much of the potential uses for the data become limited. Bernstein and Sweeney (2012) discussed three factors that affect the ability of data stewards to share with surveillance programs: "1) rules and regulations governing how and why the data are collected and released; 2) the availability of resources to put the data into a form that can be shared, and 3) the willingness to use those resources." Birth defects surveillance systems have legislative or public health authority to carry out their activities. This has allowed them access to medical records and administrative datasets. Appendix B contains a complete listing of birth defects surveillance programs' authority.

Informed consent to access medical records for the surveillance function of public health programs has generally not been required. This practice is necessary since gathering informed consent would negate the strength of population-based approach and introduce bias in the surveillance data. Mai et al. (2007) discussed the importance of this approach and the potential bias this introduces. Only one state (Wisconsin) has required opt in.

While recognizing the need for state programs to collect the information, some states have attempted to limit the duration of data access. In recent years, some states, including Minnesota and New Hampshire, have passed legislation that allows them to develop an opt out system but the program needs to remove identifiers after the child reaches a certain age. For

example, New Hampshire statue states that "the program shall not obtain any individually identifiable health information for any individual who does not have a confirmed birth condition diagnosis and shall retain the name and address only of any such individual for a period not to exceed 2 years" (http://www.lawserver.com/law/state/new-hampshire/nh-statutes/new_hampshire_revised_statutes_141-j_5). Gill, Miller, Broussard, and Reefhuis (2012) studied the New Hampshire opt out law and found significant differences in race/ethnicity and maternal age between those who opted out and those who did not.

Another challenge for some of the state programs is the availability of the state hospital discharge data. While many have access to the discharge data with identifiers to allow for record linkage, some have only had access to de-identified data files, which limit its linkage capacity.

Strong Data Analytic Foundation

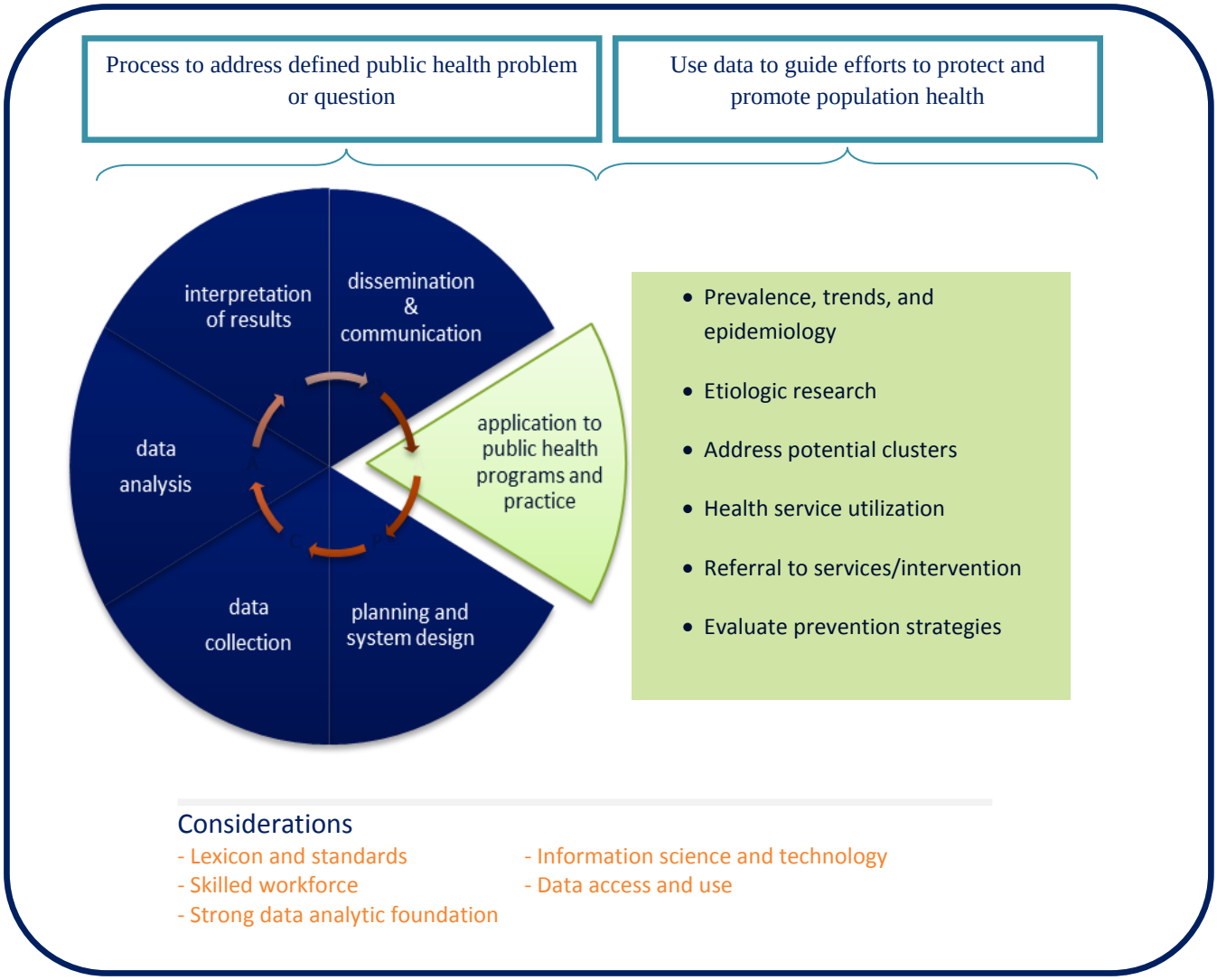
Rolka et al. (2012) note that the "root of effective disease control and prevention is an informed understanding of the epidemiology of a particular disease based on sound scientific interpretation of evidence," and discuss how surveillance programs must move "raw data" to "consumable information" that can be used for public health action. A strong analytic staff and data processing tools are needed to handle the complexity of information systems. Garne et al. (2011) discussed a computer algorithm developed for the EUROCAT, a collaboration of birth defects systems in Europe. They concluded that their computer algorithm can be used to decrease the case load for clinical review, especially for large populations, and understanding if a defect is isolated or multiple is key for certain utility, such as survival. Other software and tools, such as a data linkage tool called *Fine-grained Record Integration and Linkage* or FRIL is helpful in assisting programmatic staff with data analysis (Jurczyk, Lu, Xiong, Cragan, & Correa, 2008). Beyond software and tools developed with birth defects surveillance functions,

tools from the cancer surveillance such as Registry Plus (http://www.cdc.gov/cancer/npcr/tools) need to be explored for potential application to birth defects.

IIb. Conceptual Framework

The CDC's definition of public health surveillance comprises two major areas: process cycle to address a defined public health problem and utility of data to guide efforts to protect and promote population health (Figure 4). The concepts for the framework are drawn from Hall et al. (2012). The set of processes for public health surveillance include: 1) planning and system design, 2) data collection, 3) data analysis, 4) interpretation of results of analysis (i.e., generation of information), 5) dissemination and communication of information, and 6) application of information to public health programs and practice. The last step of applying the data to guide efforts to protect and promote population health is essential. Specific for birth defects surveillance, the key objectives focus on using the data for on-going monitoring of prevalence and trends, epidemiologic and etiologic research, health service utilization, service intervention, and evaluation of prevention strategies. Data utility also provides an important feedback loop to data processing to ensure that the data collected are relevant. Against this backdrop, the five areas (considerations) identified in CDC's vision for public health surveillance in the 21st century must be addressed to ensure effective and efficient surveillance practice.

Figure 4. Public Health Surveillance Framework for Monitoring Birth Defects



III. Methods

IIIa. Study Design and Data Sources

A mixed methods approach using three data sources (TABLE II) is employed to address the study questions. The first data source is the National Birth Defects Prevention Network (NBDPN) Program Survey that was self-administered by state birth defects program directors/ managers; for this DrPH research study, an emphasis is placed on the data focusing on: 1) characteristics of population-based birth defects surveillance systems, 2) intended purposes of birth defects surveillance systems, and 3) major surveillance considerations (lexicon and standards, information science and technology, skilled workforce, data access and use, and data analytic foundation) as they relate to birth defects. A second data source is a directory of state

TABLE II. STUDY QUESTIONS, DATA SOURCES AND ACCESS

Study Questions	Data Sources	Data Access
• What are the characteristics, e.g., methods and infrastructure, of population-based birth defects surveillance systems in the United States?	• NBDPN Program Survey • NBDPN Annual Report Directory	Secondary data sources: • Access to NBDPN Program Survey results through SurveyMonkey.com • Access to an Excel output file of the NBDPN Annual Report directory information Primary data collection: • Notes from semi-structured interviews
• To what extent are population-based birth defects surveillance programs in the U.S. addressing the intended purposes of the systems?	• NBDPN Annual Report Directory • Semi-structured interviews	
• How prepared are birth defects surveillance programs to address the major considerations identified in "CDC's Vision for Public Health Surveillance in the 21st Century"?	• NBDPN Program Survey • Semi-structured interviews	

birth defects surveillance programs that was collected for the 2012 NBDPN Annual Report. Lastly, a third data source consists of qualitative data from semi-structured interviews with selected surveillance program directors.

IIIb. Data Collection and Management

The NBDPN Program Survey was developed by an ad-hoc NBDPN workgroup, and feedback on the questions was solicited from the NBDPN Executive Committee and selected members. The questions were entered into SurveyMonkey (www.surveymonkey.com) and piloted by the ad-hoc NBDPN workgroup members. After the initial pilot, the survey was e-mailed to all state primary contacts listed in the NBDPN 2011 Annual Report (NBDPN, 2011), and data collection occurred between January 13 – February 15, 2012. E-mail and phone call reminders were sent to state programs. During the final data cleaning process, 10 state programs were followed-up to clarify responses and/or asked to complete the survey to ensure a complete response from all programs in the U.S. The survey included 55 questions that focused on: operational status of the state surveillance system, catchment area, case ascertainment methodology, public health authority (legislative or administrative), staffing, funding sources, data analysis software, geocoding practice, family history information, potential for follow-up studies, data sources and reporting, impact of electronic medical records, coding practice, staff, data quality, health service utilization, priority areas, and challenges for surveillance systems (see Appendix C for the Survey).

The responses that were collected from SurveyMonkey were exported to a CSV file, which required recoding to ensure a seamless import into SAS 9.3 for analysis. Additional

cleaning was performed and some responses were collapsed, when applicable, in the final data presentation.

Data from the second source (NBDPN Annual Report Directory) were collected via a web-based interface that was designed to provide metadata about the surveillance programs, with the primary purpose of collecting the information for 2012 NBDPN Annual Report Directory that was published in Birth Defects Research Part A (NBDPN, 2012). The data collection was coordinated by the NBDPN Data Committee, and a request was sent to all state birth defects contacts to update their program information via a state-specific, password-protected webpage. This survey included metadata on program status, population-covered, legislation, case definition, surveillance methods, case ascertainment, data collected, data collection methods and storage, data analysis software, data quality assurance, system integration, funding, and program contacts. The results were exported to a CSV file from the SQL server. The variables were renamed to ensure a proper export to SAS for comparative analysis with the data collected from the NBDPN Program Survey.

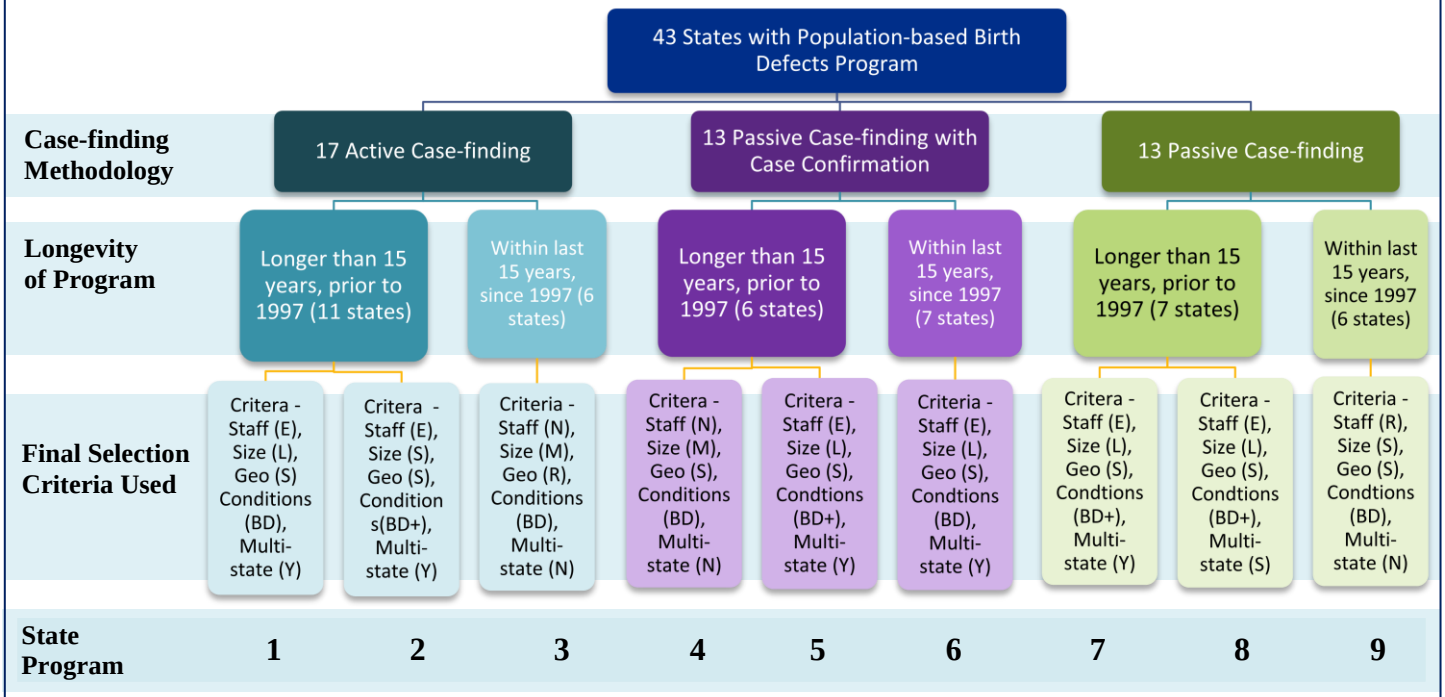
The final data source was obtained from semi-structured interviews to further probe the issues identified in the CDC Strategic Framework for Public Health Surveillance in the 21st Century," and the purpose of birth defects programs. Since some of the questions asked in the NBDPN Program Survey cover aspects of the issues raised in the CDC Strategic Framework, responses were used to help inform the semi-structured interviews (see TABLE III).

TABLE III. CROSSWALK BETWEEN ISSUES LISTED IN CDC STRATEGIC FRAMEWORK FOR PUBLIC HEALTH SURVEILLANCE AND CORRESPONDING NBDPN PROGRAM SURVEY QUESTIONS (APPENDIX C)

Issues (Considerations)	Corresponding NBDPN survey questions
Lexicon and standards	Definitions (Questions 19-21)
Information science and technology	Electronic Medical Records (Questions 37-41)
Skilled workforce	Staff (Question 17)
Data access and use	Laws, Regulations, or Authority (Questions 6-15)
Strong data analytic foundation	Analytic Software and Geocoding (Question 22-25)

The semi-structured interviews were conducted with nine program directors. Guest, Bunce, and Johnson (2006) found that saturation ("point at which no new information or themes are observed in the data") occurred within the first 12 interviews, but as few as six individuals may be sufficient to get high-level themes. Purposive sampling of state programs was used to obtain a diverse perspective. Figure 5 shows a stratification scheme for state selections. First, states with an operational birth defects surveillance program were grouped by their primary case-finding approach: active, passive with case confirmation, or passive without case confirmation (three groups). A second level further stratified the programs by whether they were established before or after 1997 (six groups); the year was chosen to distinguish newer programs from their more established counterparts, given the enhanced national efforts at that time for birth defects surveillance with the creation of the NBDPN and increased funding from CDC to support state-based birth defects surveillance. These six groups were again stratified so that each primary case-finding approach was correlated to two established and one newer program (a total of nine programs); the criteria used to ensure diversity in programs in this final selection included core

Figure 5: Stratification Scheme for Semi-structured Interview Selection



Secondary criteria used for final state selection:
Staff - core staff experience in birth defects surveillance:
N – new; R- recent; E - experienced
Size – size of live birth (LB) population for the catchment area:
S - less than 50,000 LB; M - 50,000 to less than 100,000 LB; L – greater than 100,000 LB
Geo - geographic coverage:
S -State-wide; R - regions (selected counties)
Conditions – conditions ascertained:
BD - major birth defects; BD+ - major birth defects and other conditions
Multi-state - engagement in multi-state data collaborations:
Y - Yes; N - No; S – Somewhat

staff experience in birth defects surveillance, live birth population size for the program's catchment area and whether it is state-wide or selected counties, case inclusion beyond structural and genetic conditions, and engagement in multi-state data collaborations.

TABLE IV lists the targeted states by selection criteria. Some of the information in the table is based on the 2012 NBDPN Annual Report Directory. A full list of state programs with selected program characteristics is listed in Appendix A.

Written notes were taken using AudioNotes (http://luminantsoftware.com) during the semi-structured interviews, which lasted for approximately 90 minutes. The notes were transcribed in AudioNotes and converted to plain text files for import into Atlas.ti, a qualitative analysis software (http://www.atlasti.com).

TABLE IV. SELECTION CRITERIA FOR SEMI-STRUCTURED INTERVIEWS

State	TX	IA	MN	MD	MI	FL	IL	NY	VT
Case Finding[1]	A	A	A	P	P	P	P+	P+	P+
Longevity of System[2]	E	E	N	E	E	N	E	E	N
Core Staff Experience in BD Surveillance[3]	E	E	N	N	E	E	E	E	R
Size of Live Birth Population[4]	L	S	M	M	L	L	L	L	S
Geographic Coverage[5]	S	S	R	S	S	S	S	S	S
Outcomes Covered[6]	BD	BD+	BD	BD	BD+	BD	BD+	BD+	BD
Multi-state Data Collaborations[7]	Y	Y	N	N	Y	Y	S	Y	N

[1]Case-finding methodology: A - Active; P+ - Passive + case confirmation; P- Passive
[2]Longevity of program: E- Longer than 15 years (prior to 1997); N - Within last 15 years (since 1997)
[3]Core Staff Experience in Birth Defects Surveillance: N – new; R- recent; E - experienced
[4]Size of Live Birth Population: S - less than 50,000 LB; M - 50,000-less than 100,000 LB; L - 100,000 + [4]Live birth (LB)
[5]Geographic Coverage: S -State-wide; R - regions (selected counties)
[6]Outcomes Covered: BD - major birth defects; BD+ - major birth defects + other conditions
[7]Engagement in Multi-state Data Collaborations: Y - Yes; N - No; S – Somewhat

IIIc. Analysis Methods

The NBDPN Program Survey data were analyzed in SAS. The output CVS file from SurveyMonkey was first restructured and fields were renamed so that the survey could be imported into SAS. A data codebook was developed in Excel that contains all questions and their responses to guide data analysis.

Descriptive analyses, including frequency of responses, were performed in SAS, and responses were also examined by state and compared to the information in the NBDPN Annual Report Directory (NBDPN, 2012). TABLE V provides an example of the mapping that was completed. There were some overlap between the two data sources; although some of the questions were similar in content, their responses were recorded differently. For example, Q3 in the NBDPN Program Survey allowed respondents to select an answer from a list of specific defects, whereas the NBDPN Annual Report Directory offered an open-ended text field as a response to a similar question. Also, the NBDPN Program Survey included expanded questions for some of the topics, e.g. legislation, as well as questions on programmatic changes over time that were not included in the NBDPN Annual Report Directory.

All the data presented in the tables were obtained from the NBDPN Program Survey except for the table on intended purposes of surveillance programs, which was obtained from the NBDPN Annual Report Directory. For each variable examined, a ratio was calculated by dividing the percent of active case-finding programs by percent of passive case-finding programs. Also, p values were calculated using Fisher's exact test of independence.

TABLE V. QUESTION MAPPING FROM 2012 NBDPN PROGRAM SURVEY TO ANNUAL REPORT DIRECTORY OF STATE BIRTH DEFECTS SURVEILLANCE PROGRAMS

2012 Program Survey	NBDPN Annual Report Directory of State Birth Defects Surveillance Programs
Q1. What is the current status of your population-based birth defects surveillance system in your state or territory? (We mean a system that uses more than birth certificates for case identification).	Section 1 - General Program Status
Q2. What proportion of your state's or territories' birth population is covered by your state's birth defect surveillance system (e.g.: statewide = 100%)	Section 1 - General Birth Population: Is it Statewide?
Q3. Which birth defects does your surveillance system currently identify? • Checklist of potential responses: e.g. NBDPN list less than 46 conditions, Critical Congenital Heart Defects (CCHD) list, Environmental Public Health Tracking (EPHT) list of 12 conditions, Developmental conditions	Section 2 - Case Condition Conditions covered • Open-ended text field completed by state program contact
Q4: Which pregnancy outcomes does your surveillance system categorize? Checklist of potential responses.	Section 2 - Case Condition Pregnancy outcomes
Q5: Does the legal public health authorization language in your state or territory explicitly mandate reporting of birth defects or authorize the program to identify and collect information from health facilities? Q6: If yes, was the authority established through... Q7: Does your state or territory have any laws, regulations, or authority that negatively impact the surveillance function of case record abstraction?	Section 1 - General Legislation: if legislation or rule, cite the law and year enacted.

The information gathered from the nine semi-structured interviews was coded by C. Mai (primary coder) for themes using a grounded theory approach. First, the primary coder read the interview notes twice and then developed a codebook from the transcribed information focusing on intended purposes of birth defects surveillance programs and the major considerations outlined in the CDC Strategic Framework for Public Health Surveillance (see Appendix E). The

themes were then marked in Atlas.ti for each interview. A second coder used the developed codebook to independently code three of the nine interviews in Atlas.ti. The primary coder examined the results from both coders for the three overlapping interviews and made changes, as needed, to the primary coded files. The primary coded files were used to write the study findings.

IIId. Validity Considerations

Yin (2009) stressed the importance of using "multiple sources of evidence or triangulation in order to encourage convergent lines of inquiry." The mixed methods approach using various data sources should help minimize validity concerns. The primary data source, NBDPN Program Survey, is self-administered by the primary contact person for each state birth defects program. Some of the questions are complex and multi-layered, and misinterpretation is possible. For example, the survey attempted to determine the number of major birth defects ascertained by a state program, but several respondents misunderstood the categories and did not realize that the Environmental Public Health Tracking (EPHT) list of 12 conditions is a subset of the NBDPN list of 47 conditions; therefore, NBDPN list might have been selected without selecting the EPHT list. The use of a second survey dataset, NBDPN Annual Report Directory, along with follow-up to obtain clarification on states' survey responses increased the validity of findings for this research study. Additionally, the semi-structured interviews provided further insights and possible alternative explanations for changes affecting the practice of birth defects surveillance.

Other threats to validity include selection and interviewer bias. An attempt was made to minimize bias with purposive sampling and a priori selection of states. The states were selected using multiple criteria to increase diversity in respondents. Another consideration was the

duration of the data collection. The period from initial data collection (January/February 2012) to final data cleaning and write-up (late summer/fall 2013) could have produced some programmatic changes. For the four states that completed the NBDPN Program Survey a year and half later, they were asked to respond to their program as of 2012 to help minimize the changes.

IIIe. Institutional Review Board (IRB)

IRB determination was submitted to both UIC Office for the Protection of Research Subjects and CDC National Center on Birth Defects and Developmental Disabilities Human Subject Office. UIC granted the project an exemption status on March 1, 2013 (UIC Research Protocol # 2013-0179), and CDC determined that the project was non-research (public health practice: surveillance).

IV. Results

IVa. Intended Purposes of Population-based Birth Defects Surveillance Programs

Information on the purposes/objectives for population-based birth defects surveillance programs were obtained from the NBDPN Annual Report Directory. All programs engaged in the following monitoring activities: generating prevalence data, examining trends, and/or performing cluster investigations as warranted. More active case-finding programs indicated using their data for ecological and etiological studies (77% vs. 65%) and for planning/services (94% vs. 65%), whereas more passive case-finding programs indicated referral of children with affected conditions to medical and social services in the community.

To further understand the intended purposes of the programs, the semi-structured interviews with nine program directors included a question about the original purposes of their

programs and how their programs have changed through the years. The major themes that emerged regarding the impetuses of their respective programs were: 1) an inability to satisfactorily address community concerns owning to a lack of data on birth defects, 2) the availability of new funding opportunities, and 3) a need to help affected children and their families. Partnership engagement played a key role in establishing and shaping the directions of the programs. One interviewee said that, "They [i.e., partners] have been very involved in what are the priorities and activities of the registry. They have been there when we had some setbacks with regards to funding; we had a few years where funding was reduced and we had to reset our priorities and they were there to help guide us as we made decisions about what we could and couldn't do with reduced funding. They advocated on behalf of the registry and actually last year that funding was restored."

Given the range of possible uses of quality population-based data beyond on-going monitoring, programs have to strategically prioritize their efforts. One interviewee stated, ""When I came [to the program], I always tried to have contacts with MCH because I used to work there. My vision is that we are not just here for landfills, like the early studies. We are here as part of a whole health department. We started to provide data to other programs, like HIVs, early interventions, etc. Over time, we have expanded. It also depends on where you get the money." Another interviewee noted, "When most people see tables with counts, etc., they don't think about whether where the data come from. They just want to see the outputs." This sentiment was noted often by program directors. They realize the forces driving the establishment of their program, such as cluster investigations or provision of services for families, but in the end, the program can only remain relevant if the data are utilized to guide public health actions and serve families and communities.

TABLE VI. REPORTED OBJECTIVES OF POPULATION-BASED BIRTH DEFECTS SURVEILLANCE PROGRAMS BY CASE-FINDING STATUS

Purpose	Objectives	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)		Ratio[1]	P value[2]
		#	%	#	%		
Surveillance	• Develop timely baseline birth defects rates • Monitor trends and relationships to environmental factors • Perform cluster investigations	17	100 %	26	100 %	1.0	
Research	• Provide basis for ecologic and etiologic studies (research)	13	77%	17	65%	1.2	0.51
Planning / Prevention	• Provide data for services planning • Provide basis for prevention strategies • Evaluate efficacy of preventive services	16	94%	17	65%	1.4	0.06
Healthcare and human services	• Refer children to services and resources • Evaluate services utilization	11	65%	20	77%	0.8	0.49

[1]Ratios are calculated by dividing the % of active case-finding programs by % of passive case-finding programs.
[2]P values are calculated using Fisher's exact test.

IVb. Manuscript 1: Public Health Practice of Population-based Birth Defects Surveillance Programs in the United States

Prepared for submission to the Journal of Public Health Management and Practice

Public Health Practice of Population-based Birth Defects Surveillance Programs in the United States

Author names (with academic credentials)
Affiliations (including title(s), department, and name and location of institutions of primary employment)

Author Biography. An author biography (a brief autobiographical sketch from each author including pertinent education and work experience, in 75 words or less

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The findings and conclusions in this study are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

The material is based upon a Dissertation, submitted in partial fulfillment of the requirements for the doctoral degree at the School of Public Health of the University of Illinois at Chicago.

Abstract

Context: Birth defects remain a leading cause of infant mortality in the United States (U.S.) and contribute substantially to health care costs and life-long disabilities. The World Health Assembly adopted a resolution highlighting the importance of addressing birth defects given their impact on infant and child morbidity and mortality.

Objective: To understand the current practice of U.S. population-based birth defects surveillance.

Design: The National Birth Defects Prevention Network (NBDPN) conducted a survey of U.S. population-based birth defects activities that included questions about operational status, case ascertainment methodology, program infrastructure, data collection and utilization, as well as priorities and challenges for surveillance programs. Birth defects contacts in the U.S., including District of Columbia and Puerto Rico, received the survey via e-mail; follow-up reminder via e-mails and phone calls were made to ensure 100% response from all programs.

Results: Forty-three states perform population-based surveillance for birth defects, covering approximately 80% of the live births in the U.S. Seventeen primarily employ an active and 26 employ a passive case-finding approach. These programs all monitor major structural malformations; however, passive case-finding programs more often monitor a broader list of conditions, including developmental conditions and newborn screening conditions. Active case-finding programs are more likely to use clinical reviewers, cover broader pregnancy outcomes, and collect more extensive information, such as family history. Overall, many programs reported an ability to conduct follow-up studies of children with birth defects during infancy and early childhood with a few able to follow-up to adulthood.

Conclusions: The breadth and depth of information collected at a population level by birth defects surveillance programs in the U.S. serves as an important data source to guide public health action. Collaborative efforts at state and national levels can help harmonize data collection and increase utility of birth defects programs.

KEY WORDS: birth defects, surveillance, public health practice, population-based

Birth defects are common, affecting one in every 33 babies in the United States (U.S.); costly, over $2.6 billion spent on just hospital costs in a given year; and deadly, contributing to one in every 5 infant deaths.[1] These are conditions that: 1) result from a malformation, deformation, or disruption in one or more parts of the body; 2) are present at birth; and 3) have a serious, adverse effect on health, development, or functional ability.[2] Although the U.S. lacks a unified national population-based surveillance system to collect data on major birth defects, most states currently have a program to monitor these conditions.[3] The first state statute that established a program to capture birth defects from reporting sources was enacted in New Jersey in 1926. However, the proliferation of systems in the U.S. to conduct population-based birth defects surveillance did not occur until the last few decades as a response to: community concerns about environmental exposures, such as Thalidomide;[4-6] evaluation of prevention strategies, such as folic acid fortification; and referrals of affected children and families to medical and social services.[7]

In May 2010, the 65th World Health Assembly adopted Resolution WHA 63.17 that highlighted the importance of surveillance, research, prevention, and intervention actions to address birth defects given their impact on infant and child morbidity and mortality.[8] The resolution called upon member states to "develop and strengthen surveillance systems for birth defects in order to have accurate information available for making decisions on prevention and control of these birth defects and to continue providing care and support to individuals affected by birth defects." In the U.S., the National Birth Defects Prevention Network (NBDPN) was established in 1997 as a national organization to address birth defects surveillance, research, and prevention by maintaining a network of state and population-based birth defects programs (www.nbdpn.org). The NBDPN publishes an annual data report that includes state-specific

prevalence data on 47 birth defects and an accompanying directory containing a descriptive metadata profile of each state/territory program.[3] The information in the directory has mainly been used to understand the data collection methodology of each program. However, it has been 20 years since a detailed assessment of birth defects programs in the U.S. was last conducted by CDC.[9] The purpose of this study is to describe the current practice and approach to collecting population-based birth defects data across the U.S.

Methods

The National Birth Defects Prevention Network (NBDPN) conducted a survey of population-based birth defects activities in the U.S. that included questions about birth defects surveillance status, case ascertainment methodology, program infrastructure, data collection and utilization, as well as priorities and challenges for surveillance programs. The survey questions were piloted by several state programs and then entered into SurveyMonkey™ (www.surveymonkey.com). The survey was then e-mailed to birth defects contacts in the 50 U.S. states, District of Columbia, and Puerto Rico in January 2012, with periodic e-mail reminders sent to the birth defects contacts. During the final data cleaning stage in fall 2013, the lead author (C. Mai, CDC liaison to NBDPN) contacted states that did not complete the survey or whose answers required clarification to ensure completed responses from all programs. Survey responses were also cross-checked with any available information from NBDPN data report's annual directory and discrepancies were resolved by checking the information with program staff or existing programmatic materials.

The data collected were exported to SAS 9.3 for cleaning and analysis (SAS Institute Inc, Cary, NC). Descriptive analyses were performed by stratifying the 43 operational programs by

their primary case ascertainment methodology (active or passive case-finding) and calculations (ratio and p values) were performed between the two case-finding approaches. The open-ended responses to the survey questions regarding the three areas or activities of highest priority and three most important challenges for the program were reviewed and manually coded using categories created from the responses.

The study protocol was reviewed and approved by the University of Illinois at Chicago institutional review board (IRB, protocol # 2013-0179) and by CDC's National Center on Birth Defects and Developmental Disabilities human subject protection office.

Results

Of the 50 U.S. states, District of Columbia, and Puerto Rico surveyed in this study, 43 indicated that they conduct population-based surveillance for birth defects; the total geographic area covered by these 43 programs include approximately 80% of the live births in the U.S. Thirty-nine of these programs were consistently operational (on-going) and captured all births within their state catchment areas except for California, Georgia, and Minnesota (Figure 1). Three state programs conducted birth defects surveillance but data collection was not always done routinely and one state restarted its surveillance program after an organizational transition. Of the remaining nine states, three were planning to develop a program, and six indicated no birth defects surveillance activities.

Public health surveillance programs are sometimes distinguished by their case-finding approaches, whether programmatic staff collect the primary data for the conditions of interest or passively receive information from data sources. Of the 43 population-based surveillance programs that collect birth defects data, 17 programs employ an active case-finding methodology

where staff are sent to hospitals and provider offices to perform primary collection of medical information and birth defects data while 26 programs predominately use a passive case-finding approach that relies on reported data from providers or administrative datasets where programs' staff may or may not perform definitive case confirmation of the information.

Table 1 provides funding sources and methodology used by state programs. The top three funding sources include state general funds, Federal Title V block grant, and CDC birth defects cooperative agreements. Programs on average rely on two funding sources, with one state obtaining funds from greater than three sources for core surveillance activities (data not shown). These programs all monitor major structural malformations; however, more passive case-finding programs cover a broader list of conditions, including developmental conditions (23.1% compared to 11.8%), newborn/infant hearing (38.5% compared to 5.9%) and newborn screening conditions (42.2% compared to 11.8%). All programs include live births but more active case-finding programs include other pregnancy outcomes, most notably for pregnancy outcomes less than 20 weeks gestation (52.9% compared to 11.5%) and pregnancy terminations at any gestation (76.5% compared to 15.4%).

Table 2 presents information on coding, quality procedures for case confirmation, and abstraction practices of the surveillance programs. The disease classification system used by the majority of passive case-finding programs (92.3%) is the International Classification of Diseases, Clinical Modification, Version 9 (ICD-9-CM) while the active case-finding programs uses CDC's more detailed, expanded coding structure of the British Paediatric Association modification of ICD-9-CM (CDC/BPA). The active case-finding programs predominately use trained data abstractors and clinician reviewers to code birth defect cases. Most of these case-finding programs routinely abstract both maternal (82.4%) and fetus/infant (100%) medical

records at delivery and tertiary hospitals. However, only 2 programs routinely request medical records from the mother's obstetric care providers for all or selected conditions.

Given the complexity in the case definition for selected birth defects, surveillance programs use various strategies to ensure accuracy of the conditions collected. The most common strategies employed are medical/record review of the documentation and data quality assurance performed by program staff (31 of 43 operational programs). In addition, programs that use active case-finding more often use clinical reviewers, such as dysmorphologists (board-certified pediatricians who specialize in birth defects) (35.3% compared to 3.8%) and geneticists (70.6% compared to 15.4%). Access to medical records is often done through secure file transfers for active case-finding programs while passive case-finding programs use web-based health information ports or internal health department electronic uploads and/or transactions.

Each program collects a set of demographic and clinical information on infants with birth defects. Table 3 focuses on selected data elements collected by surveillance programs that are beyond the basic demographic and clinical information. Most programs have geocoded data and collect maternal residency at date of delivery. Very few programs are collecting maternal residency at date of conception or during the pregnancy time period and even fewer are conducting specific prenatal surveillance to identify potential cases of birth defects as the pregnancy progresses. More active case-finding programs routinely collect and record information on family history. Reasons cited by programs for not collecting family history information include a lack of legislative or other authority and lack of data collection methodologies requisite to the task.

Many surveillance programs can conduct follow-up studies of children with birth defects during infancy and early childhood (up to 5 years of age), with a few able to follow-up to adulthood (up to 18 years of age) (Table 3). Fifteen programs can access or link to cost/charge data during the first year of the child's life while nine can follow-up beyond the first year of life. Furthermore, 12 programs are able to access or link to health care service data during the first year of life, and 6 programs can follow-up beyond the first year of life. However, when asked if they utilize cost or charges data, only 6 programs indicated its use for economic analysis, 6 do so for program planning, 2 do so for program planning, 3 do so for needs assessment, and 6 do so for legislative requests.

Finally, 38 out of 43 birth defects surveillance programs responded to the question asking them to identify three areas or activities of highest priority, and 39 programs responded to the question asking them to identify three most important challenges. Using created categories that were based on manually coding the participants' responses, the activities of highest priority are case ascertainment/data quality improvements (76%), utilization of data for referrals or prevention (39%), and data dissemination (21%). The most important challenges of programs include funding/sustainability (72%), staffing issues (38%), and data quality/data system improvements (38%).

Discussion

During the past 40 years, the number of birth defects programs in the U.S. has increased from 3 programs in the early 1970s to 43 programs by 2013. In 1994, Lynberg and Edmonds published a comprehensive review of these programs, and reported that of the 23 operational programs during that time, seven states used active and 16 used passive case-finding

methodology. Since then, 20 additional states have established surveillance programs, with half of the new programs primarily using an active case-finding approach and the other half using a passive case-finding approach.

Various factors, such as resources and legislation, affect the approaches employed by population-based birth defects surveillance programs. A dichotomous category is used to classify the case-finding approaches in presenting the results in this study, but it should be recognized that many of these programs incorporate varying strategies that fall on a continuum of programmatic interventions to ensure the accuracy and completeness of data collection for the population ascertained. However, to understand general characteristics of these programs, it is useful to examine their case-finding approaches, since the data collected during this stage form the basis of the database for birth defect surveillance. The passive multiple source case-finding approach used by 60% of the birth defects surveillance programs in the U.S. obtains primary case information from hospital/provider reporting and/or administrative datasets, such as hospital discharge data. This approach offers several benefits, including the ability for a surveillance program to cover a broad list of conditions while considering resource constraints and potential improvement in timeliness that can be important for referring affected individuals to medical and social services. All of the birth defects surveillance programs collect data on structural malformations, but more passive case-finding programs ascertain additional conditions, such as developmental disabilities, newborn/infant hearing, and newborn genetic and metabolic screening. The resources required by a surveillance program to receive the information from existing data sources enable a program to cover a range of conditions. A concern for this approach is the accuracy of the information reported or obtained from administrative datasets. Salemi et al.[10] found that the Florida program could increase its positive predictive value and

generate more accurate prevalence estimates with the addition of case verification to their passive case-finding program.

Additionally, access to specialized medical experts to assist surveillance programs with enhanced case review, disease coding, and clinical classification can improve the accuracy of case information given the complexity of some of the birth defects conditions collected. This is especially important for the programs that use a more detailed coding system. Most of the specialized clinical reviewers, such as dysmorphologists, geneticists, and cardiologists, work for a birth defects program with active case-finding methodology where more detailed medical information on the cases is often captured in verbatim text in the database. Lin et al.[11] discussed the role that clinicians can play in providing not only diagnostic interpretations of the abstracted medical information but also in data interpretation for cluster analyses and research. Resource constraints can sometimes limit a program's access to clinicians. Lin et al.[11] found that the birth defects surveillance programs that have clinical support mainly employ the clinicians part-time or as consultants.

In addition to the demographic and case information collected by surveillance programs, this study focused on examining expanded data collection elements. As shown in Table 3, most programs have access to geocoded data and collect data on maternal residency at date of delivery. The number of programs with geocoded case data is similar to those reported by Ying et al.[12] However, only a few programs with active case-finding approaches conduct prenatal surveillance to identify potential cases of birth defects that are prenatally diagnosed as the pregnancy progresses, which is consistent with the pregnancy outcomes included in the surveillance program. Since the active case-finding programs rely on their own staff for primary data collection, they are able to collect more comprehensive data about the cases. The majority

(70.6%) were able to routinely collect and record family history information. This data can be useful to examine recurrence of selected birth defects and help inform prevention strategies. Additionally, many of these programs reported a capacity to conduct follow-up studies of children beyond infancy.

As the life expectancy for children born with birth defects increases,[13-16] population-based birth defects surveillance data can be used to better understand health outcomes and service utilization of these children. Approximately one third of the programs reported the ability to access or link their birth defects data to cost and/or health care utilization information during a child's first year of life; but very few are currently using the cost or charge data for program planning, needs assessment, or legislative requests. Increased utilization of these types of data will assist programs to better understand the financial and social burden of birth defects for the state and on local communities.

This study has a number of strengths. The response rate is 100% of all operational population-based surveillance programs in the U.S. This offers a current snapshot of the practice of birth defects surveillance programs in the U.S. Steps were taken to validate the data provided in the survey with the information in the NBDPN annual report and programmatic materials as well as to follow-up with state programs.

A number of weaknesses should be noted. First, the survey was self-administered and as such, was subject to programmatic interpretations of the questions and categorical responses. Second, data collection and cleaning of the survey lasted about one and one-half years and programmatic changes could have occurred during that time period. Although some changes could be expected, the overall effort in conducting birth defects surveillance should be relatively stable. Third, birth defects programs can vary in their data collection approaches, which might

not be captured well in the survey. This was evidenced in the dichotomous grouping of the programs by primary case-finding status. Given the range of data sources and intensity in ascertaining the information, some of the passive case-finding programs have steps in place to perform active verification of the reported case information. However, the case-finding categories can be useful to examine overall activities in the U.S. The NBDPN reports on estimating national estimates for birth defects stratify the data by case-finding strategies.[17,18]

Conclusion

Population-based birth defects surveillance activities in the U.S. have increased during the past few decades and continue to evolve to address community concerns about the impact of birth defects. The modern concept of the scope of surveillance was reframed in the 1960s to address newfound public health concerns for both communicable and chronic diseases by systematically collecting and evaluating pertinent data and disseminating the information for public health action.[19] The use of population-based surveillance for birth defects mirrors that of the public health community to conduct population-based surveillance for chronic conditions, such as cancer, where the impact can be tremendous for affected populations. The data collected offers a rich source of information to guide public health practice and improve the lives of families and communities.

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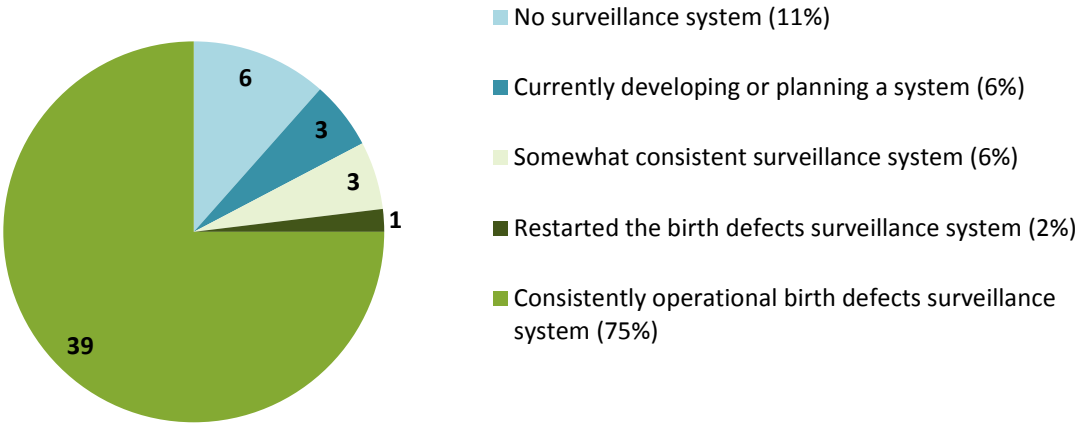
Figure 1. Status of State Birth Defects Surveillance Programs, 2012 (50 states, DC, and PR)



State-wide coverage for 39 operational systems except for the following states:
CA - Covers about 70,000 live births (LB) annually in 2 regions
GA - Covers about 35,000 LB annually in the metropolitan Atlanta counties
MN - Covers around 80% of the state population (about 70,000 LB annually)

Table 1: Population-based Birth Defects Surveillance Program Structure and Methodology by Case-finding Status

Program Structure	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)		Ratio[1]	P value[2]
	No.	%	No.	%		
Funding[3]						
Title V MCH /SSDI	9	52.9%	16	61.5%	0.9	0.75
State General Funds	8	47.1%	9	34.6%	1.4	0.53
CDC birth defects surveillance	6	35.3%	8	30.8%	1.2	1.001.(
CDC environmental public health tracking (EPHT)	2	11.8%	6	23.1%	0.5	0.45
University/Academia	2	11.8%	0	0.0%	---	0.15
State fees, e.g. vital statistics, newborn screening, or dedicated fund	1	5.9%	6	23.1%	0.3	0.21
Other sources	1	5.9%	3	11.5%	0.5	1.00
Conditions ascertained[4]						
Structural malformations (all 46 birth defects on NBDPN list)	10	58.8%	18	69.2%	0.9	0.53
Structural malformations (less than 46 birth defects on NBDPN list)	7	41.2%	8	30.8%	1.3	
Developmental disabilities	2	11.8%	6	23.1%	0.5	0.45
Newborn/infant hearing	1	5.9%	10	38.5%	0.2	**0.03**
Newborn genetic and metabolic screening	2	11.8%	11	42.3%	0.3	**0.04**
Pregnancy outcomes covered[5]						
Live births	17	100.0%	26	100.0%	1.0	
Fetal deaths	15	88.2%	15	57.7%	1.5	**0.04**
Miscarriages (spontaneous abortions, <20 wks gestation)	9	52.9%	3	11.5%	4.6	**0.01**
Stillbirths (> 20 wks gestation)	15	88.2%	15	57.7%	1.5	**0.04**
Pregnancy terminations (any gestation)	13	76.5%	4	15.4%	5.0	**<.001**

No. – number of programs; % - percent; Title V MCH/SSDI – Title V Maternal and Child Health Services Block Grant/State Systems Development Initiative; CDC – Centers for Disease Control and Prevention; NBDPN – National Birth Defects Prevention Network

[1]Ratios are calculated by dividing the % of active case-finding programs by % of passive case-finding programs for the category.

[2]P values are calculated using Fisher's exact test.

[3]Multiple responses allowed for each type of funding source.

[4]Multiple responses allowed for conditions beyond structural malformations.

[5]Multiple responses allowed for each type of pregnancy outcome.

Table 2: Birth Defects Data: Coding, Quality Procedure for Case Confirmation, and Abstraction Practices

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)		Ratio[1]	P value[2]
	No.	%	No.	%		
Disease classification coding system utilized						
ICD-9-CM	8	47.1%	24	92.3%	0.5	**<.003**
CDC/BPA (6 digit code) or modified	15	88.2%	6	23.1%	3.8	**<.001**
ICD-10	2	11.8%	6	23.1%	0.5	.44
Person responsible for assigning a disease classification code to a birth defects case						
Data abstractor	11	64.7%	6	23.1%	2.8	**.01**
Certified hospital coder (e.g. Registered Health Information Technicians - RHITs or Registered Health Information Administrator - RHIA)	1	5.9%	9	34.6%	0.8	.06
Trained disease coder	4	23.5%	7	26.9%	0.9	1.00
Clinician or clinical reviewer	9	52.9%	6	23.1%	2.3	**.05**
Epidemiologist	0	0.0%	1	3.8%	---	1.00
Background of data abstractor or other staff who review medical records for case identification or case verification						
Health information management technology with RHIT/RHIA credential	10	58.8%	2	7.7%	7.7	**<.001**
RN-Nursing or Nurse Consultant	13	76.5%	2	7.7%	10.0	**<.001**
Other health care professional	8	47.1%	1	3.8%	12.2	**.001**
None, trained in-house	4	23.5%	8	30.8%	0.8	.73
Data quality procedures utilized to assess accuracy of the birth defects case status (a true case)						
Dysmorphologist clinical reviewer	6	35.3%	1	3.8%	9.2	**.01**
Geneticist clinical reviewer	12	70.6%	4	15.4%	4.6	**<.001**
Cardiologist clinical reviewer	9	52.9%	3	11.5%	4.6	**.005**
Pediatric clinical reviewer (on the personnel list)	4	23.5%	3	11.5%	2.0	.41
Medical records or health records review of the documentation	14	82.4%	12	46.2%	1.8	**.03**
Quality of the data source (e.g. pathology, cytogenetic lab, genetics clinic, specialty clinic, etc)	11	64.7%	11	42.3%	1.5	.22
Corroborating procedure that is linked to the diagnosis	11	64.7%	8	30.8%	2.1	**.03**
Data quality assurance procedure performed by staff	14	82.4%	17	65.4%	1.3	.30
Other, e.g. electronic edits, re-abstraction, etc.	3	17.6%	6	23.1%	0.8	1.00
Surveillance program with the ability to classify birth defect cases into isolated, multiple, and syndromes (not using disease codes)						
No	5	29.4%	14	53.8%	0.6	.005
Yes	7	41.2%	3	11.5%	3.6	
Sometimes	4	23.5%	6	23.1%	1.0	

Abstraction Practices						
Data abstractors go to the delivery and tertiary hospitals to abstract the medical records of the fetus and/or infant						
Yes; consistently for all birth defects or conditions reportable to the program	17	100.0%	3	11.5%	8.7	**<.001**
Yes; consistently for selected birth defects or conditions	---	---	4	15.4%	---	
Yes; consistently for selected data sources	---	---	2	7.7%	---	
Sometimes; for selected conditions; selected data sources; or for special projects	---	---	3	11.5%	---	---
No or rarely, only as required (e.g. community investigations)	---	---	14	53.8%	---	**<.001**
Data abstractors go to the delivery hospital to abstract the medical records of the mother						
Yes; consistently for all birth defects or conditions reportable to the program	14	82.4%	2	7.7%	10.7	**<.001**
Yes; consistently for selected birth defects or conditions	0	0.0%	2	7.7%	0.0	
Sometimes; for selected conditions; selected data sources; or for special projects	2	11.8%	1	3.8%	3.1	---
No or rarely, only as required (e.g. community investigations)	1	5.9%	21	80.7%	0.1	**<.001**
Data abstractors request medical records from the mother's obstetric care provider to obtain additional information on the mother's pregnancy						
Yes; consistently for all birth defects or conditions reportable to the program	1	5.9%	0	0.0%		.15
Yes; consistently for selected birth defects or conditions	1	5.9%	0	0.0%		
Sometimes; for selected conditions; selected data sources; or for special projects	2	11.8%	2	7.7%	1.5	---
No or rarely, only as required (e.g. community investigations)	13	76.5	24	88.9%	0.9	.40
Electronic health information transaction						
Electronic transaction method used to receive a reported birth defect case or for case identification or case finding						
Web based health information ports	5	29.4%	13	50.0%	0.6	.22
Secure File Transfer	14	82.4%	12	46.2%	1.8	**.03**
Internal health department electronic upload or transaction:	2	11.8%	6	23.1%	0.5	.45
External electronic download transaction or other type of download, e.g. encrypted e-mail, secure mail, CD	6	35.3%	10	38.5%	0.9	1.00

No. – number of programs; % - percent; ICD-9-CM - International Classification of Diseases, Ninth Revision, Clinical Modification; ICD-10 - International Classification of Diseases, Tenth Revision; CDC/BPA - Centers for Disease Control and Prevention/British Paediatric Association

Multiple responses allowed for all categories.

[1]Ratios are calculated by dividing the % of active case-finding programs by % of passive case-finding programs for the category.

[2]P values are calculated using Fisher's exact test.

Table 3: Selected Data Collection Elements by Population-based Birth Defects Surveillance Programs

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)		Ratio[1]	P value[2]
	No.	%	No.	%		
Geocoding						
Program routinely geocode or have access to geocoded data for **birth defect CASES**						
Yes	12	70.6%	16	61.5%	1.2	.76
No	4	23.5%	9	34.6%	0.7	
Unsure	1	5.9%	1	3.8%	1.5	
Program routinely geocode or have access to geocoded data for **ALL LIVE BIRTHS** (i.e. denominator data)						
Yes	7	41.2%	15	57.7%	0.7	.41
No	7	41.2%	9	34.6%	1.2	
Unsure	3	17.6%	2	7.7%	2.3	
Maternal residency						
Time period collected on maternal residency						
Maternal residency at date of delivery	17	100.0%	22	84.6%	1.2	.14
Maternal residency at date of conception	4	23.5%	1	3.8%	6.1	.07
Maternal residency collected during the pregnancy time period	3	17.6%	1	3.8%	4.6	.28
Prenatal Surveillance						
Conducts specific prenatal surveillance to identify potential cases of birth defects that are prenatally diagnosed as the pregnancy is progressing during the current time period						
Yes	3	17.6%	0	0.0%	---	.06
Yes, from selected data sources	4	23.5%	5	19.2%	1.2	1.00
Yes, for selected diagnosis	0	0.0%	2	7.7%	---	.51
No	10	58.8%	21	80.8%	0.7	.17
Family History						
Program routinely collects (and records) information on **family history, (1st degree such as bio mother, bio father, siblings or greater) of birth defects** in relation to index case						
Yes	12	70.6%	6	23.1%	3.1	**.004**
No	5	29.4%	20	76.9%	0.4	
Program able to identify **siblings** within database by tracking through the biological mother						
Yes	5	29.4%	5	19.2%	1.5	**.05**
Probably; but only for siblings that are also born in the state	11	64.7%	13	50.0%	1.3	
No	1	5.9%	8	30.8%	0.2	

Follow-up studies						
Program has the capacity to conduct follow-up studies of children with birth defects						
Yes, under 1 year of age	---	---	4	15.4%	---	**.02**
Yes, through 5 years of age	4	23.5%	6	23.1%	---	
Yes, through 18 years of age	5	29.4%	---	---	---	
Yes, over age 18 years	3	17.6%	2	7.7%	---	
No or unsure	5	29.4%	14	53.8%	---	---
Program has access or can link to cost/charge or health care service data during the FIRST YEAR of life						
No access/link	7	41.2%	13	50.0%	0.8	.16
Yes, access or link to cost/charge data	7	41.2%	8	30.8%	1.3	1.00
Yes, access or can link to healthcare resource utilization data	9	52.9%	3	11.5%	4.6	**.005**
Program has access or can link to cost/charge or health care service data BEYOND first year of life						
No access/link	8	47.1%	22	84.6%	0.6	**.02**
Yes, access or link to cost/charge data	7	41.2%	2	7.7%	5.4	**.02**
Yes, access or can link to healthcare resource utilization data	6	35.3%	0	0%	---	**.002**
Programs' utilization of cost or charges data						
Never utilized	9	52.9%	16	61.5%	0.9	.75
Economic analysis (such as cost-benefit analysis or cost-effectiveness analysis)	5	29.4%	1	3.8%	7.7	**.03**
Program planning-justification	2	11.8%	0	0.0%	---	.15
Needs Assessment	1	5.9%	2	7.7%	0.8	1.00
Legislative request	1	5.9%	2	7.7%	0.8	1.00

No. – number of programs; % - percent

[1]Ratios are calculated by dividing the % of active case-finding programs by % of passive case-finding programs for the category.

[2]P values are calculated using Fisher's exact test.

Supplemental Digital Content (SDC)

Supplemental Table 1
National Birth Defects Prevention Network (NBDPN) Survey of
Population-based Birth Defects Surveillance Programs in the United States

Program status/structure (Figure 1 and Table 1)

- What is the current status of your population-based birth defects surveillance system in your state or territory? (We mean a system that uses more than birth certificates for case identification)?
- What proportion of your state's or territories' birth population is covered by your state's birth defect surveillance system (e.g.: statewide = 100%)
- Which birth defects does your surveillance system currently identify?
- Which pregnancy outcomes does your surveillance system categorize?
- What percent do the following funding sources currently contribute to the annual costs of running your birth defects surveillance program? This is funding just for your surveillance activities and does not include any research grants, e.g. NBDPS. Check all that apply.

Coding, Quality Procedure for Case Confirmation, and Abstraction Practices (Table 2)

- What disease classification coding system does your surveillance system utilize? (Check all that apply).
- Who is responsible for assigning a disease classification code of the major and minor birth defects to a birth defects case?
- What type of background or experience have you utilized when hiring a data abstractor or other staff who will review/read medical records information for case identification or case verification?
- Does your surveillance system utilize data quality procedures to assess accuracy of the birth defects case status (a true case)?
- Does your surveillance system have the ability to classify birth defect cases into isolated, multiple, and syndromes (not using disease codes)?

Selected Data Collection Elements by Population-based Birth Defects Surveillance Programs (Table 3)

- Does your program routinely geocode, or have access to geocoded data for birth defect CASES?
- Does your program routinely geocode, or have access to geocoded data for the set of ALL LIVE BIRTHS (i.e., denominator data)?
- What time periods does the program collect maternal residency information?
- Does your program conduct specific prenatal surveillance to identify potential cases of birth defects that are prenatally diagnosed as the pregnancy is progressing during the current time period?
- Does your surveillance system routinely collect (and record) information on Family history (1st degree (e.g. bio mother, bio father, siblings) or greater) of birth defects in relation to the index case.

    - If no, please identify reasons (check all that applies).
    - Is the surveillance system able to identify siblings within your database by tracking through the biological mother?
- Does your surveillance system have the capacity to conduct follow-up studies of children with birth defects? Check all that apply.
    - If yes, can you add a module to your surveillance system's database for a follow-up study?
    - If yes, do you already have a module for a follow up study in your surveillance system's database?
- Does your surveillance program currently have access or link to cost/charge or health care service* data during the FIRST YEAR of life? (*Note: Health care service data could include comprehensive information about exams performed, well childcare visits, immunizations, physician/outpatient visits, hospital admissions, treatments and procedures, etc. These types of data do not necessarily include associated dollar amounts.)
    - If yes for access or link to cost/charge data or healthcare resource utilization data during the first year of life, which data sources?
    - If yes for access or link to cost/charge data or healthcare resource utilization data during the first year of life, which data sources?
- Does your surveillance program currently have access or link to cost/charge or health care service* data BEYOND the first year of life? (*Note: health care service data could include comprehensive information about exams performed, well childcare visits, immunizations, physician / outpatient visits, hospital admissions, treatments and procedures, etc. These types of data do not necessarily include associated dollar amounts.)
    - If yes for access or link to cost/charge data or healthcare resource utilization data BEYOND the first year of life, which data sources?
- How has your program utilized cost or charges data?

Open-ended Questions:

- What three areas or activities are of highest priority for your birth defect surveillance program/system during 2012 thru 2013?
- What are three most important challenges your birth defects surveillance program/system will face during 2012 thru 2013?

IVc. Manuscript 2: Opportunities for Advancing Public Health Surveillance of Birth Defects in the United States

Prepared for submission to Public Health Reports

Opportunities for Advancing Public Health Surveillance of Birth Defects in the United States

Author names (with academic credentials)

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The findings and conclusions in this study are those of the authors and do not necessarily represent the official position of the Centers for Disease Control and Prevention.

The material is based upon a Dissertation, submitted in partial fulfillment of the requirements for the doctoral degree at the School of Public Health of the University of Illinois at Chicago.

Target journal: Public Health Reports

ABSTRACT

Objective: To assess how United States (US) population-based birth defects surveillance programs are addressing current and emergent needs using the *CDC strategic framework* for public health surveillance (lexicon and standards, legal authority, technological advances, workforce, and analytic capacity).

Method: A mixed method approach comprising a survey and semi-structured interviews was used to examine the *CDC strategic framework* considerations. Programs' legal authorities, clinical information collected, data sources, and types of workforce were obtained from a survey completed by US birth defects programs. The interviews were conducted with nine program directors using a purposive sampling to supplement survey data; themes were identified through a grounded analytical approach.

Result: Most birth defects programs (86%) have legislative mandate to conduct surveillance whereas the remaining rely on other types of legal authority. Central to these programs is multiple data source case ascertainment that includes prenatal, postnatal, public health and pediatric data, such as cytogenetic laboratories, hospital discharge summaries, outpatient clinics, vital statistics and newborn genetic and metabolic screening. Programs are proactively addressing the changing medical information collection landscape by working with data sources to obtain remote access and improve medical record functionalities. This study indicates that in order to better address challenges these programs would do well to: devote more attention to bi-directional system communication for data transactions, enhance workforce knowledge of information technology, and strengthen analytic skills through multi-state data collaborations.

Conclusion: The *CDC strategic framework* is a useful tool to assess current and emergent surveillance-program needs, strengthen programmatic effectiveness and better guide efforts to improve population health.

INTRODUCTION

In July 2012, the Centers for Disease Control and Prevention (CDC) released its "Vision for Public Health Surveillance in the 21st Century" report, which highlighted six major areas (considerations) that should be addressed to advance public health surveillance, including: 1) lexicon and conceptual framework; 2) global health surveillance; 3) information sciences and technological advances; 4) surveillance work force; 5) access to and use of public health surveillance data; and 6) analytic challenges.[1] These considerations were identified by CDC scientists and managers as important to address to meet continuing and emerging challenges and form the basis of the *CDC strategic framework* for public health surveillance.[1] This framework can be used to determine current and future needs of population-based birth defects surveillance programs.

Population-based surveillance of birth defects has been established in most geographic areas in the United States (US) in order to understand the impact of these serious conditions on communities through on-going data collection and utilization for on-going monitoring, etiologic research, prevention strategies and evaluation, as well as health service referrals for affected individuals. The approaches for birth defects surveillance by health departments or their bona fide agents have stayed relatively constant over the past few decades, yet changes in the way that data are collected, a shifting emphasis toward developing our understand of long-term health outcomes, and advances in surveillance methods are altering the practice of surveillance programs. A systematic examination of these surveillance systems to examine current and future challenges is therefore necessary. In this paper, we use the *CDC strategic framework* to assess the extent to which surveillance developments are being incorporated into practice.

METHODS

A mixed method approach comprising a survey and semi-structured interviews was used to examine the major considerations outlined in the *CDC strategic framework*. First, questions related to lexicon and standards, legal authority, technological advances, types of workforce, and analytic capacity from 2012 National Birth Defects Prevention Network (NBDPN) Program Survey were examined. All birth defects programs in the United States and Puerto Rico completed the survey; initial data collection occurred during January-February 2012 and final

data cleaning and follow-up were completed by fall 2013. SAS 9.3 was used for cleaning and analysis (SAS Institute Inc, Cary, NC). This survey data were used for tables 1-4 in describing the operations and structure of the programs.

Second, semi-structured interviews were conducted with nine program directors using purposive sampling to further understand how programs are addressing the *CDC Strategic Framework* considerations. Figure 1 presents the stratification scheme for state selections. States with an operational birth defects surveillance program were grouped by their primary case-finding approach: active, passive with case confirmation, or passive without case confirmation (three groups).[2] A second level further stratified the programs by whether they were established before or after 1997 (six groups); the year was chosen to distinguish newer programs from their more established counterparts given enhanced birth defects surveillance activities starting around this time period. These six groups were again stratified so that each primary case-finding approach was correlated to two established and one newer program (a total of nine programs); the criteria used to ensure diversity in programs in this final selection included population size, engagement in multi-state data collaborations, case inclusion beyond structural and genetic conditions, and funding base.

The selected program directors were provided a link to the *CDC Strategic Framework* report prior to the interviews, which were conducted in November/December 2013. Program directors were asked to provide qualitative comments on whether and how well birth defects programs were addressing the considerations. Written notes were taken using AudioNotes© during the semi-structured interviews, which lasted between 90-120 minutes. A codebook was developed from the transcription data, which guided the final coding of the interviews in Atlas.ti™ using a grounded analytical approach. The major themes from the interviews were examined and used to guide the study findings.

The study protocol was reviewed and approved by the University of Illinois at Chicago institutional review board (IRB protocol # 2013-0179) and by CDC National Center on Birth Defects and Developmental Disabilities human subject protection office.

RESULTS

Lexicon and Conceptual Framework

The first major consideration in the *CDC strategic framework* concerns lexicon, standards and conceptual framework. Several themes emerged during the semi-structured interviews. First, birth defects surveillance programs fit within the larger public health surveillance practice, which often uses jargon that can easily be misunderstood by stakeholders. The terms "surveillance" and "monitoring" often elicit connotations associated not necessarily with public health surveillance, but with Orwellian "big government." Given concerns over the role of government in data collection and monitoring, public health programs must assert programmatic use of the collected data and surveillance as necessary to ensure population health and safety in order to ameliorate public concern. For example, directors should focus on how the data are used, e.g. understanding disproportionately affected individuals or risk factors, without lingering on the terms (labels).

Second, certain birth defects surveillance terms can be confusing. Since birth defects can encompass a range of conditions, from structural malformations to other adverse conditions and outcomes at birth, programs need to be clear about their case definitions. Table 1 presents the reported findings from the survey data of whether the programs utilize clinical case definitions and instructions for each data field collected. Most programs with active case-finding (94.1%) indicated the use of specific instructions for each data field completion (94%) and all utilize clinical case definitions; for passive case-finding programs, a little more than half of the programs (53.8%) had data field completion instructions and 61.5% reported that they utilize clinical case definitions. Guidelines and standards that are being developed and fine-tuned through NBDPN were mentioned often during the interviews as necessary to ensure shared understanding of the terms used. The majority of the programs reported using a national guideline (NBDPN or National Birth Defects Prevention Study) for their clinical case definition (Table 1).

Legal authority

Thirty-seven (86%) of the operational birth defects surveillance programs obtain their data collection authority from statute, and 16 of those also have specific authority outlined in rules and regulations (Table 1). The remaining 6 operational programs rely on a rule or regulation for their data access. The majority of the programs did not report any negative impact, either directly or indirectly, that affects the reporting or identification of cases (33 out of

43 programs) or case record abstraction (36 out of 43 programs). Sources of restrictions included a lack of access to all pregnancy outcomes, lack of legislative enforcement, hindered access to selected datasets, and restrictive "opt in/opt out" rules.

A topic raised during the interviews was the idea that surveillance programs need to be politically acceptable. In reference to the *CDC strategic framework* considerations of data access and use, an interviewee said, "The one thing missing from confidentiality is that we are almost all governmental entities. The issue of politically palatability is important and if the public turns against us, then we are not going to be able to do what we do."

Another interview theme was the expansion of data access and linkage with data sources. For programs that expand surveillance past infancy to examine long-term outcomes of children born with birth defects, clarification is needed regarding program authority to access and link to new databases to determine health, education progress, and cost utilization. In addition, several interviewees discussed the development of and access to data warehouses that are used to link data from multiple state programs to understand the outcomes of affected individuals. These data warehouses offer a data source that might help researchers to understand the health outcomes of children with birth defects. The program directors cited difficulties linking or accessing data to non-health data sources, such as education and criminal justice databases. Without explicit authority or a community of practice for doing this, the challenge then lies in the ability of the program managers to articulate a benefit for the shared populations that these programs serve while addressing any privacy concerns.

Information Sciences and Technological Advances

Three primary themes regarding technological advances emerged during the interviews. First, programs are experiencing data flux as medical information transitions from paper to electronic formats. Table 2 shows programs reporting changes to the level of detail of clinical information that result from the transition from historical hard copy to electronic medical records, with active case-finding systems reporting more fluctuations. Negative effects of electronic records include: an increase in condensed information with lack of primary documentation of sources, poorly organized files making it difficult for data abstractors to cull through the information, and repetitive information without improvements in detail. One benefit

noted is the feasibility of multiple record uploads, which helps institutionalize the reporting process so that there is less dependency on reporting source staff.

Second, as medical information becomes electronic, remote access to the data systems containing the medical information has greatly increased the efficiency of record review and verification by allowing for: reduced travel time for data abstractors who formerly had to visit each birthing facility; increased time to review the medical information (given there is no longer a dependence on medical personnel time to provide program staff access to the medical information); and quicker medical record retrieval. The drawback is information access restrictions. For instance, researcher access can be restricted to only certain parts of medical charts or to particular program staff. Again data systems' designs might block certain search and/or copying features. Constant quality control needs to be performed to ensure that the types of information collected by programs are accessible. Third, health information exchanges are becoming a reality in certain regions of the country, and although most birth defects surveillance programs are not participating in these exchanges yet, preparation must occur now. Adopting industry standards, such as HL7, and leveraging federal funding sources are important toward preparing birth defects surveillance programs to bi-directionally communicate and share medical information. This exchange will eventually shift the emphasis of surveillance programs from how the medical information is obtained to what types of data are collected. One interviewee said, "One of these days, it's going to be harder to tell passive/active systems apart. If you can look at [medical information from] your desk, it is going to be easier for passive systems to get decent data."

Workforce

Table 3 presents the number of staff by program functions for birth defects surveillance programs. Active case-finding programs have dedicated staff (1 or more FTE positions) for epidemiologist/statistician, data / information technology / web support, and data abstractors while maintaining other staff (less than 1 FTE position) for director / program manager and clinical reviewers. Passive case-finding programs focus staff for director / program manager with 18 programs stating that they have dedicated managers. They also employ other staff (less than 1 FTE position) for epidemiologist/statistician and data/information technology web support while many of these programs have no clinical reviewers (69%) or data abstractors (50%).

Several workforce themes emerged during the interviews, including the need for more specialized information technology (IT) skills, cross-training, and strategic thinking. As programs become more dependent on IT systems and processes, they often struggle to train staff to understand IT functions or hire personnel with IT backgrounds. One interviewee noted, "You have more chance to teach public health people IT concepts than the other way around. It's from the people I see through the years… You need to find more people like me, a bimorph [who can converse in both worlds]. "

Cross-training and active participation beyond job duties for program staff are important. Suggestions from the interviews include: 1) engage staff in different aspects of managing the surveillance system, 2) allow staff input in decision-making, and 3) create a participatory environment to keep staff engaged and invested. Programs managers should have a clear vision for the program and strategically prioritize the workload to generate outcomes. Also, tapping into expertise from academic and national resources such as CDC and NBDPN can enhance state surveillance program capacity to maximize data utilization.

Analytic Capacity and Challenges

The final consideration examined is analytic capacity, including database management, which is important to ensure effective utilization and dissemination of the data. The top databases used by the 43 operational birth defects programs are Microsoft® Access (53%) and Sequel server/Oracle (51%). The primary analytic software include SAS® (70%s), GIS software (40%), and Microsoft® Excel (37%).

A primary theme for enhancing analytic capacity emerged as interviewees discussed a "community of learning," which would periodically convene analytic staff to discuss common analytic issues via webinars or conference calls. State program analytic staff often lacks opportunities to interact with other analytic staff to learn different skills, such as linkage with non-health data sources. Also, gathering ideas for data interpretation and presentation to disseminate to various stakeholders through information sharing and technical demonstrations can enhance the analytic capacity within and among programs. These interactions should include practical data application.

DISCUSSION

The *CDC Strategic Framework* for public health surveillance provides a structure for programs to examine areas of strengths and weaknesses in their surveillance practice given the changing health data collection environment, which requires accounting for diminishing resources, increasing attention to government data collection systems and privacy protection as well as increased in health data exchanges. This information gathered can be used to focus efforts to improve how surveillance programs address current and emerging needs.

A shared understanding of lexicon and concepts can facilitate communication about the intent and utility of surveillance programs. Hall et al. note, "Public health surveillance is not defined by the system used to collect data but by the purpose of the data collection — the specific public health question that the data will be used to answer and the link to disease prevention and control."[3] Describing concretely why population data are collected and how they will be used will help improve stakeholder support for the program.

Balancing between the need for a public health program to access data and the public's concerns over privacy is an on-going issue for birth defects surveillance systems, as more information shifts to electronic formats. Boundaries change as data become linked and integrated. A salient issue raised during the interviews was that the activities of public health programs need to be politically acceptable. It is not enough that public health professionals create results that are valued, but the benefit to society must outweigh the consequences."[4]

The criteria that determine what medical information can be accessed by surveillance programs are in flux as health care providers transition their data systems, and program staff are learning to understand the new format of the medical information. Hsiao et al. reported that as of 2012, 72% of physicians had adopted some type of electronic system and that 40% had adopted capabilities needed for a basic electronic health record system.[5] This changing data landscape offers opportunities and challenges. Data collectors must learn to decipher medical records that sometimes suffer from imprecise or verbose data entry; however, the electronic medical records offer an opportunity for remote access to data sources. Programs must also prepare practices to receive data from health information exchanges and ensure compatible functionality between systems. On-going data quality vigilance will help programs as new data from various sources are used for case finding and verification.

A feature of birth defects surveillance programs is their multiple data source ascertainment methodologies. Ascertaining cases from multiple data sources increases case identification that might be missed by a single source, especially for programs that ascertain cases from all pregnancy outcomes or rely on provider reporting and need to perform case verification.[6,7] Additionally, birth defects surveillance programs have generally focused on ascertaining cases at birth, but the need to monitor health outcomes of these children as they age could shift the activities performed by surveillance programs. This again will require linking to new data sources or using information collected from existing data sources.

Having a skilled and dedicated workforce is essential to ensuring an effective surveillance program. Drehobl et al. stress the importance of a workforce analysis—including reviewing workforce availability and identifying existing gaps and future needs.[8] The staffing pattern for birth defects surveillance programs reflects programmatic emphasis, whether on reporting systems or clinical accuracy of information gathered; however, a prominent need was the ability of public health staff to better understand IT language and systems. Staff that normally do not interface with IT systems are doing so more often and are learning how these systems facilitate efficient data collection and analysis. Also, enhancing the analytic knowledge base of the workforce through a community of peer collaborative learning is important. Beyond on-going training, a national peer collaborative learning mechanism might improve analytic skills through hands-on learning. The NBDPN created a mechanism whereby multiple states collaborate on analytic projects and staff participate either as lead investigators or members of a research group and are able to participate in the development of a project from inception to publication to information dissemination. Through collaborations, expertise from various members can be tapped.

Strengths and Limitations

Survey responses were obtained for 100% of population-based birth defects programs in the U.S. A mixed method approach was used to further understand how birth defects programs were addressing major considerations affecting surveillance practice. However, reporting bias was a potential threat to external validity. The use of purposive sampling was used to obtain a diverse perspective to minimize reporting bias.

CONCLUSION

A changing surveillance environment and diminished resources are altering the practice of population-based data collection of health outcomes such as birth defects. Birth defects surveillance programs have been operational across the U.S. for several decades and offer a rich source of data that can be used to address community concerns and to help affected children and their families but need to continuously focus on data improvement and utilization to ensure relevancy within a dynamic environment. The *CDC Strategic Framework* for public health surveillance serves as a useful tool to assess current and emerging challenges, which, in turn, can help program leadership to manage resources to improve program effectiveness, especially as regards the translation of data into improved public health outcomes.

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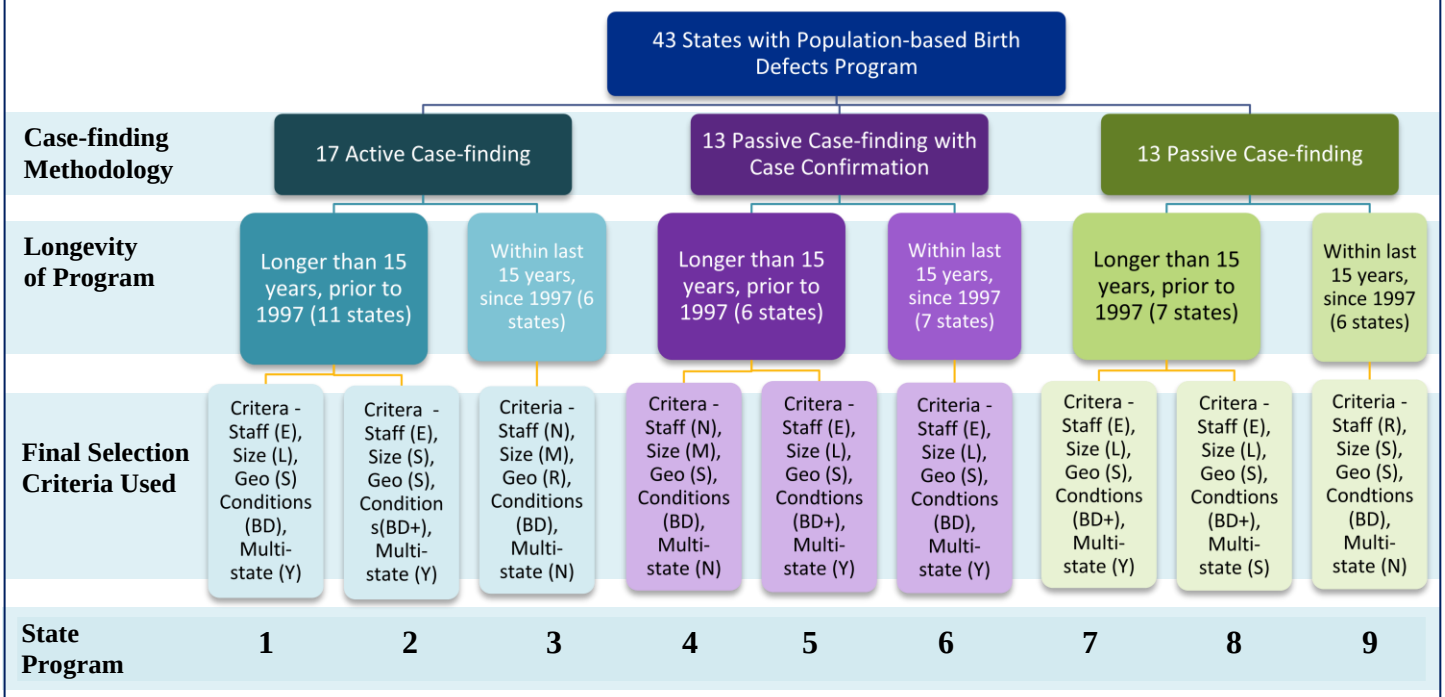
316 **Tables and figures:**

Figure 1: Selection Criteria for Nine Semi-structured Interviews



Secondary criteria used for final state selection:

Staff - core staff experience in birth defects surveillance:
N – new; R- recent; E - experienced

Size – size of live birth (LB) population for the catchment area:
S - less than 50,000 LB; M - 50,000 to less than 100,000 LB; L – greater than 100,000 LB

Geo - geographic coverage:
S -State-wide; R - regions (selected counties)

Conditions – conditions ascertained:
BD - major birth defects; BD+ - major birth defects and other conditions

Multi-state - engagement in multi-state data collaborations:
Y - Yes; N - No; S – Somewhat

Table 1. Case Definition and Legal Authority of Population-based Birth Defects Surveillance Programs

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)		Ratio[1]	P value[2]
Case Definition	No.	%	No.	%		
Utilize a clinical case definition, such as inclusion and exclusion criteria, for birth defects included in the surveillance system						
No	0	0.0%	10	38.5%	--	**.003**
Yes	17	100%	16	61.5%	1.6	
If yes, what does your surveillance system use? (multiple choices allowed)						
Developed our own system	9	52.9%	7	43.8%	1.2	.73
NBDPN Abstractor guidelines	8	47.1%	10	62.5%	0.8	.50
Data dictionary for each data element collected or ascertained	4	23.5%	2	12.5%	1.9	.66
NBDPS	4	23.5%	0	0.0%		.10
NBDPS modified	2	11.8%	1	6.3%	1.9	1.00
Specific instructions in place to complete each data field collected for each birth defect case						
No	1	5.9%	8	30.8%	0.2	**.01**
Yes	16	94.1%	14	53.8%	1.8	
Unsure	0	0.0%	4	15.4%	--	
Legal Authority						
Legislation	9	52.9%	11	42.3%	1.3	.87
Rule or regulation	1	5.9%	3	11.5%	0.5	
Both	6	35.3%	11	42.3%	0.8	
Other, e.g. public health authority	1	5.9%	1	3.8%	1.5	
Impact of Authority						
Any laws or regulations that negatively impact (directly or indirectly) surveillance function of the **reporting of cases or the identification of cases to the program**						
No	14	82.4%	19	73.1%	1.1	.73
Yes	2	11.8%	6	23.1%	0.5	
Unsure	1	5.9%	1	3.8%	1.5	
Any laws, regulations, or authority that negatively impact (directly or indirectly) the surveillance function of **case record abstraction (i.e., the ability to access medical records and other information?)**						
No	16	94.1%	20	76.9%	1.2	.38
Yes	1	5.9%	5	19.2%	0.3	
Unsure	0	0.0%	1	3.8%	--	

No. – number of programs; % - percent

[1]Ratios are calculated by dividing the % of active case-finding programs by % of passive case-finding programs for the category.

[2]P values are calculated using Fisher's exact test.

Table 2. Level of Information Observed in Electronic Health Records

Electronic health information	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)		Ratio[1]	P value[2]
Any change in the **level of detail of clinical information** compared to historical hard copy medical record as a result of electronic medical record (EMR):	No.	%	No.	%		
Stayed the same	4	23.5%	8	32.0%	0.7	.25
Increased	2	11.8%	1	4.0%	2.9	
Varied	6	35.3%	4	16.0%	2.2	
Decreased	3	17.6%	1	4.0%	4.4	
Unsure	2	11.8%	7	28.0%	0.4	
Any change in the **level of completeness in the content** compared to historical hard copy medical record as a result of electronic medical record (EMR):						
Stayed the same	0	0.0%	5	20.0%	--	**.05**
Increased	4	23.5%	3	12.0%	2.0	
Varied	5	29.4%	4	16.0%	1.8	
Decreased	5	29.4%	1	4.0%	7.4	
Unsure	3	17.6%	8	32.0%	0.5	

Five passive case-finding programs had missing responses.
No. – number of programs; % - percent
[1]Ratios are calculated by dividing the % of active case-finding programs by % of passive case-finding programs for the category.
[2]P values are calculated using Fisher's exact test.

Table 3. Data Sources Accessed by Population-based Birth Defects Surveillance Programs

Data Sources	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)		Ratio[1]	P value[2]
Prenatal Data Sources	No.	%	No.	%		
Prenatal Pathology Reports	8	47.1%	2	7.7%	6.1	**.007**
Prenatal Diagnostic Centers (MFM)	9	52.9%	3	11.5%	4.6	**.005**
Prenatal Ultrasound database	7	41.2%	1	3.8%	10.7	**.004**
Prenatal Genetic counselors	5	29.4%	2	7.7%	3.8	.09
Prenatal Obstetricians	5	29.4%	2	7.7%	3.8	.09
Prenatal sites Logbooks	6	35.3%	0	0.0%		**.002**
Prenatal Cytogenetic Laboratories	8	47.1%	5	19.2%	2.4	.09
Prenatal Hospital-based Cytogenetic Laboratories	5	29.4%	3	11.5%	2.5	.22
Postnatal Sources						
Postnatal Delivery hospital discharge (in-patient)	17	100.0%	18	69.2%	1.4	**.01**
Postnatal Delivery hospital discharge (out-patient)	14	82.4%	12	46.2%	1.8	**.03**
Postnatal Labor and Delivery Logbooks	9	52.9%	2	7.7%	6.9	**.003**
Postnatal Emergency rooms	8	47.1%	2	7.7%	6.1	**.007**
Postnatal Newborn Nursery Logbooks	10	58.8%	2	7.7%	7.6	**<.001**
Postnatal NICU Logbooks	9	52.9%	1	3.8%	13.8	**<.001**
Postnatal NICU Reports	9	52.9%	5	19.2%	2.8	**.04**
Pediatric (tertiary) hospital discharge (in-patient)	15	88.2%	12	46.2%	1.9	**.009**
Pediatric (tertiary) hospital discharge (out-patient)	13	76.5%	8	30.8%	2.5	**.005**
Postnatal Pathology Reports	10	58.8%	3	11.5%	5.1	**.002**
Postnatal Cytogenetic laboratories	7	41.2%	7	26.9%	1.5	.51
Postnatal Hospital-based cytogenetic laboratories	5	29.4%	7	26.9%	1.1	1.00
Postnatal Newborn Hearing Screening	5	29.4%	10	38.5%	0.8	.74
Postnatal Newborn Genetic and Metabolic Screening	6	35.3%	11	42.3%	0.8	.75
Specialty and Pediatric Data Sources						
Specialty clinics: hospital-based/other outpatient clinics	12	70.6%	9	34.6%	2.0	**.03**
Pediatric cardiology	8	47.1%	7	26.9%	1.7	.21
Pediatric genetics	10	58.8%	10	38.5%	1.5	.23
Pediatric orthopedics	9	52.9%	5	19.2%	2.8	**.04**
Pediatric urology	10	58.8%	5	19.2%	3.1	**.01**
Pediatric Developmental	4	23.5%	3	11.5%	2.0	.41
Pediatric surgery	10	58.8%	5	19.2%	3.1	**.01**
Pediatric neurology (e.g. NTDs)	8	47.1%	7	26.9%	1.7	.21
Pediatric Orofacial	9	52.9%	7	26.9%	2.0	.11
Other specialty clinic	4	23.5%	4	15.4%	1.5	.69
State and other data sources						
Vital Records	11	64.7%	16	61.5%	1.1	1.00
Physician reports	4	23.5%	7	26.9%	0.9	1.00
Hospital discharge data set	9	52.9%	12	46.2%	1.1	.76
Medicaid data set	3	17.6%	5	19.2%	0.9	1.00
Children with Special Health Care Needs data set	1	5.9%	7	26.9%	0.2	.12
EHDI Registry	2	11.8%	9	34.6%	0.3	.15
Other data sets-registries	1	5.9%	3	11.5%	0.5	1.00

No. – number of programs; % - percent;

[1]Ratios are calculated by dividing the % of active case-finding programs by % of passive case-finding programs for the category.

[2]P values are calculated using Fisher's exact test.

Table 4. Number of Personnel by Case-finding Types for Population-based Birth Defects Surveillance Programs

Personnel Type	Number of FTEs within a program	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
		No.	%	No.	%
Director / Program Manager	No FTEs	0	0%	8	31%
	<1 FTE	10	59%	9	35%
	1 or more FTE	7	41%	9	35%
Epidemiologist / Statistician	No FTEs	4	24%	5	19%
	<1 FTE	5	29%	13	50%
	1 or more FTE	8	47%	8	31%
Clinical Reviewer	No FTEs	1	6%	18	69%
	<1 FTE	12	71%	6	23%
	1 or more FTE	4	24%	2	8%
Data Manager/ IT / Web Support	No FTEs	2	12%	6	23%
	<1 FTE	7	41%	6	23%
	1 or more FTE	8	47%	5	19%
Data Abstractors	No FTEs	0	0%	13	50%
	<1 FTE	1	6%	6	23%
	1 or more FTE	16	94%	5	19%

FTE – full-time equivalent personnel

V. Discussion

This study focuses on three areas: 1) examining the characteristics, e.g., methods and infrastructure, of the population-based birth defects surveillance systems in the U.S.; 2) understanding the intended purposes of the population-based birth defects surveillance programs; and 3) applying the CDC *Strategic Framework* for Public Health Surveillance in the 21st Century to assess current and future needs of birth defects surveillance programs. Most states have a population-based birth defects surveillance program, and the utility of these programs depends in part on their application of their data to understand the impact of birth defects on the affected families and communities.

Va. Recommendations

Recommendations to improve the practice of birth defects surveillance in the United States at the state and national levels address:

1) Vision and strategic direction

Given resource constraints and increased pressure to demonstrate impact of programmatic activities, program directors and others in leadership positions need to both articulate a clear vision of the direction of the program and prepare to capitalize on opportunities for success. This includes the ability to capitalize on emergent medical and health data collection technologies to improve efficiency in both data collection for birth defects programs and data linkage to databases on health and other outcomes of individuals with birth defects.

2) National standards for conducting birth defects surveillance

National standards for birth defects surveillance will facilitate collaborative efforts and help guide state programs toward achieving national performance measures. The work begun

by the NBDPN to develop national standards for birth defects focusing on data quality and utility is important given the range of ascertainment methodologies for birth defects across the U.S. However, there is an emergent need for birth defects surveillance programs to prepare for health information exchange by adapting existing standards for exchange and communication, such as mapping clinical coding systems like Systematized Nomenclature of Medicine – Clinical Term (SNOWMED CT) with the International Classification of Diseases, Version 10 - Clinical Modification (ICD-10-CM) for birth defects. The birth defects surveillance community needs to ensure that programs are prepared for changing health information transmission.

3) Stakeholder engagement

Continued engagement of partners is important for the success of ongoing and emergent efforts of birth defects surveillance programs. Partners played a key role in shaping the development of the systems, and their continued engagement can help shape a program's direction, not only by enhancing program utility but also by providing an important voice for policy makers and the local community.

Being aware of the wider community and their perception of public health and surveillance activities can help birth defects program staff better frame their work. Surveillance is an important tool to advance public health work, but it must be done within the context of stakeholders and community buy-in.

4) Enhancement of "Community of Learning and Collaborative Projects"

National efforts to collaborate on multi-state pooled data projects, such as developing national prevalence estimates for specific birth defects, have been successful in generating data that are widely used and in increasing understanding of the epidemiology of some rare birth defects. Given limited resources, continued support for these collaborative projects will be

important as we move forward. Another benefit of the multi-state collaborative projects is that they hone the analytic and scientific writing skills of birth defects program staff. These projects allow program staff to learn while doing since the work is spread across programs. This is an important opportunity since some of the staff are often in a program where they are the only analytic staff and in a community of learning, staff benefit from exchanges, decision making process, and following through to completion on tasks.

Periodic webinars and training opportunities to allow program staff to develop and sharpen analytic skills are needed. Many programs lack on-going training for staff, who could benefit from acquaintance with scholarship on topics such as analyzing health outcome datasets, linking birth defects data with other databases, and data quality techniques. Also, development of analytic and other tools for data process and linkage can enhance program efficiency.

5) Enhancement of Data Dissemination and Communication Channel

Programs need to develop better data dissemination and communication plans to ensure the data are interpreted and understood by their intended audiences. It is not enough just to publish data reports, tables or peer-reviewed papers; programs need to provide interpretation of the data and present them with the respective audiences in mind. Program should consider using various data dissemination channels, such as the publication of scientific articles along with synopses of their key findings for the broader audiences, creation of queryable public datasets, and framed social messaging.

Vb. Conclusion and Leadership Implications

Birth defects surveillance has been conducted in the United States for several decades and can be used for epidemiologic, research, prevention, education, and health service planning to help communities understand the impact of these conditions and to assist families with

affected children. Given a lack of a national system for population-based surveillance, continued coordination and support of state birth defects surveillance programs are needed. Advancing the field will require concerted efforts at both state and national levels given the impact of diminished resources and the changing field of health information data.

This study not only presented the structure and operations of the population-based birth defects surveillance programs in the U.S. but also applied the CDC *Strategic Framework* for public health surveillance to identify areas to enhance programmatic effectiveness. Several recommendations with leadership implications for advancing the work of population-based birth defects surveillance in the U.S. were presented. First, program directors should be able to articulate clearly their vision for the program and to show how their data are impacting the health of their communities and affected families. Second, leadership must be provided to guide the efforts for developing and adapting national standards specific for birth defects surveillance. This includes standards to improve data quality and utility across programs in the U.S. so as to enhance multi-state collaborations as well as standards that are currently used by public health programs, such as IT messaging standards like SNOWMED, that require adaptation for birth defects specific purposes. Third, leaders should actively engage in collaborations to develop and strengthen data linkages to databases on health and other outcomes of individuals with birth defects. As individuals with birth defects live longer, a better understanding of the survival patterns, health disparities, and health outcomes is needed. Birth defects programs are in a position to be able to address these important issues through data linkage with other health outcome databases. Fourth, stakeholder engagement played a key role in establishing and shaping the direction of many programs and continued development of the partnerships is key to ensuring that the programs stay relevant and meet the needs of the community. Finally, leaders

should ensure not only the translation and dissemination of the collected data, but the evaluation of the programmatic products in meeting the needs of the intended audiences.

Surveillance is a core public health activity, and programs developed to monitor health conditions need to ensure that they are responding effectively and efficiently to public health problems. Birth defects surveillance programs offer a rich source of data that can be used to address community concerns and to help affected children and their families but need to continuously focus on data improvement and utilization to ensure relevancy within a dynamic environment.

Appendices

Appendix A: Selected Characteristics of Birth Defects Surveillance Program

State	Case Finding[1]	Longevity of System[2]	Core Staff Experience in BD Surveillance[3]	Size of Live Birth Population[4]	Geographic Coverage[5]	Outcomes Covered[6]	Multi-state Data Collaborations[7]
LA	A	N	N	M	S	BD	N
MN	A	N	N	M	R	BD	N
NH	A	N	E	S	S	BD	N
SC	A	N	R	M	S	BD	N
DE	A	N	R	S	S	BD+	N
MA	A	N	E	M	S	BD+	Y
NC	A	E	E	L	S	BD	Y
OK	A	E	R	M	S	BD	N
PR	A	E	N	S	S	BD	S
UT	A	E	E	M	S	BD	Y
AR	A	E	E	S	S	BD	S
AZ	A	E	D	M	S	BD	Y
CA	A	E	D	M	R	BD	S
GA	A	E	D	M	R	BD	Y
HI	A	E	N	S	S	BD	N
TX	A	E	E	L	S	BD	Y
IA	A	E	E	S	S	BD+	Y
FL	P	N	E	L	S	BD	Y
MS	P	N	R	S	S	BD	N
ND	P	N	R	S	S	BD	N
TN	P	N	N	M	S	BD	N
WI	P	N	E	M	S	BD	N
CT	P	N	R	S	S	BD+	N
MD	P	E	N	M	S	BD	N
MO	P	E	N	M	S	BD	N
NE	P	E	N	S	S	BD	S
VA	P	E	N	L	S	BD	N
WA	P	E	N	M	S	BD	N
WV	P	E	E	S	S	BD	N
MI	P	E	E	L	S	BD+	Y
KY	P+	N	N	M	S	BD	N
ME	P+	N	R	S	S	BD	N
NV	P+	N	R	S	S	BD	N
OH	P+	N	R	L	S	BD	N

RI	P+	N	E	S	S	BD	N
VT	P+	N	R	S	S	BD	N
IN	P+	N	R	M	S	BD+	N
AK	P+	E	E	S	S	BD	N
NM	P+	E	N	S	S	BD	N
CO	P+	E	E	M	S	BD+	Y
IL	P+	E	E	L	S	BD+	S
NJ	P+	E	E	L	S	BD+	S
NY	P+	E	E	L	S	BD+	Y

[1]Case-finding methodology: A - Active; P+ - Passive + case confirmation; P- Passive
[2]Longevity of program: E- Longer than 15 years (prior to 1997); N - Within last 15 years (since 1997)
[3]Core Staff Experience in Birth Defects Surveillance: N – new; R- recent; E - experienced
[4]Size of Live Birth Population: S - less than 50,000 LB; M - 50,000-less than 100,000 LB; L - 100,000 + [4]Live birth (LB)
[5]Geographic Coverage: S -State-wide; R - regions (selected counties)
[6]Outcomes Covered: BD - major birth defects; BD+ - major birth defects + other conditions
[7]Engagement in Multi-state Data Collaborations: Y - Yes; N - No; S – Somewhat

Note: These states either do not have a surveillance system or have suspended activities in 2012: AL, DC, ID, MT, OR, PA, SD, WY.
Source: 2012 NBDPN Annual Report Program Directory, BDRA with updates by C. Mai

Appendix B: Birth Defects Surveillance Program Authority & Reportable Outcomes

Source: 2012 National Birth Defects Prevention Network (NBDPN) Annual Report

State	Annual Births	Population Coverage	Legislation/Rule	Year Enacted	Outcomes Covered
AK	11,000	State	7 AAC 27.012	1996	Major birth defects
AR	41,000	State	Senate Bill Act 214	1985	Major structural birth defects
AZ	87,053	State	Legislation and rule	1988	Major birth defects & genetic diseases
CA	70,000	Selected counties	Legislation and rule (1982), recodified (1996)	1982	Serious structural birth defects
CO	66,346	State	Colorado Revised Statutes (CRS) 25-1.5-101 - 25-1.5-105	1985	Structural birth defects, fetal alcohol syndrome, selected genetic and metabolic disorders; muscular dystrophy; selected developmental disabilities; very low birth weight; others with medical risk factors for developmental delay
CT	43,000	State	Legislation Sec. 19a	1991	All major structural birth defects; biochemical, genetic and hearing impairment; any condition which places a child at risk for needing specialized medical care ICD-9 codes 740 - 759.9 and 760.71
DE	12,000	State	House Bill No. 197	1997	Selected birth defects, developmental disabilities if due to a birth defect, selected metabolic defects, genetic diseases, infant mortality, congenital infections, autism
FL	213,234 in 2011	State	Section 381.0031(1,2) F.S., allows for development of a list of reportable conditions	1999	major structural malformations and selected genetic disorders

State	Annual Births	Population Coverage	Legislation/Rule	Year Enacted	Outcomes Covered
GA	138,000	State	Reportable under State Laws Official Code of GA Annotated	updated in 2003	major birth defects, genetic diseases, FAS and CP
GA (CDC)	50000	Metropolitan Atlanta	State Laws Official Georgia Code Annotated (OCGA)		Major structural or genetic birth defects
HI	18,913	Island-wide	HRS §321.421 to 426; HRS §321.41 to 44	2002	All outcomes identified on the ICD-9 and CDC/BPA codes
IA	37,831	State	Iowa Code 136A, Iowa Administrative Code 641-4.7	1986; Last revised 2009	major birth defects, Duchene/Becker muscular dystrophy, fetal deaths w/ and w/o birth defects, newborn screening disorders
IL	170,000	State	Illinois Health and Hazardous Substances Registry Act (410 ILCS 525)	1985	ICD-9-CM Codes 740.0 through 759.9; infants positive for controlled substances; very low birth weight (< 1500g); fetal death; death during the newborn hospital stay; serious congenital infections; congenital endocrine, metabolic or immune disorders; congenital blood disorders; other conditions such as retinopathy of prematurity, intrauterine growth retardation, FAS
IN	89,000	State	IC 16-38-4-7 Rule 410 IAC 21-3	2001	ICD-9-CM Codes 740-759.9, Fetal Alcohol Spectrum Disorder, Pervasive Developmental Disorder, fetal deaths, metabolic disorders & hearing loss, selected neoplasm, congenital blood disorders, and certain eye disorders
KS	41,338	State	K.S.A. 65-1,241 through 65-1,246	2004	Thirteen anomalies (and "other" congenital

State	Annual Births	Population Coverage	Legislation/Rule	Year Enacted	Outcomes Covered
					anomalies) are listed on the birth certificate and are reported
KY	56,000	State	KRS 211.651-211.670	1992	major birth defects, genetic diseases, fetal mortality
LA	~61,000	State	Law: LA R.S. 40:31.41 - 40:31.48, 2001. DHH Rule: LAC 48	2001	major structural birth defects and selected genetic diseases
MA	75,000	State	MA General Laws Statue amended in 2002 and expanded birth defects monitoring program. Regulations in 2009.	1963	major structural birth defects and chromosomal anomalies of medical, surgical or cosmetic significance
MD	75,000	State	Health-General Article, Section 18-206; Annotated Code of MD	1982	Selected birth defects until 2009, and now all significant birth defects
ME	12,814	State	22 MRSA c. 1687	1999	Selected major birth defects: NTD, clefts, gastroschisis, omphalocele, trisomy 21, reduction deformities of upper and lower limb, hypospadias and major heart defects
MI	112,000	State	Public Act 236 of 1988	1988	Congenital anomalies, certain infectious diseases, conditions caused by maternal exposures and other diseases of major organ systems
MN	73,000	2 counties (~50% of births); statewide soon	MS 144.2215-2219	2004	major "reported birth defects" as defined by CDC and ICD-9 codes up to 1 year of age
MO	79,000	State			ICD9 codes 740-759, plus genetic, metabolic, and other disorders
MS	42,000	State	Section 41-21-205 of the Mississippi	1997	Live births and reportable fetal deaths with birth

State	Annual Births	Population Coverage	Legislation/Rule	Year Enacted	Outcomes Covered
			Code of 1972		defects (fetal death of 20 completed weeks of gestation or more, or a weight of 350 grams or more) shall be reported.
NC	122,000	State	NCGS 130A-131	1995	major birth defects
ND	9234	State	North Dakota Century code 23-41	1941	selected birth defects (NTDs, congenital heart defects, cleft lip and palate, chromosomal anomalies) and other risk factors that may lead to health and developmental problems
NE	27,000	State	Laws 1972	1972	All birth defects using CDC list
NH	12,500	State	RSA 141:J, NH Administrative Rules	2008	all major birth defects and genetic diseases recommended by the CDC/NBDPN
NJ	110,000	State	NJSA 26:8-40.20 et seq., NJAC 8:20 - Amended: 1990, 1991, 1992, 2005, Readopted: 2010, Rule Amendments Adopted: 2009; Re-adopted 2010	1983	All birth defects (structural, genetic, and biochemical), all Autism Spectrum Disorders, and severe hyperbilirubinemia, are required to be reported; all special needs and any condition which places a child at risk (prematurity, asthma, cancer, developmental delay) are also reported but not required.
NM	30,000	State	In January 2000, birth defects became a reportable condition. (change in regulations)	2000	740-760.71, Currently focused on major birth defects of interest to Environmental Public Health Tracking.
NV	Nearly 40,000	State	NRS 442.300 - 442.330 - Regulation = NAC 442	1999	Major birth defects and genetic diseases
NY	250,000 - 300,000	State	PH Law Art. 2, Rules & Regulations, State	1982	Major malformations

State	Annual Births	Population Coverage	Legislation/Rule	Year Enacted	Outcomes Covered
			Sanitary Code		
OH	145,000	State	Ohio Revised Code (ORC) 3705.30 - 3705.36, signed into law in July, 2000. Ohio Administrative Code OAC)	2000	Major birth defects recommended by NBDPN, disorders on state newborn bloodspot panel, disorders related to infant hearing loss
OK	55,000	State	63 O.S. Section 1-550.2	1992	modified 6-digit ICD-9-CM codes for birth defects and genetic diseases (CDC/BPA)
PR	45,000	Island-wide	Yes, Law 351	2004	Selected major birth defects
RI	11,000	State	Title 23, Chapter 13.3 of RI General Laws	2003	major birth defects and genetic diseases
SC	60,682	State	A281,R308,H4115	2004	Neural tube defects, cardiovascular defects, musculoskeletal defects, orofacial clefts
TN	85,000	State	TCA 68-5-506	2000	45 major structural birth defects
TX	401,599	State	Health and Safety Code	1993	all major structural birth defects and fetal alcohol syndrome
UT	55,000	State	Birth Defect Rule (R398-5)	1999	major structural malformations; newborn metabolic conditions; stillbirths
VA	101,202	State	Health Law	1986; 2006	Major birth defects and genetic diseases
VT	6500	State	Act 32 (TITLE 18 VSA §5087)	2003	Major birth defects and genetic diseases, very low birth weight (less than 1500 grams)
WA	90,000	State	Notifiable Conditions: WAC 246-101	2000	From 1987 to 1991 (active surveillance), and from 1991 to the 2000 (passive surveillance), the cases reportable to the Birth Defects Registry included those with ICD-9-CM codes 740-759, selected primary cancers, selected metabolic

State	Annual Births	Population Coverage	Legislation/Rule	Year Enacted	Outcomes Covered
					conditions, and FAS/FAE. Since the adoption of the Notifiable Conditions law in 2000, conditions subject to mandatory reporting are neural tube defects, orofacial clefts, limb deficiencies, abdominal wall defects, hypospadias/epispadias and Down Syndrome.
WI	~69,000	State	Wisconsin Statutes Rules	2000; 2003	Structural malformations, deformations, disruptions, or dysplasias; genetic, inherited, or biochemical diseases
WV	21,000	State	State Statute Section 16-5-12a	1991; 2002	congenital anomalies of ICD-9 codes 740-759, 760, 764, 765, 766

Note: These states either do not have a surveillance system or have suspended activities: AL, DC, ID, MT, OR, PA, SD, and WY.

Appendix C: NBDPN survey of state birth defects surveillance programs

This survey of state birth defects surveillance programs is intended to describe the current status of program operations, and to obtain projections of activities over the next two years. This includes issues related to funding, ascertainment, data quality, and prevention. Please answer all questions to the best of your ability.

I. General Information

1. For which State or Territory are you answering this survey?

State:

Other than a State or Territory (please specify):

2. What is the current status of your population-based birth defects surveillance system in your state or territory? (We mean a system that uses more than birth certificates for case identification).

- ◯ N/A: never had a birth defects surveillance system
- ◯ Currently developing or planning surveillance system is in the planning stage
- ◯ Previously stopped and restarted the birth defects surveillance system
- ◯ A consistently operational birth defects surveillance system
- ◯ Other (please specify)

II. Scope and legal authority of the birth defects surveillance system program

3. What proportion of your state's or territories' birth population is covered by your state's birth defect surveillance system (e.g.: statewide = 100%)

4. Which birth defects does your surveillance system currently identify?
Note: 7 critical congenital heart defects (CCHD) include hypoplastic left heart, total anomalous pulmonary venous return, Tetratology of Fallot, tricuspid atresia, pulmonary atresia with intact septum, truncus arteriosus, and d-TGA.

- ☐ NBDPN list of 46 conditions
- ☐ NBDPN list less than 46 conditions
- ☐ CCHD list of 7 conditions
- ☐ CCHD list less than 7 conditions
- ☐ EPHT list of 12 conditions
- ☐ EPHT list less than 12 conditions
- ☐ Developmental conditions (autism,muscular dystrophy, etc)
- ☐ Newborn/infant hearing loss (EHDI)
- ☐ Newborn genetic and metabolic screening panel

Comments:

5. Which pregnancy outcomes does your surveillance system categorize:

Pregnancy Outcome

	Yes	No
Miscarriages (spontaneous abortion) (<20 weeks gestation)	☐	☐
Fetal Deaths	☐	☐
Pregnancy terminations (any gestation)	☐	☐
Stillbirths (> 20 weeks gestation)	☐	☐
Live births	☐	☐

Comments (please specify)

6. Does the legal public health authorization language in your state or territory explicitly mandate reporting of birth defects or authorize the program to identify and collect information from health facilities?

- ◯ No (skip the next question)
- ◯ Yes
- ◯ Unsure

7. If yes, was the authority established through: (check all that apply)

- ☐ Legislation
- ☐ Rule or regulation
- ☐ Administrative action
- ☐ Other (please specify)

8. Does your state or territory have any laws, regulations, or authority that negatively impact (directly or indirectly) the surveillance function of case record abstraction (i.e., the ability to access medical records and other medical information?)

- ◯ No (skip the next question)
- ◯ Yes
- ◯ Unsure

9. If yes, how does it facilitate or hinder with regards to the abstraction of medical records?

10. Does your state or territory have any laws or regulations that negatively impact (directly or indirectly) surveillance function of the reporting of cases or the identification of cases to the program?

- ◯ No (skip the next question)
- ◯ Yes
- ◯ Unsure

11. If yes, how does it facilitate or hinder with regards to the reporting of cases?

12. Does your legislation require that your surveillance system secure consent from parent for IDENTIFICATION of potential birth defects cases:

- ◯ No (skip the next question)
- ◯ Yes
- ◯ Unsure

13. If yes (for identification):

- ☐ General opt out consent
- ☐ Cultural opt out consent
- ☐ Religious opt out consent
- ☐ Other type of consent (please specify):

14. Does your legislation require that your surveillance system secure consent from parent for RELEASING child's name for referral to services?

- ◯ No (skip the next question)
- ◯ Yes
- ◯ Unsure

15. If yes (for releasing child's name):

- ☐ General opt out consent
- ☐ Cultural opt out consent
- ☐ Religious opt out consent
- ☐ Other type of consent (please specify):

16. With respect to case identification, is your surveillance system primarily a dynamic or fixed system according to descriptions below?

Dynamic: you are permitted to perform case follow up and/or identify a diagnosis in the future regardless of the age of the case (e.g., as a result of new information or a new cytogenic test), in order to complete and improve the classification and listing of a birth defect in the surveillance system.

Fixed: you are permitted to perform case follow up and/or identify a new diagnosis or information within a set and fixed period of time (that may be related to the program's legal authority).

- [ ] Unsure if dynamic
- [ ] Dynamic, no restrictions
- [ ] Dynamic, some restrictions based on program objectives (e.g. special projects or closing out a cohort)
- [ ] Dynamic, however program resources or capacity prevents full utilization of this attribute
- [ ] Dynamic, other (please describe below)
- [ ] Unsure if fixed
- [ ] Fixed, restrictions based on legal authority
- [ ] Fixed, restrictions based on other administrative limitations
- [ ] Fixed, other (please describe below)

If other, please describe:

III. Resources and Funding

17. Please list the following staff members for your current birth defect surveillance program. Estimate the FTE per role rather than for an individual staff (since some individuals may perform multiple roles.

	Previously Had	Current FTE	Current FTE in-kind	Future Need
Director				
Program Manager - administrator				
Epidemiologist				
Statistician				
Clinical review, general				
Clinical review, Pedriatric cardiology				
Clinical review, Geneticist				
Clinical review, Dysmorphologist				
Clinical review, Epidemiologist				
Data manager				
IT or systems administrator (includes programming and security)				
Data abstractors (includes staff who perform medical record reviews for case verification)				
Referral to services				
Prevention, intervention, outreach - education				
Web based transactions and maintenance				
Other support staff				

Please specify "other" response or provide comments:

18. What percent do the following funding sources currently contribute to the annual costs of running your birth defects surveillance program? This is funding just for your surveilance activities and does not include any research grants, e.g. NBDPS. Check all that apply

	Percent (%)
Title V Maternal Child Health programs	
State General Funds	
CDC Birth Defects Surveillance Grants	
CDC Environmental Public Health Tracking Grants	
March of Dimes	
University or academic programs	
Other sources (specify below)	

Please specify "other" sources from federal, state, foundation, etc.

IV. Surveillance System Attributes

19. Does your surveillance system have specific instructions to complete each data field collected for each birth defect case?

- ◯ No
- ◯ Yes
- ◯ Unsure

20. Does your surveillance system utilize a clinical case definition (ie, inclusion and exclusion criteria) for each or selected birth defects included in the surveillance system? (This does not include case definition per legislation.)

- ◯ No
- ◯ Yes
- ◯ Unsure

Comment

21. If yes, what does your surveillance system use?

- [ ] Developed our own system
- [ ] NBDPN Abstractor Guidelines
- [ ] Data dictionary for each data element collected or ascertained
- [ ] NBDPS (National Birth Defects Prevention Study)
- [ ] NBDPS modified
- [ ] Other (please list):

22. What database and data analysis software does your surveillance system utilize? (Check all that apply)

	Database	Analytic
Epi Info	☐	☐
Access	☐	☐
Sequel server	☐	☐
SAS	☐	☐
GIS software	☐	☐
Maptitude	☐	☐
SaTScan	☐	☐
SPSS	☐	☐
Excel	☐	☐
Oracle	☐	☐

Other (please specify)

23. Does your program routinely geocode, or have access to geocoded data for birth defect CASES?

- ◯ No
- ◯ Yes
- ◯ Unsure

Comment

24. Does your program routinely geocode, or have access to geocoded data for the set of ALL LIVE BIRTHS (ie. denominator data) ?

- ◯ No
- ◯ Yes
- ◯ Unsure

Comment

25. What time periods does the program collect maternal residency information?

- ☐ Maternal residency at date of delivery
- ☐ Maternal residency at date of conception
- ☐ Maternal residency is collected during the pregnancy time period
- ☐ Other (please specify)

26. What disease classification coding system does your surveillance system utilize? (Check all that apply).

- ☐ ICD-9-CM
- ☐ ICD-9-BPA modified
- ☐ CDC/BPA (6 digit code)
- ☐ CDC/BPA modified
- ☐ ICD-10
- ☐ Other (please specify)

27. Does your program conduct specific prenatal surveillance to identify potential cases of birth defects that are prenatally diagnosed as the pregnancy is progressing during the current time period?

- ☐ No
- ☐ Yes
- ☐ Yes, from selected data sources
- ☐ Yes, for selected diagnosis
- ☐ Unsure

Comment

28. Does your surveillance system routinely collect (and record) information on Family history (1st degree (e.g. bio mother, bio father, siblings) or greater) of birth defects in relation to the index case.

- ◯ No
- ◯ Yes
- ◯ Unsure

Comment

29. If no, please identify reasons (check all that applies).

- ☐ No legislative authority or other permission
- ☐ Not part of case identification-data collection methodology
- ☐ Not part of surveillance program objectives
- ☐ Other (please specify)

30. Is the surveillance system able to identify siblings within your database by tracking through the biological mother?

- ◯ No
- ◯ Yes
- ◯ Probably, but only for siblings that are also born in our state
- ◯ Unsure

Comment

31. Does your surveillance system have the capacity to conduct follow-up studies of children with birth defects? Check all that apply.

- [ ] No
- [ ] Yes, under 1 year of age
- [ ] Yes, ages 1 thru 5
- [ ] Yes, ages 6 thru 10
- [ ] Yes, ages 11 thru 18
- [ ] Yes, over age 18 years
- [ ] Unsure

Comment

32. If yes, can you add a module to your surveillance system's database for a follow up study?

- ◯ No (skip the next question)
- ◯ Yes
- ◯ Unsure

Comment

33. If yes, do you already have a module for a follow up study in your surveillance sytem's database?

- ◯ No
- ◯ Yes
- ◯ Unsure

Comment

34. Does your surveillance system have the capacity to link birth defects data to death certificate data to study survival of live born infants with birth defects?

- ◯ No
- ◯ Yes
- ◯ Unsure

Comment

35. What data sources does your surveillance system utilize to identify potential cases?

	Use for Case Finding (Active Case Ascertainment)	Frequency Case Finding	Receive/Get access to case reports from the data source	Frequency Receiving case reports from the data source
Prenatal Pathology Reports				
Prenatal diagnostic centers (MFM)				
Prenatal Ultrasound database				
Prenatal Genetic counselors				
Prenatal Obstetricians				
Prenatal sites Logbooks				
Prenatal Cytogenetic Laboratories-in state (free standing)				
Prenatal Cytogenetic laboratories-out of state (free standing)				
Prenatal Hospital based cytogenetic lab-in state				
Prenatal Hospital based cytogentic lab-out of state				
Prenatal Emergency Room				
Postnatal Delivery hospital discharge (in-patient)				
Postnatal Delivery hospital discharge (out-patient, includes emergency room)				
Postnatal Labor &Delivery Logbooks				
Postnatal Emergency rooms				
Postnatal Newborn Nursery Logbooks				
Postnatal NICU Logbooks				
Postnatal NICU Reports				

Postnatal Pediatric (tertiary) hospital discharge (in-patient)				
Postnatal Pediatric (tertiary) hospital discharge (out-patient, includes emergency room)				
Postnatal Pathology Reports				
Postnatal Cytogenetic laboratories in-state (free standing)				
Postnatal Cytogenetic laboratories out-of-state (free standing)				
Postnatal Hospital based cytogenetic lab-in state				
Postnatal Hospital based cytogenetic lab-out of state				
Postnatal Newborn Hearing Screening				
Postnatal Newborn Genetic and Metabolic Screening				
Specialty clinics: hospital based or other outpatient clinics				
Pediatric cardiology				
Pediatric genetics				
Pediatric orthopedics				
Pediatric urology				
Pediatric Developmental				
Pediatric surgery				
Pediatric neurology (e.g. NTDs)				
Pediatric Orofacial				
Other specialty clinic (specify below)				
Vital Records				
Birth certificates				
Fetal death certificates				
Death certificates				
Other type of vital record (specify below)				
WIC				
Physician reports				
Community Health Centers				

Local Health Departments				
Admin Data Sets- Registries				
Hospital discharge data set				
Medicaid data set				
Children with Special Health Care Needs data set				
National Death Index				
Environmental data set				
EHDI Registry				
Other data sets-registries (specify below)				

Please specify "other" response or provide comments:

36. Please select from the drop down menu,

	Responses
Do data abstractors go to the delivery hospital to abstract the medical records of the mother?	
Do data abstractors go to the delivery hospital to abstract the medical records of the fetus/infant?	
Do data abstractors go to the tertiary hospital to abstract the medical records of the infant?	
Do data abstractors request medical records from the mother's obstetric care provider to obtain additional information on the mother's pregnancy?	
Do data abstractors request medical records from the child's primary pediatric care provider to obtain additional information on the infant?	

Comment

37. What type of electronic transaction method is used to receive a reported birth defect case or for case identification or case finding?

- [ ] Web based health information ports
- [ ] Secure File Transfer
- [ ] Internal health department electronic upload or transaction:
- [ ] External electronic download transaction (describe below)
- [ ] Other type of download or transaction (describe below)

Please describe.

38. What types of electronic medical records are used by the hospitals and other health care settings that the surveillance system accesses for data collection or case identification?

	% of inpatient hospitals using this method	% of other health care settings using this method
Electronic medical record - primary documents or primary documentation is scanned.		
Electronic medical record - documentation is keyed in a prescribed template or format.		
Electronic medical record - information is keyed in an ad hoc or verbatim format (e.g. bedside or mobile laptop or Ipad as patient care) is administered.		
Other (specify below)		

If selected "other", please specify.

39. What methods of access are available to the surveillance system to retrieve or access the electronic medical record information?

	% of inpatient hospitals where you use this method	% of other health care settings where you use this method
Web based health information ports		
Secure File Transfer		
Remote access		
Hospital site access to the electronic medical record		
Electronic transaction		

40. How has the electronic medical record (EMR) affected the level of detail of clinical information when compared to the historical hard copy medical record?

The level of detail of clinical information as a result of the EMR has:

- [ ] Stayed the same
- [ ] Increased
- [ ] Varied
- [ ] Decreased
- [ ] Unsure

Comment

41. How has the electronic medical record affected the level of completeness in the content when compared to the content in the historical hard copy medical record? (e.g. the types of information reports)

The level of content completeness as a result of the EMR has:

- [ ] Stayed the same
- [ ] Increased
- [ ] Varied
- [ ] Decreased
- [ ] Unsure

Comment

42. Who is responsible for assigning a disease classification code of the major and minor birth defects to a birth defects case?

- ☐ Data abstractor
- ☐ Certified hospital coder (e.g. Registered Health Information Technicians - RHITs or Registered Health Information Administrator - RHIA)
- ☐ Trained disease coder
- ☐ Clinician or clinical reviewer
- ☐ Epidemiologist
- ☐ Other (please specify)

43. What type of background or experience have you utilized when hiring a data abstractor or other staff who will review-read medical records information for case identification or case verification?

	Previous hire	Current hire
Health information management technology with RHIT credential	☐	☐
Health information management technology with RHIA credential	☐	☐
Health information management technology without RHIT or RHIA credential	☐	☐
RN-Nursing	☐	☐
Nurse Consultant	☐	☐
Public health nurse	☐	☐
Physician Assistant	☐	☐
Other health care professional	☐	☐
None, trained the abstractor in house	☐	☐
None, the program has no abstractor	☐	☐

Please specify or provide comments

44. Does your surveillance system utilize data quality procedures to assess accuracy of the birth defects case status (a true case)?

	No	Yes	Sometimes	Unsure
Dysmorphologist clinical reviewer	☐	☐	☐	☐
Geneticist clinical reviewer	☐	☐	☐	☐
Cardiologist clinical reviewer	☐	☐	☐	☐
Epidemiologist clinical reviewer	☐	☐	☐	☐
Pediatric clinical reviewer (this is on the personnel list)	☐	☐	☐	☐
Medical records or health records review of the documentation	☐	☐	☐	☐
Quality of the data source (e.g. pathology, cytogenetic lab,genetics clinic, specialty clinic, etc)	☐	☐	☐	☐
Corroborating procedure that is linked to the birth defect diagnosis	☐	☐	☐	☐
Data quality assurance procedure performed by staff	☐	☐	☐	☐
Other (specify below)	☐	☐	☐	☐

Please specify "other" response or provide comments:

45. Does your surveillance system have the ability to classify birth defect cases into isolated, multiple, and syndromes (not using disease codes)?

- ◯ No
- ◯ Yes
- ◯ Sometimes
- ◯ Unsure

If checked yes or sometimes, please explain the process/system used:

V. Assessment of surveillance system-program functions

46. The following statements ask you to assess activities related to the functions of your birth defects surveillance program-system. You are asked to respond to a series of statements in relation to changes over the past 24 months, and anticipated plans over the next 12-24 months.

	During 2010-2011	In 2012 thru 2013
CASE IDENTIIFICATION: The number of data sources that the birth defects surveillance system uses for case identification		
The level of surveillance activity related to the identification of prenatally diagnosed cases		
The level of case finding for case identification		
The number of cases assigned per data abstractor for case finding or case verification		
The number of structural birth defects that the surveillance system ascertains based on the current disease coding system		
ANALYSIS and DATA UTILIZATION: The level of consistent and ongoing monitoring statistical analyses performed by the surveillance systems		
The level of Statistical analyses performed for program evaluation		
The level of statistical analyses performed for other reasons		
The number of research activities for collaborative data projects using surveillance data		
The number of cluster investigations or responses to community inquiries using surveillance data		
The number of research activities for non collaborative data projects using surveillance data		
The amount of information disseminated using printed materials		

The amount of information disseminated using web based materials		
The amount of information disseminated using other education tools, or public outreach		
The amount of information disseminated using social media (e.g. facebook, twitter, text messaging, etc)		
DATA QUALITY: The use of data quality evaluations to measure the timeliness of the case identification process		
The level of case verification by medical records review to assess the accuracy or completeness of diagnostic information in a case record-abstract		
The use of data quality procedures to monitor the frequency of reporting from data sources		
The use of data quality procedures to assess the number of data sources that contributed to the identification of a respective birth defects case		
The use of data quality procedures to assess the unique contribution from a data source to the identification of respective birth defects case		
The use of data quality procedures to evaluate errors in data abstraction during data collection activities (case finding or case verification)		
INTERVENTION and PREVENTION: The numbers of children identified from the birth defects surveillance program-system for referrals to services		
The level of prevention or intervention activities for FAS		

The level of prevention or intervention activities for pre-conception risk factors		
The level of recurrence risk prevention or intervention for NTDs		
The level of prevention or intervention collaborations with partners		

Health Service Utilization

VI. Utilization of health services and economic data

47. Does your surveillance program currently have access or link to cost/charge or health care service* data during the FIRST YEAR of life?

(*Note: Health care service data could include comprehensive information about exams performed, well childcare visits, immunizations, physician / outpatient visits, hospital admissions, treatments and procedures, etc. These type of data do not necessarily include associated dollar amounts.)

	ACCESS TO	LINK TO
No	☐	☐
Yes, Cost/charge data	☐	☐
Yes, Healthcare resource utilization data	☐	☐
Unsure	☐	☐

Comment

48. If yes for access or link to cost/charge data or healthcare resource utilization data during the first year of life, which data sources?

	ACCESS TO	LINK TO
Medicaid paid claims data	☐	☐
Hospital discharge data (only for birth hospitalization)	☐	☐
Hospital discharge data (include more than birth hospitalization)	☐	☐
HMO or private insurance	☐	☐
Physician billing system	☐	☐
CHIP	☐	☐
Other (specify below)	☐	☐

Please specify other type of billing or payer data base.

49. If yes for access or link to cost/charge data or healthcare resource utilization data during the first year of life, which data sources?

	ACCESS TO	LINK TO
Medicaid paid claims data	☐	☐
Hospital discharge data (only for birth hospitalization)	☐	☐
Hospital discharge data (include more than birth hospitalization)	☐	☐
HMO or private insurance	☐	☐
Physician billing system	☐	☐
CHIP	☐	☐
Other (specify below)	☐	☐

Please specify other type of billing or payer data base.

50. Does your surveillance program currently have access or link to cost/charge or health care service* data BEYOND the first year of life?

(*Note: health care service data could include comprehensive information about exams performed, well childcare visits, immunizations, physician / outpatient visits, hospital admissions, treatments and procedures, etc. These type of data do not necessarily include associated dollar amounts.)

	ACCESS TO	LINK TO
No	☐	☐
Yes, Cost/charge data	☐	☐
Yes, Healthcare resource utilization data	☐	☐
Unsure	☐	☐

Comment

51. If yes for access or link to cost/charge data or healthcare resource utilization data BEYOND the first year of life, which data sources?

	ACCESS TO	LINK TO
Medicaid paid claims data	☐	☐
Hospital discharge data (only for birth hospitalization)	☐	☐
Hospital discharge data (include more than birth hospitalization)	☐	☐
HMO or private insurance	☐	☐
Physician billing system	☐	☐
CHIP	☐	☐
Other (specify below)	☐	☐

Please specify other type of billing or payer data base.

52. How has your program utilized cost or charges data ?

- ☐ Never utilized
- ☐ Economic analysis (such as cost-benefit analysis or cost-effectiveness analysis)
- ☐ Program planning-justification
- ☐ Needs Assessment
- ☐ Legislative request
- ☐ Other, please specify.

Concluding Page

VII. Concluding Comments

53. What three areas or activities are of highest priority for your birth defect surveillance program-system during 2012 thru 2013?

1.
2.
3.

54. What are three most important challenges your birth defects surveillance program-system will face during 2012 thru 2013?

1.
2.
3.

55. Please list any comments:

Thank you for completing this survey. If you have any questions, please e-mail nbdpn@nbdpn.org.

Comment Page

56. Please list any comments:

Thank you for completing this survey. If you have any questions, please e-mail nbdpn@nbdpn.org.

Appendix D: Semi-structured Interview Guide

Participant name and state:

_____

1) *Recruit and schedule phone interview:*
   Initial call to birth defects surveillance program directors from states selected using the sampling scheme

   Hi [Name],

   I'm currently enrolled in the Doctor of Public Health (DrPH) Program at the University of Illinois, Chicago, and have decided to focus my dissertation on the current practice of population-based birth defects surveillance programs in the United States. I'm hoping to better understand how our programs are poised to address current and emerging needs.

   I am analyzing the 2012 NBDPN survey that included many questions on the infrastructure and practice of birth defects programs. In addition, I'm interviewing nine program directors to further discuss the issues raised in a recently released MMWR article, "CDC's Vision for Public Health Surveillance in the 21st Century"; these include standards, workforce, IT, as well as data access, use, and analytic capacity.

   Your program has been selected for the qualitative data collection. I'm hoping that you'll be willing to talk to me about your thoughts on these issues as well as the intended purposes of your surveillance program.

   [Response from State Birth Defects Program staff]

   Please let me know when you are available to talk. The call should last about 90 minutes.

   [Respond to any questions, discuss schedule availability, etc.]

   I will send you an e-mail confirmation along with a link to the MMWR article.
   I look forward to talking to you soon.

   Thank you.

2) *Semi-structured telephone interview:*

   Hi [Name],

   Thanks again for your willingness to talk to me about your program and your thoughts on how birth defects surveillance programs can better address the issues outlined in the MMWR article.

[If clarification is needed based on this person's response to the 2012 NBDPN Program Survey, ask here.]

Let's start with the intended purposes of your surveillance system. Please describe the original purposes and if they have changed through the years.

[Discuss intended purposes of the program.]

Now, we'll shift to discuss the issues outlined in the MMWR article. I hope you had a chance to review it. It covered five areas that I'm interested in examining specifically for birth defects surveillance: standards, IT, workforce, data access and use, and analytic foundation. The MMWR article does address global surveillance but we are going to focus this discussion only on domestic activities.

I plan to discuss each of these in details and get your thoughts.

Let's start with lexicon and standards…

[Move through the other issues:

- Information science and technology
- Skilled workforce
- Data access and use
- Strong data analytic foundation]

Do you have any other comments?

I appreciate you taking the time to talk to me today. If you have other thoughts, please feel to call or e-mail me.

Thanks again.

Appendix E: Codebook for Semi-structured Interviews

Code	#	Description	Example
Question 1: Intended purposes of the surveillance system			
Impetus (Driver)– need better data	1.1	A reason why a program got started was because of a lack of data to address community needs	"…as a result a report that was released from the [state] legislature that had studied a number of communities in [state] and looked at disparities in that community with health outcomes… some of the recommendations included the need for the surveillance systems that did not exist at the time, and one of those was the birth defects registry."
Impetus (Driver) – funding opportunity	1.2	A reason why a program got started was because of a funding opportunity	"So using the findings from that report the department put together what we call a legislature budget request and requested funding from the [state] legislature to begin a registry. And we were actually funded and that's how we got started."
Impetus (Driver) – helping families	1.2a	To improve the well-being families and babies; to reduce infant mortality	"It was set-up as a response to [state] infant mortality."
Purpose - partnership engagement	1.3	The role partners played in shaping the intended purposes of the program	"[The partners] have been very involved in what are the priorities and activities of the registry. They have been there when we had some setbacks with regards to funding; we had a few years where funding was reduced and we had to reset our priorities and they were there to help guide us as we made decisions about what we could and couldn't do with reduced funding. They advocated on behalf of the registry and actually last year that funding was restored."
Purpose – support and perceived value	1.4	The support and perceived value of the birth defects program from partners and communities Show the value of the data provided by the surveillance program	"They [Partners] hear from families that it is valuable and they see it as valuable." "The registry has been sustained because of an interest to respond to public concerns. You know that often time, there is nothing there, but you are really assuring them that it's not different from other areas. Often times, it's assuring to be able to look at the concern is a valuable tool."
Purpose – activities	1.5	The activities that the program was established to do	"…conceived as a surveillance system (BD and children with adverse outcomes) and provides case management services of families affected by such pregnancies with the goal of prevent problems arising from these adverse outcomes."
Purpose - evolving	1.6	The objectives/purpose of what the program should be doing changing over the years	"..evolved over the years because we have moved from being solely passive to incorporating some active surveillance components tied into opportunities that we have received from CDC to environmental public health tracking."
Resources	1.7	Availability or lack of availability of resources	"We were aware of centers of excellence like in [State] and other places, but we couldn't afford that."

Code	#	Description	Example
Assessment of approach	1.8	The program assesses or evaluates its practice and implemented changes.	"We tried to take a comprehensive approach over the last few years that kind of looking at what are the strengths and what are the limitations with regard to timeliness and data quality of those issues."
Reporting and accountability	1.9	The program has to report to some authority	"We do an annual report for an oversight board. There are specified people who nominate people to attend [representative from various programs]."

Code		Description	Example
Question 2: How prepared are birth defects surveillance programs to address the major considerations identified in "CDC's Vision for Public Health Surveillance in the 21st Century"?			
Question2a: Lexicon and conceptual framework			
Lexicon - jargons	2a.1	Jargons used by surveillance programs are hard to translate to partners	"They don't have a good understanding about how one activity can complement the other and how we can address an emerging issue, such as what we are going to try to do with the critical congenital heart defect."
Lexicon - similar understanding	2a.2	Similar understanding of common lexicon and concepts (everyone on the same page)	"Lexicon to me refers to everyone on the same page, doing the same thing. There are a lot of differences with birth defects. This was a problem with cancer. The one thing cancer has that birth defects lack is that they end up making their diagnostic through microscopic pathologic exam. That's the gold standard for cancer, but we can't do that for birth defects."
Foundation of public health	2a.3	Collecting data serves as basis or foundation of public health	"Literally, surveillance and tracking are the foundation of what we do with regards to collecting information on disease including birth defects and then using that information, analyzing and interpreting and trying to develop interventions that we can evaluate and have then a positive impact on reducing morbidities and mortalities."
Reframing how to think about surveillance terms	2a.4	Reframe how we think about the terms we use, e.g. opt-in/opt-out, active/passive	"To the degree that we can, we need to make sure we have population-based data. It is not opt in / opt out. It's not a question of whether you want to or not... There are some things you need to hold as principal, but we have to do so in a politically palatable way, have a clear need (from the public) and use only for what we said we need it for."

Code		Description	Example
Clinical diagnosis and practice affecting public health surveillance	2a.5	Diagnosis and practice of birth defects affecting the information available for public health surveillance Understanding and using similar diagnostic descriptions of conditions	"This is an example of how simply settling in on a diagnostic description of conditions can impact health care but also on the ability to conduct surveillance in a uniform manner so that the information is uniform from doctor to doctor, hospital to hospital." 'The other thing birth defects can do with quality of diagnosis is severity. For example, need detailed severity of conditions. The more specific can get, then the better you can look at it more in medical risks, a lot of potential on how careful to classify clarity of conditions."
Standard codes and messaging	2a.6	Standard codes and messaging such as ICD, SNOWMED, etc.	"We talk about SNOWMED and ICD-10; we are moving into a new coding system and suddenly there is more specificity, we need to learn how to use it. We need to push it to make it more relevant."
Birth defects surveillance guidelines and standards	2a.7	Development and use of national guidelines and standards for birth defects (NBDPN)	"…having those standards and having those guidelines provide individuals like me at the state level with good reference, framework documentation to take to the leadership of my agency whenever this becomes a topic within my own state health department."
National efforts	2a.8	National resources such as National Birth Defects Prevention Network (NBDPN), CDC, etc.	"I think that one of the things that is really helping is the fact that we do have this network this partnership now, and that CDC is committed to pulling together this kind of information. I mean those help tremendously to have those clinically correct illustrations of infants with conditions and that's one of the things that would help. I hope that CDC would continue to work on and maybe to develop some illustrations for other conditions. And those are extremely useful for the work that we are doing, and we are able to include those and incorporate information around those illustrations in a way that we are finding more appealing to health care providers and families."
State sharing and collaboration	2a.9	States sharing knowledge/learning from each other	"…learn from other states"
Proactive outreach	2a.10	Proactively using data and information to outreach to community	"We are planning on developing letters and data tables and sending them out to each of the county medical associations and the some of these professional specialties and say here we are, this is what we have, how can we work together?"
Tools	2a.11	Tools and resources	"…to develop some abstractor manuals and manual tools to help review medical records and to make decisions about what the data in the medical records mean."

Code		Description	Example
Communications	2a.12	Messaging and communication of public health surveillance	"When you publish something, you need to define things clearly, especially if they are not in a scientific journal. Your politicians, stakeholders are not epidemiologists. Instead of saying population-based, talk about covering all babies born in [the states]. Trying to effectively communicate is one of the hardest things we do."
Question 2b: Information sciences and technological advances			
Preparation from state health department	2b.1	State health department not ready for changing technology	"I don't even know how much public health is providing with regard to electronic medical records. The data exchange activity is lead by another department in our state."
Changing how data are obtained	2b.2	Health data being used for public health is changing	"As we move into informatics that is coming, the line between active and passive will disappear. It's what you do with the data that matters not how you get possession of the data (come in)."
Information in electronic medical records (EMR) – Positive	2b.3	Positive impact of medical information in an electronic format	"Generally, we accept if a doctor says the results, then we will accept that as verification of the condition. At that level of verification, most of the hospitals give us what we need. There are some that send us a CD with PDF files. We can search."
Information in EMR - negative	2b.4	Negative impact of medical information in an electronic format	"We are doing more of going out to hospitalsdirectly and reviewing NICU department logs, radiology, and other datasets in the hospitals to identify data. I think a lot of information that will be lost. As a state and as a Network, we need to evaluate the implication of the lost of the data and what is the value of electronic medical records. Are there other ways to provide information that we are not getting from electronic medical records? We need to rethink what we are doing."
Positive – remote access	2b.5+	Positive effects of remote access	"We have gained remote access to the information related to birth defects. This has allowed us to view the medical records from our offices. This has cut down the time that we need to travel to the hospitals. That part is good."
Negative – remote access	2b.6-	Challenges with remote access	"They each have their own search function and it can get complicated."
Evaluate data source / Quality assurance to access to information	2b.7	Examine the data sources and information in electronic medical records periodically	"The role of electronic medical records is still in the preliminary phase. It is something that we are paying attend to. We'll probably have to evaluate it yearly to see progress. All the hospitals in our state are using different systems."

Code		Description	Example
Cost	2b.8	Resources to build a system	"One of the things I see happening is, if looking at public health surveillance, one of the challenges is to try to build a system that we can afford. We need to develop a system that is not too burdensome on the people who provide data. You come up with methods that seem to work. You design low cost, impact types of system."
Health information exchange	2b.9	Access to medical information through health information exchange	"We have not explored Health Information Exchanges. I think there is potential, but we have not fully worked out on it. I hope to learn from other states."
IT opportunities	2b.10	Changing data provides opportunities to enhance data utilization	"Given the fact that there is so much change going on with electronic medical records and the data system we would love to look at our data over a life span and something that we are all moving towards so that this brings all things in."
Barriers to IT integration	2b.11	Barriers to integration of IT systems	"It is also the political will. It's a territorial issue around individual programs."
Future is near (potential)	2b.12	Potential for the systems to "talk" to each other and automation	"I'm seeing is that within 10-12 years, the data that will become available and mineable for public health surveillance, is just going to explode, very reliable, right out of the doctor's mouth."
Systems planning	2b.13	High level systems planning	"We have centralized IT support. But it is not like we ever sit down with them to have large-scale, high-level discussions. Instead of having stand-alone data systems, is there any possibility to have a warehouse, where a variety of surveillance data can be integrated and access by the birth defects registry?"
Thinking outside the box	2b.14	Thinking of what is possible with new data (not on what need now, but what can get in the future)	"What we need to do is to think about ways, best place to get information, get as much information as we can from systems, so that we can shift through the information (for proper processing and evaluation). They are not trying to take advantage of what is available."
Investment into system development	2b.15	Federal money available now to develop systems	"The interest is being driven by federal requirements, Medicare/Medicaid, throwing around a lot of money. What I worry is that the incentives money will disappear. What will continue is that CMS will continue to require doctors to use approved certified software."
New IT terminology	2b.16	IT terms that public health need to understand	"We need to worry about what they call on-boarding and validation. What to do with them? How much detail to check? If you find problem, what do you do with it? What is the path the data follow? "

Code		Description	Example
Question 2c: Surveillance work force			
Specialized IT skills	2c.1	Informaticians People who specialize in public health informatics	"The systems that are informatics along with transport issues are very technical. You need support to health department with informatics team; syndromic surveillance has it this way. What we are doing is simple to them but we need to tap into their infrastructure."
Vision from leaders	2c.2	Vision from leaders and managers of where the program is going	"Absolutely we need priority setting, strategic planning and vision."
Cross-training	2c.3	Staff who can perform multiple tasks using different skill sets	"It's the program that needs to learn programmer terms. They don't. The programs, mainly nursing staff, are not interested in learning computers. As younger people enter and move toward management positions, then there is more innate understanding of how computers work."
Active participation beyond job duties	2c.4	Active participation in decision making	"…what has been helpful is to equip people in meeting that might not directly impact their piece [job to have a better understanding of the program]."
Clinicians	2c.5	Staff with clinical background	"We would have to maintain contract relations with clinicians and when we need it, we need to reach out to clinical experts in our state or at CDC for input."
On-going training	2c.6	Training staff on an on-going basis	"We don't offer on-going training to our staff…"
Hiring	2c.7	Ability to hire or hire the types of personnel needed	"One of the things we are tapping into is the abstractors who are working for the allied programs, like mortality review."
Lack of staff	2c.8	Sufficient number of staff to carry out activities	"And I'm not sure that state health departments have the staff to deal with some of these complex issues you need a whole lot more than an epidemiologist to understand what all of this data means together and then to turn it into some action or policy or prevention and then to be able to evaluate it."
Workforce skills	2c.9	The type of workforce (people trained) to carry out necessary tasks	"But we lack individuals who can make these systems and integrate these systems and allow us to use the information for a number of surveillance activities."
Expertise from university and CDC	2c.10	Utilizing expertise from the universities and CDC	"We have an on-going collaborative relationship with the University of South Florida and this partnership with March of Dimes and CDC. Those partnerships and data sharing agreements. We are beginning to see publications from that collaboration."

Code		Description	Example
Multi-state collaborative projects / collaborations with different skills	2c.11	Synergistic collaborations with different skills	"One thing that NBDPN has been doing and I have never seen work so well and so effectively is the Data Committee. The teams they put together have no comparison. It's a great example of pulling collaborators together, people with different skill sets. This where birth defects surveillance is above the other programs: forming a team of people with different abilities to do analytics projects."
Opportunities	2c.12	People at right places and identify opportunities	"There are a lot of opportunities. We have to reach out to a number of channels to promote ourselves. It is a limitation of state health departments. We don't do that. We have all these data, sitting in draw. We don't have staff to analyze and interpret the data for policy and evaluation."
Question 2d: Access and use of public health surveillance data covering legal, policy, ethnical, regulatory, and practice concerns related to data sharing			
Data access	2d.1	Positive effects of data access Boundaries for data access Data access beyond traditional case finding sources, e.g., FERPA, criminal system	"But I know that we worried about having the amount of health info the state agencies and education agencies are gathering and holding and using with regards to their individual health information their child health information, and so there are real concerns for both for the department of health and the general public and the health care providers about pulling all of this information together and what is done with it and how it is used."
Legislative authority for birth defects using birthing / pediatric sources	2d.2	Legislation requiring the reporting of data/health information from providers	"[State] has not been a state that is willing to fine hospitals/providers for not reporting, even for infectious diseases."
Legislative authority for long-term outcomes	2d.3	Authority to obtain data on children with birth defects beyond the birthing period	"…our case definition is 20 weeks gestation to the first year of life, so those are real limitations for us at least for birth defects surveillance. What we can do with it we have the authority and statue to do public health research public health education, so we count some of the work that we are doing now where we are looking through the 10 -12 years of our dataset that we have to look at what is going on with infants that are born with birth defects over that course or that span."
Data warehouse	2d.4	Centralized place for data from multiple sources and programs	"…We have a data warehouse. We feed information into the database."

Code		Description	Example
Relationship with reporting sources	2d.5	Relationship with reporting sources at hospitals, clinics, etc. to ensure they report or understand the need of the program to access the medical data	"…how to work with hospital staff. We are going around to emphasize the importance to hospital staff. But I think that there is a lot of staff turnover, and hospital staff sees that it is just another thing they have to do without getting the bigger picture of why they have to report to us. We are not just trying to get information, but we do it to inform public health. We go out and teach periodically but with a high staff turnover, it's is hard to keep up."
Public concerns about government having access to data	2d.6	Concerns from the public about government having access to data, e.g. too much data, types of data	"There seems to be a growing concern by the public about state agencies even federal agencies having this much information, this much health information on individuals, families and their children."
Changing primary data sources	2d.7	Data sources are changing	"I don't know what the implications are with birth defects, but our cancer registry has been able to reach out to physicians who are diagnosing and treating patients because more and more those patients are not ending up and receiving service in a hospital. I don't know if we have a good understanding of how that is occurring in kids with birth defects and can we get direct reports from physician offices and what is the value of that data, what does it mean for the surveillance program, what does it bring?"
Politically palatable (acceptable)	2d.8	Agreeable / acceptable politically	"This was a well-done piece, but the one thing missing from confidentiality is that we are almost all governmental entities. Whatever we do, it is politically palatable. …. The issue of politically palatable is important and if the public turns against us, then we are not going to be able to do what we do."
Privacy protection	2d.9	Protect privacy of individuals public health surveillance monitor	"We have an overriding obligation to get the maximum value out of the data and also an overriding obligation to protect the privacy and confidentiality of the data to the maximum possible."
Concept of monitoring	2d.10	Perception of monitoring	"I can see how surveillance and registry turn off people when discuss in abstract. I don't know if there are better terminologies. Even if we come up with different terms, it would quickly associate with the same thing as soon as people know what it meant. It's not the terms but the concept of monitoring. Anything terminology that makes people uncomfortable, such as surveillance or something being wrong with a child, will make people uncomfortable."
Question 2f: Analytic challenges such as database management			

Code		Description	Example
Databases / data systems	2f.1	Databases for data collection of variables	"We can't continue to have stand alone systems. We know that is not going to happen with health exchanges. We need staff that can integrate these systems and have the ability to construct the systems."
Data steward commitment to data integration / sharing	2f.2	Leadership committed to data integration	"They had a commitment to create a maternal linked file."
New data sources	2f.3	New data sources opened as data become more available	"And when you start pulling in data from Department of Education (linked data); for example, if you get demographic information, they have their own coding system, how do you interpret and have the data to complement the current sources?"
Limitation – data analysis	2f.4	Limited people to analyze the amount of data available for public health action	"We are at least getting the linked file, but we have tremendous limitation in analyzing the data (people who can evaluate the data) and turning the information into public health action."
Linking datasets	2f.5	Different datasets link	"We are linking our birth defects registry to a number of data sets. Now, the state has created a maternal and child linked file."
Bringing in expertise from university / CDC	2f.6	Partnering with experts from universities and CDC	"We partnership with university. They bring unique skills that we would never have, along with CDC. There are health economists, clinical expertise, GIS expertise. That is all beyond what we are able to do. I think this is really key that health departments develop the relationship with the university and clinical experts."
Priority setting	2f.7	Setting priorities	"One of things I want to do is to set priorities for our program."
Data interpretation and utilization	2f.8	Interpreting the analyzed data for public health action	"We are trying to develop a series of fact sheets that actually takes the data and interprets them in a way that are useful ..."
Tools for families	2f.9	Materials and tools developed for families of affected children	"…actually getting ready to develop a resource document that could be provided to families that says "here is where you can get services in your area."
Across the lifespan	2f.10	Changing health care practice for individuals living longer	"So I think there is a tremendous opportunity to learn from individuals who are living longer than they ever have, and to re-educate the medical community about how you are going to care for these individuals as they turn 20, 30, 40, 50, 60, 70 years old. I think there is a whole new set of information interventions; you know things that need to happen that haven't been happening."

Code		Description	Example
Data quality	2f.11	Strategies/activities performed by program to ensure/improve quality of data collected	"And, we have an entire quality assurance program; we do re-case-finding, re-case abstraction, etc. I think the program is a leader in this; he is trying to get staff to write about it. We do all sorts of data quality assurance; but I still worry about data quality. Overall, the program does a pretty good job, but areas that can still be improved, including pregnancy outcomes."

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Vita

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Education:

1989-1994	University of California, Davis Davis, California	B.S. Nutrition Sciences, 1994
1994-1996	University of California, Berkeley Berkeley, California	M.P.H. Public Health Nutrition, 1996

Professional Experience:

11/2000-present	Birth Defects Branch, Division of Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, Georgia *Public Health Analyst/Project Officer*
4/1999–11/2000	Battelle Memorial Institute, on-site at the Centers for Disease Control and Prevention, Atlanta, Georgia *Health Research Scientist/Program Coordinator*
4/1997–1/1999	Santa Clara County Public Health Department, Maternal and Child Health, Santa Clara, California *Perinatal Services Coordinator* *Health Education & Nutrition Consultant*
8/1996–7/1997	Santa Clara County Public Health Department, Child Health and Disability Prevention Program, Santa Clara, California *Health Education Specialist*

Awards:

2012 CDC Domestic Excellence in Surveillance and Health Monitoring Award to the Critical Congenital Heart Defects Working Group (member)
2005 National Birth Defects Prevention Network President's Award – honoring an NBDPN member who has made significant contributions important to the mission and goals of the National Birth Defects Prevention Network
1996 U.S. Public Health Services Traineeship
1995 California Dietetics Association Scholarship
1995 Etta Dobbs Award for Outstanding Public Health Nutritionist
1994 Kristen Maxwell Memorial Award Outstanding Student, UC Davis

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1. **Mai CT**, Rickard R, Block S, O'Halloran A, Flood T, Copeland G, Stanton C, Wang Y, Contreras D, Ehrhardt J, Kirby R for the National Birth Defects Prevention Network. Validation of Transposition of the Great Arteries Using Surgical Procedure Codes. National Birth Defects Prevention Network 16th Annual Meeting, February 25-27, 2013, Atlanta, GA.
2. RS Rickard, J Donnelly, RS Kirby, **CT Mai**, D Law, C Druschel, A Caton for the National Birth Defects Prevention Network. Improving the early detection of biliary atresia in the United States: a missed opportunity? Accepted for 17th Annual Maternal and Child Health Epidemiology Conference, December 14-16, 2011, New Orleans, LA.
3. Leslie deRosset, Alina Flores, **Cara Mai**, Onameyore Utuama. Using a Promotora model to increase folic acid consumption among Latinas in North Carolina: It Really Works! 3rd National Summit on Preconception Health and Health Care, June 12-14, 2011, Tampa-St. Petersburg, FL.
4. Jennifer Donnelly, Russel Rickard, **Cara Mai**, Russell Kirby, David Law, Charlotte Druschel, and Alissa Caton. Diagnosing Biliary Atresia in the United States: Are we losing the race against the clock? National Birth Defects Prevention Network 13th Annual Meeting, March 8-10, 2010, National Harbor, MD.
5. SE Parker**, CT Mai**, MJ Strickland, RS Olney, RS Rickard, RE Meyer, L Marengo, Y Wang, and SS Hashmi. Multi-State Study of the Epidemiology of Clubfoot, 2001-2005. National Birth Defects Prevention Network 12th Annual Meeting, February 23-25, 2009, Nashville, TN.

6. **Cara T. Mai**, Assia Milller. Birth Defects: What's in a Name? National Birth Prevention Network 11th Annual Meeting, February 11-13, 2008, Washington, DC.
7. CJ Alverson, **Cara T. Mai**, Adolfo Correa. Requirements for Reliable Estimation of Prevalence of Birth Defects. National Birth Prevention Network 11th Annual Meeting, February 11-13, 2008, Washington, DC.
8. Sheree Boulet, Quanhe Yang, **Cara Mai**, Russell Kirby, Julianne Collins, Timothy Flood, Robert Meyer, Mark A. Canfield, Joe Mulinare, James M. Robbins. Trends in the Post-Fortification Prevalence of Spina Bifida and Anencephaly in the U.S. Accepted for presentation at the Twelfth Annual Maternal and Child Health Epidemiology, December 6-8, 2006, Atlanta, GA.
9. Julianne Collins; Mark Canfield; Kay Pearson; Russell Kirby; Amy Case; **Cara Mai**; Judy Major; Barbara Frohnert; Jane Dean; Russel Rickard; Joe Mulinare. Preventing the Recurrence of Neural Tube Defects in the United States: Current Status and Recommendations. National Conference on Health Promotion, September 12-14, 2006, Atlanta, GA.
10. Mark A. Canfield, Nataliya Yuskiv, Margaret Honein, **Cara T. Mai**, et al. National Prevalence Estimates for Selected Birth Defects in the U.S., 1999-2001. National Birth Defects Prevention Network 9th Annual Meeting, January 30-February 1, 2006, Arlington, VA.
11. Cynthia Cassell, **Cara Mai**, Russel Rickard. Interstate Data Exchange for Birth Defects Surveillance: A Battle Worth Fighting? National Birth Defects Prevention Network 9th Annual Meeting, January 30-February 1, 2006, Arlington, VA.
12. April Montgomery, Mittie Moffett, **Cara Mai**. Survey of Referral of Children to Services by Birth Defects Programs. National Birth Defects Prevention Network 8th Annual Meeting, January 23-26, 2005, Scottsdale, AZ.
13. Mark A. Canfield, Julianne Collins, Lorenzo D. Botto, Laura J. Williams, **Cara T. Mai**, Russell S. Kirby, Kay Pearson, and Joe Mulinare. Changes in the Birth Prevalence of Selected Birth Defects After Flour Fortification With Folic Acid in the United States: Findings From A Multi-State Population-Based Study. National Birth Defects Prevention Network 8th Annual Meeting, January 23-26, 2005, Scottsdale, AZ.