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

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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. State population-based surveillance systems have been established to monitor birth defects, yet no recent systematic examination of their efforts in the U.S. has been conducted.

Objective: To describe 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 reminders via e-mails and telephone were used to ensure a 100% response rate.

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 more often use clinical reviewers, cover broader pregnancy outcomes, and collect more extensive information, such as family history. Over half of the programs (24 out of 43) reported an ability to conduct follow-up studies of children with birth defects.

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] 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.[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 increase in the number 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: 1) community concerns about exogenous exposures, such as the use of teratogenic medications during pregnancy (e.g., Thalidomide) or exposure to environmental hazards (e.g., toxic waste); 2) evaluation of prevention strategies, such as folic acid fortification; and 3) referrals of affected children and families to medical and social services.[4]

In May 2010, the 65th World Health Assembly adopted Resolution WHA 63.17, highlighting the importance of surveillance, research, prevention, and intervention actions to address birth defects given their impact on infant and child morbidity and mortality.[5] 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 been used mainly 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 the Centers for Disease Control and Prevention (CDC).[6] The purpose of this study is to describe the current practices and approaches 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 (see selected survey questions used, Supplemental Digital Content Table 1). The survey questions were piloted by several state programs and then entered into SurveyMonkey™ (www.surveymonkey.com). The survey was e-mailed in January 2012 to birth defects surveillance contacts listed in the NBDPN annual report program directory for the 50 U.S. states, District of Columbia, and Puerto Rico with periodic e-mail reminders sent.[3] During the final data cleaning stage in fall 2013, the states that did not complete the survey or whose answers required clarification were contacted via phone to ensure completed responses from all programs. Survey responses were also crosschecked with available information from the NBDPN data report's annual directory and discrepancies were resolved by checking the information with program staff or existing programmatic materials.

The data were then imported to SAS 9.3 (SAS Institute Inc, Cary, NC) for cleaning and analysis. Descriptive analyses were performed by stratifying the 43 operational programs by their primary case ascertainment methodology (active or passive case-finding). Active case-finding methodology is when staff is sent to hospitals and provider offices to perform primary collection of medical information and birth defects data while passive case-finding approach relies on reported data from providers or administrative datasets where programs' staff may or may not perform definitive case confirmation of the information with active record review. 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 into categories.

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. Of the remaining nine states, three were planning to develop a program, and six indicated no birth defects surveillance activities. The 43 programs conducting surveillance cover a catchment area including approximately 80% of the live births in the U.S. Thirty-nine of the 43 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.

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 while 26 programs predominately use a passive case-finding approach.

Table 1 provides funding sources and methodology used by state programs. The top three funding sources include Federal Title V block grant, state general funds, 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). Programs all monitor major structural malformations; however, a greater number of the passive case-finding programs cover a broader list of conditions, including developmental conditions (23.1% compared to 11.8%), and newborn screening for hearing loss (38.5% compared to 5.9%) and metabolic and endocrine conditions (42.3% compared to 11.8%). All programs include live births but more of the 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 care hospitals. However, only two 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 physicians who specialize in birth defects) (35.3% compared to 3.8%) and geneticists (70.6% compared to 15.4%), to assess accuracy of the birth defects case status compared to programs that use passive case-finding. 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 beyond the basic demographic and clinical information collected by surveillance programs. Most programs have geocoded data and collect maternal residency at date of delivery. Very few programs collect maternal residency at date of conception or during the pregnancy time period and even fewer systematically collect information on prenatal diagnosis to identify potential cases of birth defects as the pregnancy

progresses. More active case-finding programs routinely collect and record information on family history compared to programs that use passive case-finding. 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 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 out of 43 programs (34.5%) can access or link to healthcare cost/charge data during the first year of the child's life while nine (20.9%) can follow-up beyond the first year of life. Furthermore, 12 programs (27.9%) are able to access or link to health care service data during the first year of life, and six programs (14%) can follow-up beyond the first year of life. However, when asked if they utilize cost or charges data, only six programs (14%) indicated its use for economic analysis, two (4.7%) do so for program planning, and three (7%) do so for needs assessment or 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. 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

The number of birth defects programs in the U.S. increased from three programs in the early 1970s to 43 programs by 2013. Lynberg and Edmonds published a comprehensive review of state birth defects surveillance in 1994, and reported that of the 23 operational programs, seven states used active and 16 used passive case-finding methodology.[6] The increase from 23 to 43 programs (42%) shows a modest shift in the number of programs using an active case-finding approach (30% to 40% of programs).

In this study, a dichotomous category was used to classify birth defects case-finding approaches. 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 primary case-finding approaches, since the data collected during this stage form the basis of the database for birth defect surveillance. Active case-finding approach is considered very complete, and each diagnosis in the database is confirmed.[7] But the approach is resource intensive. The other primary method that is used by 60% of state birth defects surveillance programs relies on a passive multiple source case-finding through hospital reports and / or administrative databases. This approach offers several benefits while considering resource constraints and potential improvement in timeliness that can be important for referring affected individuals to medical and social services. A concern for this approach is the accuracy of the information reported or obtained from administrative datasets. Salemi et al.[8] 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.[9] 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.[9] 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, regardless of their case-finding methodology, 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 Wang et al.[10] 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.

As the life expectancy for children born with birth defects increases,[11-14] 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 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. Additionally, recent public health priorities to better understand long-tem outcomes of children screened with disorders on the Recommended Uniform Screening Panel in the United States and to evaluate newborn screening of critical congenital heart defects (CCHDs) offer opportunities for birth defects programs. Hinton et al. (2014) articulated a knowledge gap in understanding population-based, long-term outcomes of children with confirmed metabolic conditions and presented a feasible approach for leveraging existing public health programs, such as birth defects surveillance systems, to address this gap.[15] Birth defects surveillance programs are also positioned to play a key role in the implementation and ongoing evaluation of CCHD newborn screening through screening accuracy evaluation, costs and service utilization analyses.[16] The flexibility in population-based birth defects programs can be adapted to address current and emergent needs.

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.

This study was also subject to several limitations. 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 occurred over a one and one-half year period 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 information gathered by these programs has been used to generate prevalence data, understand risk factors, examine mortality and morbidity impact, plan for services and referral of affected infants to medical and social services, as well as evaluate prevention strategies. The breadth and depth of information collected by birth defects surveillance programs 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.

References

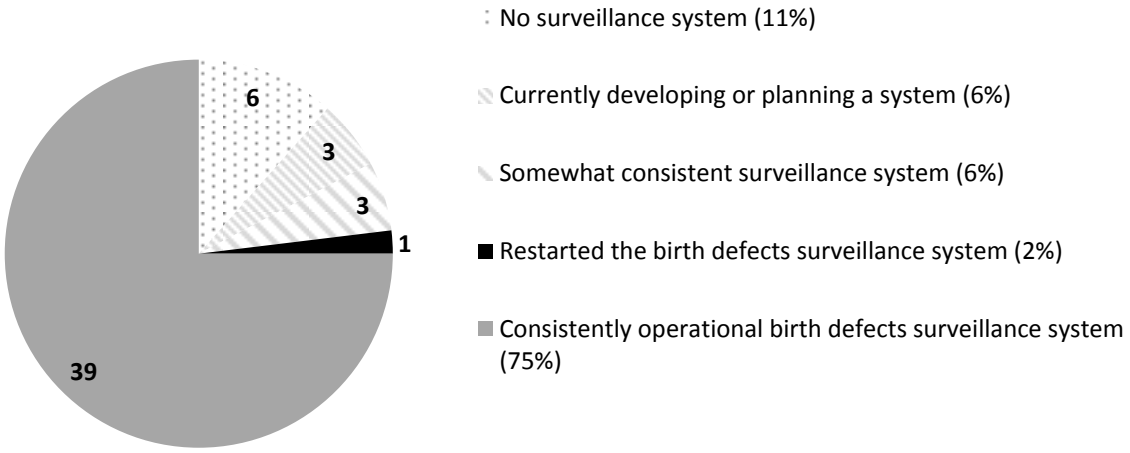
1. CDC. Ten Things You Need To Know About Birth Defects. http://www.cdc.gov/Features/BirthDefects. Updated January 24, 2011. Accessed January 24, 2014.
2. CDC. Improved National Prevalence Estimates for 18 Selected Major Birth Defects --- United States, 1999—2001. *MMWR Surveill Summ.* 2006;54(51&52):1301-1305.
3. National Birth Defects Prevention Network (NBDPN). State Birth Defects Surveillance Program Directory. *Birth Defects Res A Clin Mol Teratol.* 2012;94(12):S121-S169.
4. Edmonds LD. Birth defect surveillance at the state and local level. *Teratology.* 1997;56(1-2):5-7.
5. WHO. World Health Assembly Resolution 63.17: Birth Defects. http://apps.who.int/gb/ebwha/pdf_files/WHA63-REC1/WHA63_REC1-P2-en.pdf. Updated May 21, 2010. Accessesd January 24, 2014.
6. Lynberg MC, Edmonds LD. State use of birth defects surveillance. In: Wilcox LS, Marks JS (eds). From data to action: CDC's public health surveillance for women, infants, and children. Atlanta: US Department of Health and Human Services. 1994;217-229.
7. National Birth Defects Prevention Network (NBDPN). Guidelines for Conducting Birth Defects Surveillance. Sever, LE, ed. Atlanta, GA: National Birth Defects Prevention Network, Inc. 2004.

8. Salemi JL, Tanner JP, Kennedy S, Block S, Bailey M, Correia JA, Watkins SM, Kirby RS. A comparison of two surveillance strategies for selected birth defects in Florida. *Public Health Rep*. 2012;127(4):391-400.
9. Lin AE, Forrester MB, Cunniff C, Higgins CA, Anderka M. Clinician reviewers in birth defects surveillance programs: survey of the National Birth Defects Prevention Network. *Birth Defects Res A Clin Mol Teratol*. 2006;76(11):781-6.
10. Wang Y, O'Leary LA, Rickard RS, Mason CA; National Birth Defects Prevention Network. Geocoding capacity of birth defects surveillance programs: results from the National Birth Defects Prevention Network Geocoding Survey. *J Registry Manag*. 2010;37(1):22-6.
11. Kucik JE, Shin M, Siffel C, Marengo L, Correa A; Congenital Anomaly Multistate Prevalence and Survival Collaborative. Trends in survival among children with Down syndrome in 10 regions of the United States. *Pediatrics*. 2013;131(1):e27-36.
12. Gilboa SM, Salemi JL, Nembhard WN, Fixler DE, Correa A. Mortality resulting from congenital heart disease among children and adults in the United States, 1999 to 2006. *Circulation*. 2010;30:122(22):2254-63.
13. Wang Y, Liu G, Druschel CM, Kirby RS. Maternal race/ethnicity and survival experience of children with congenital heart disease. *J Pediatr*. 2013;163(5):1437-42.e1-2.
14. Shin M, Kucik JE, Siffel C, Lu C, Shaw GM, Canfield MA, Correa A. Improved survival among children with spina bifida in the United States. *J Pediatr*. 2012;161(6):1132-7.
15. Hinton CF, Mai CT, Nabukera SK, Botto LD, Feuchtbaum L, Romitti PA, Wang Y, Piper KN, Olney RS. Developing a public health-tracking system for follow-up of newborn

screening metabolic conditions: a four-state pilot project structure and initial findings. *Genet Med*. 2014;16(6):484-90.

16. Olney RS, Botto LD. Newborn screening for critical congenital heart disease: essential public health roles for birth defects monitoring programs. *Birth Defects Res A Clin Mol Teratol*. 2012;94(12):965-9.
17. Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, Anderson P, Mason CA, Collins JS, Kirby RS, Correa A; National Birth Defects Prevention Network. Updated National Birth Prevalence estimates for selected birth defects in the United States, 2004-2006. *Birth Defects Res A Clin Mol Teratol*. 2010;88(12):1008-16.
18. Canfield MA, Honein MA, Yuskiv N, Xing J, Mai CT, Collins JS, Devine O, Petrini J, Ramadhani TA, Hobbs CA, Kirby RS. National estimates and race/ethnic-specific variation of selected birth defects in the United States, 1999-2001. *Birth Defects Res A Clin Mol Teratol*. 2006;76(11):747-56.

Figure 1. Status of State Birth Defects Surveillance Programs, 2012 (n=52; 50 states, District of Columbia, and Puerto Rico)



State-wide coverage for 39 operational systems except for the following states:
California - Covers about 70,000 live births (LB) annually in 2 regions
Georgia - Covers about 35,000 LB annually in the metropolitan Atlanta counties
Minnesota - 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)	
	No.	%	No.	%
Funding[1]				
Title V MCH /SSDI	9	52.9	16	61.5
State General Funds	8	47.1	9	34.6
CDC birth defects surveillance	6	35.3	8	30.8
CDC environmental public health tracking (EPHT)	2	11.8	6	23.1
University/Academia	2	11.8	0	0.0
State fees, e.g. vital statistics, newborn screening, or dedicated fund	1	5.9	6	23.1
Other sources	1	5.9	3	11.5
Conditions ascertained[1]				
Structural malformations (all 46 birth defects on NBDPN list)	10	58.8	18	69.2
Developmental disabilities	2	11.8	6	23.1
Newborn/infant hearing	1	5.9	10	38.5
Newborn genetic and metabolic screening	2	11.8	11	42.3
Pregnancy outcomes covered[1]				
Live births	17	100.0	26	100.0

Program Structure	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	%	No.	%
Fetal deaths	15	88.2	15	57.7
Miscarriages (spontaneous abortions, <20 weeks gestation)	9	52.9	3	11.5
Pregnancy terminations - any gestation	13	76.5	4	15.4

No. – number of programs that selected "yes"; 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]Multiple responses allowed. Programs were asked to select all applicable responses.

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

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	**%**	**No.**	**%**
Disease classification coding system utilized[1]				
ICD-9-CM	8	47.1	24	92.3
CDC/BPA (6 digit code) or modified	15	88.2	6	23.1
ICD-10	2	11.8	6	23.1
Person responsible for assigning a disease classification code to a birth defects case [1]				
Data abstractor	11	64.7	6	23.1
Certified hospital coder (e.g. Registered Health Information Technicians - RHITs or Registered Health Information Administrator - RHIA)	1	5.9	9	34.6
Trained disease coder	4	23.5	7	26.9
Clinician or clinical reviewer	9	52.9	6	23.1
Epidemiologist	0	0.0	1	3.8
Background of data abstractor or other staff who review medical records for case identification or case verification[1]				
Health information management technology	10	58.8	2	7.7

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	%	No.	%
with RHIT/RHIA credential				
Nurse or Nurse Consultant	13	76.5	2	7.7
Other health care professional	8	47.1	1	3.8
None, trained in-house	4	23.5	8	30.8
Data quality procedures utilized to assess accuracy of the birth defects case status[1]				
Dysmorphologist clinical reviewer	6	35.3	1	3.8
Geneticist clinical reviewer	12	70.6	4	15.4
Cardiologist clinical reviewer	9	52.9	3	11.5
Pediatric clinical reviewer (on the personnel list)	4	23.5	3	11.5
Medical records or health records review	14	82.4	12	46.2
Quality of the data source (e.g. pathology, cytogenetic lab, genetics clinic, specialty clinic, etc)	11	64.7	11	42.3
Corroborating procedure that is linked to the diagnosis	11	64.7	8	30.8
Data quality assurance procedure performed by staff	14	82.4	17	65.4
Other, e.g. electronic edits, re-abstraction, etc.	3	17.6	6	23.1

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	%	No.	%
Classification of cases[1]				
Surveillance program with the ability to classify birth defect cases into isolated, multiple, and syndromes (not using disease codes)	7	41.2	3	11.5
Electronic transaction method used to receive a reported birth defect case or for case identification or case finding[1]				
Web based health information ports	5	29.4	13	50.0
Secure File Transfer	14	82.4	12	46.2
Internal health department electronic upload / transaction	2	11.8	6	23.1
External electronic download transaction or other type of download, e.g. encrypted e-mail, secure mail, CD	6	35.3	10	38.5
Abstraction Practices				
Data abstractors go to the delivery and tertiary hospitals to abstract the medical records of the fetus and/or infant [2]				
Yes; consistently for ALL birth defects or conditions	17	100	3	11.5

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	%	No.	%
reportable to the program				
Yes; consistently for selected birth defects or conditions	0	0.0	4	15.4
Yes; consistently for selected data sources	0	0.0	2	7.7
Yes, but only for selected conditions, selected data sources, or for special projects	0	0.0	3	11.5
Data abstractors go to the delivery hospital to abstract the medical records of the mother[2]				
Yes; consistently for ALL birth defects or conditions reportable to the program	14	82.4	2	7.7
Yes; consistently for selected birth defects or conditions	0	0.0	2	7.7
Yes, but only for selected conditions, selected data sources, or for special projects	2	11.8	1	3.8
Data abstractors request medical records from the mother's obstetric care provider to obtain additional information on the mother's pregnancy[2]				
Yes; consistently for ALL birth defects or	1	5.9	0	0.0

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	%	No.	%
conditions reportable to the program				
Yes; consistently for selected birth defects or conditions	1	5.9	0	0.0
Yes, but only for selected conditions, selected data sources, or for special projects	2	11.8	2	7.7

No. – number of programs that selected "yes"; % - 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

[1]Multiple responses allowed.

[2]Only one selection allowed.

Table 3: Selected Data Elements by Population-based Birth Defects Surveillance Programs by Case-finding Status

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	**%**	**No.**	**%**
Geocoding[1]				
Program routinely geocode or have access to geocoded data for birth defect cases	12	70.6	16	61.5
Program routinely geocode or have access to geocoded data for all live births (i.e. denominator data)	7	41.2	15	57.7
Maternal Residency: Time Period Collected[1]				
Maternal residency at date of delivery	17	100	22	84.6
Maternal residency at date of conception	4	23.5	1	3.8
Maternal residency collected during pregnancy period	3	17.6	1	3.8
Family History[1]				
Program routinely collects information on family history, (1st degree such as biological mother, father, siblings or greater) of birth defects in relation to index case	12	70.6	6	23.1
Program able to identify siblings within database by tracking through the biological mother	5	29.4	5	19.2

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	%	No.	%
Prenatal Surveillance[2]				
Yes, program identifies potential cases of birth defects that are prenatally diagnosed as the pregnancy is progressing	3	17.6	0	0.0
Yes, but only from selected data sources or for selected diagnosis	4	23.5	7	26.9
Follow-up studies				
Program has the capacity to conduct follow-up studies of children with birth defects[2]				
Yes, under 1 year of age	0	0.0	4	15.4
Yes, through 5 years of age	4	23.5	6	23.1
Yes, through 18 years of age	5	29.4	0	0.0
Yes, over age 18 years	3	17.6	2	7.7
Program has access or can link to cost/charge or health care service data during the first year of life[1]				
Program has access or can link to cost/charge data	7	41.2	8	30.8
Program has access or can link to health care utilization data	9	52.9	3	11.5
Program has access or can link to cost/charge or				

	Active Case-finding Programs (N=17)		Passive Case-finding Programs (N=26)	
	No.	%	No.	%
health care service data beyond first year of life[1]				
Program has access or can link to cost/charge data	7	41.2	2	7.7
Program has access or can link to health care utilization data	6	35.3	0	0
Programs' utilization of cost or charges data[1]				
Never utilized	9	52.9	16	61.5
Economic analysis (such as cost-benefit analysis analysis)	5	29.4	1	3.8
Program planning-justification	2	11.8	0	0.0
Needs Assessment	1	5.9	2	7.7
Legislative request	1	5.9	2	7.7

No. – number; [1]Multiple responses allowed. [2]Only one selection allowed.

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. National Birth Defects Prevention Study. 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. biological mother, biological 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?