

Neural Tube Defects Surveillance: A National Survey

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Neural tube defects (NTDs) are a class of birth defects characterized by defective closure of the neural tube during the first month of gestation. Reports from six states during the period of 1985-1994 demonstrated that the rate of NTDs in the United States varies by geographical area and by race/ethnicity. The reported rate ranged from 3.8 to 9.6 per 10,000 live births (CDC, '95). Recent epidemiological and basic science research has demonstrated that folic acid plays a significant role in the primary prevention of NTDs (Locksmith and Duff, '98).

In recent years the movement to establish population-based birth defects surveillance programs across the United States has gained momentum. By July 1997, programs had been established in 31 states, with organizing activities under way in several others (Edmonds, '97). These programs use a variety of case-finding methods, ascertain cases with differing sources; focus on varying segments of the perinatal and early childhood continuum; and handle coding, classification, medical record review and follow-up in a variety of ways.

We undertook a national survey of state health agencies and birth defects surveillance programs to determine how NTD surveillance data are currently being collected and applied to specific activities aimed at the prevention of NTDs. Results of this survey are presented in this report, together with a set of recommendations for the enhancement of future activities.

METHODS

We designed the survey after consulting with personnel at the Centers for Disease Control

and Prevention (CDC) and several colleagues from the birth defects surveillance community. We mailed the survey to the birth defects surveillance contact person in each state as listed in the directory maintained by the Division of Birth Defects and Pediatric Genetics at CDC. Respondents were asked to FAX their responses. Some follow-up was required to clarify certain answers; we received responses from all 50 states and the District of Columbia. Respondents were asked whether a program is conducting NTD surveillance in their state, for what time periods population-based NTD surveillance data are available, whether these data include prenatal diagnostic data, what coding/classification system is used, what specific conditions are categorized as NTDs, how useful various sources of NTD cases are in identifying NTDs, how timely case reports are in relation to the date of birth or prenatal diagnosis, whether medical records are reviewed to confirm case reports of NTDs and whether reported cases are reviewed by a clinical expert, and generally how the data are used in conjunction with intervention services.

RESULTS

Thirty-one respondents reported that there was a program that carried out NTD surveillance in their state. Four states reported that

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their states' NTD data were gathered exclusively from vital statistics sources and were excluded from the remaining analysis.

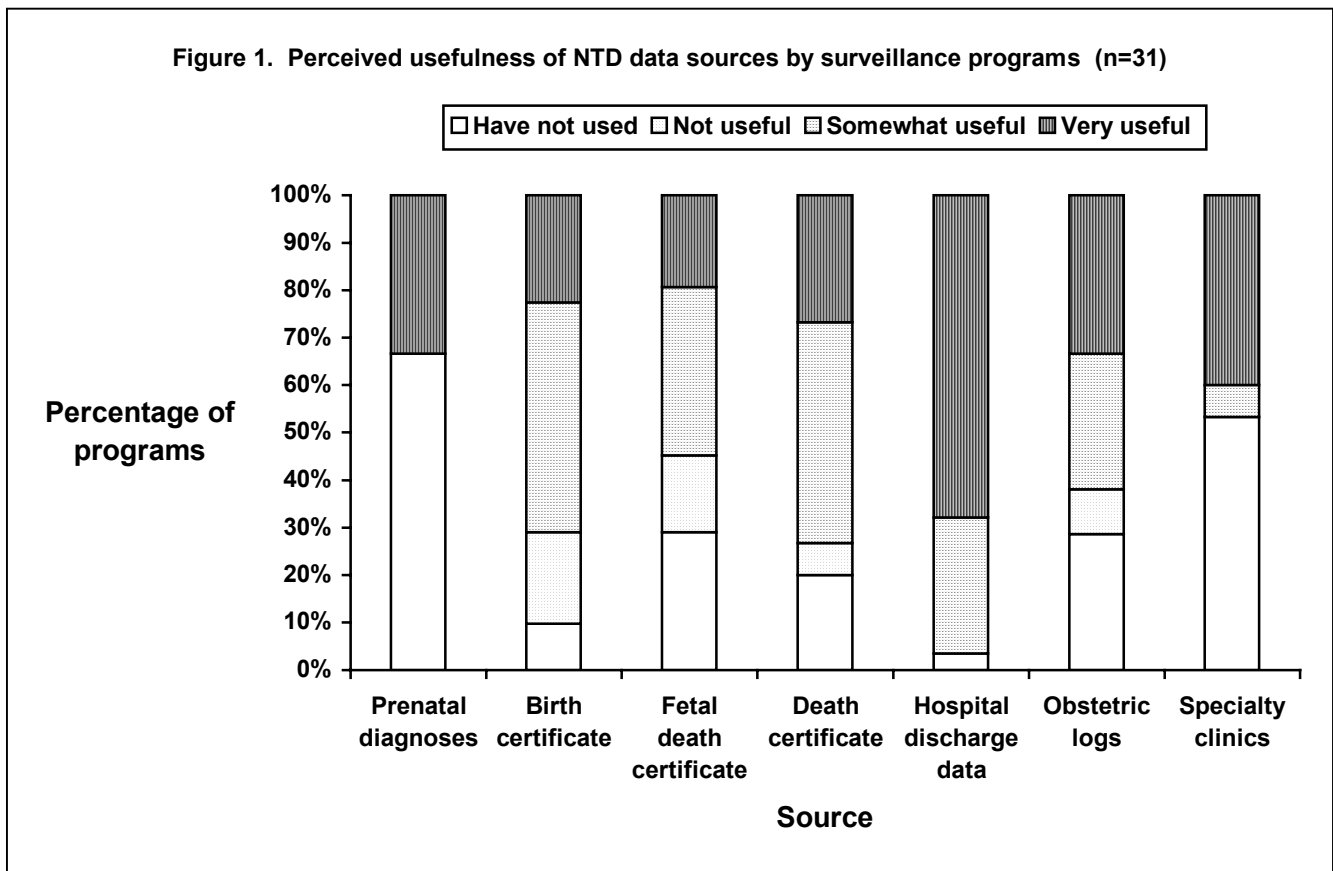
All 31 of the state NTD surveillance programs are population-based; 25 of the 31 have statewide coverage. Approximately two-thirds of the 31 programs initiated NTD surveillance after 1985. Only two programs have continuous data beginning prior to 1980, and eight have data that begin between 1980 and 1985 and continue to the present.

Thirteen respondents indicated that their states' NTD surveillance results include prenatal diagnostic data; seven reported that they began collecting these data for birth cohorts in 1994 or later. Of the 31 programs that carry out NTD surveillance, 12 use the CDC-British Pediatric Association six-digit classification system or a variant thereof, 17 use the International Classification of Diseases, Ninth Revision, Clinical Modification (ICD-9-CM), and

two use both classification systems.

Respondents were asked to select which of the 17 conditions that they categorize as NTDs. Twenty-two of the 31 programs categorized each condition as an NTD. Respondents were also given an opportunity to list additional conditions that they include as NTDs. Iniencephaly, added by eight programs, was the only condition mentioned by more than one respondent.

Respondents were asked how they use a variety of potential data sources in the ascertainment of NTDs and to rate each source on the following scale: have not used; used, not useful; somewhat useful; and very useful. For reporting these results, we assumed that lack of response indicates the program does not use that particular data source. Results are summarized in Figure 1. Most programs use a variety of data sources for NTD surveillance. Hospital discharge data were reported as being somewhat



or very useful for all 27 programs that incorporate this data source into their surveillance methodology. Respondents found vital statistics to be less useful. Only 7 indicated that birth certificates are very useful and 14 reported them to be somewhat useful. Sixteen programs reported that fetal death certificates were very or somewhat useful; 22 programs reported that death certificates were very or somewhat useful. While only 3 of 31 programs do not use birth certificates at all in NTD surveillance, 9 have not used fetal death certificate records, and 7 have not used death certificates. Several states reported using death certificates and fetal death certificates, but did not find them to be useful. All programs that access data on prenatal diagnosis found those data to be very useful, but only 10 of the 31 programs do so. Specialty clinic data are also highly valued by those programs using these sources, with 12 of 14 rating them very useful; however, more than half the programs do not currently include these sources in their surveillance strategy. Programs also access a variety of sources in addition to these, including surveillance case report forms, hospital disease indices, autopsy and pathology reports, Medicaid records, and maternal serum alpha-fetoprotein screening reports.

Respondents were asked to estimate the median number of days that elapse between an NTD event and initial case identification to the registry. Only 16 respondents were able to provide an estimate. Of these, 11 indicated that cases are identified a median of 90 days or less after the event, 3 indicated that the median was between 120 and 300 days, and 2 reported that the median lag in initial reporting to the surveillance program is more than 1 year. However, most programs also indicated a wide range, from a few days or a month after birth to as long as 5 or 6 years.

Respondents were asked whether their programs review medical records to confirm reported cases of NTDs. Twenty of the 31 re-

ported doing so. Of these, 15 reported reviewing all NTD cases, while the other 6 reported reviewing only selected diagnoses, reviewing cases at tertiary facilities, or randomly selecting cases within a larger quality assurance process. In 9 of the 20 programs, a clinical expert (dysmorphologist, pathologist, geneticist) confirms the diagnosis, and in most instances the review is conducted on the basis of case report forms rather than medical records.

Finally, respondents were asked how the NTD surveillance data are linked to several types of intervention services. These included use of the registry to identify women for folic acid counseling, to evaluate the impact of public health education campaigns, and to make referrals for specialized services, including Children with Special Health Care Needs (CSHCN) and related activities. Fifteen respondents indicated that their programs use the NTD surveillance data in some manner for intervention activities. Most of these activities focus on referrals to local and statewide programs for early intervention and for CSHCN and similar services. Three indicated that their programs use data to evaluate folic acid education campaigns. Six reported providing educational materials about folic acid or NTD recurrence prevention to women identified from their surveillance systems.

DISCUSSION

This survey demonstrated that while most states perform surveillance for NTDs, they do so in a wide variety of ways, using different classification systems and varying sources of data. In addition, half of states do not link the data to prevention or intervention activities.

One way that these surveillance data can be used is to evaluate the effectiveness of NTD prevention programs. Because not all states have surveillance systems and because of the variability between systems, surveillance data cannot presently be used to evaluate NTD programs nationally. However, surveillance sys-

tems can be used to evaluate the effect of folic acid intervention programs on NTD rates in individual states and in groups of states (Lary and Edmonds, '96). To do this accurately, surveillance systems need to have a comprehensive, ongoing system of data collection. States with a limited number of data sources may want to consider evaluating additional sources of data that others have found to be useful, as shown in Figure 1, in order to have more comprehensive data.

To evaluate the effectiveness of NTD intervention programs, analysts need NTD outcome data collected during the entire perinatal continuum. NTDs have their genesis in the failed or incomplete closure of the neural tube at approximately 28 days of gestation. NTDs can be detected antenatally through maternal serum alpha-fetoprotein screening or prenatal ultrasound tests; patients with affected pregnancies have the option to continue or electively terminate the pregnancy. Multistate, population-based surveillance has demonstrated that prenatally diagnosed cases of NTDs must be included in any comprehensive NTD data collection effort (CDC, '95). Failure to include these cases may result in an apparent reduction in NTD rates that is attributable to increases in rates of prenatal diagnosis and elective termination rather than increased folic acid use. Only a minority of surveillance programs currently use data concerning prenatal NTD diagnoses; all NTD surveillance programs should work to develop mechanisms for accessing information concerning these cases.

Evidence of the effectiveness of folic acid in preventing a portion of NTDs began accumulating in the 1980s – prior to the initiation of birth defect surveillance systems in many states. Public Health Service recommendations regarding folic acid use were published in 1991 and 1992 (CDC, '91; CDC, '92). Despite these recommendations, in 1997, only 30% of women reported consuming adequate amounts of supplemental folic acid (CDC, '97). However, there are widespread

However, there are widespread efforts to continue to increase the number of women who consume adequate amounts of folic acid. For example, in January 1998, the Food and Drug Administration implemented the fortification of grain products in the United States; and the National Folic Acid Task Force is currently implementing a national education campaign to encourage women of reproductive age to take folic acid (personal communication, Patricia Mersereau, CDC). To monitor the effect of these ongoing prevention activities states need to use the most complete data available.

In addition to using surveillance data to evaluate folic acid interventions, our survey respondents described several other ways that they can use these data. The following examples may be useful for states that are developing prevention or intervention programs. Some states mail information about NTD recurrence prevention to women who have had an NTD-affected pregnancy. Other states use case managers in local agencies to contact the family of children with NTDs to ensure that they have access to services, education, and counseling about NTD recurrence prevention. States also refer women and families to sites where they can receive counseling about NTDs and NTD recurrence prevention. Several surveillance programs also make referrals for CSHCN services and other early-intervention programs.

To be most effective, each of these interventions require timely data. We recommend that surveillance programs evaluate the timeliness of their data from each data source. Programs should then focus their efforts on reducing any excessive lag between the time of NTD diagnosis and the identification of NTD cases by the surveillance program.

The surveillance programs we surveyed were in general, but not complete, agreement regarding the conditions that they considered to be NTDs. This inconsistency may be addressed by a case definition for individual birth defects, including NTDs, as has been done for infectious

conditions under public health surveillance (CDC, '97). Case definitions for noninfectious conditions are currently being developed by the Centers for Disease Control and Prevention (personal communication, Larry Edmonds, CDC).

We excluded surveillance programs that rely exclusively on vital statistics data for three reasons. First, the validity of birth defect diagnoses identified solely from birth certificates is poor (Watkins, '96; Hexter, '90). Second, the sensitivity of NTD data in vital records has also been questioned (Greb, '87; Jorde, 84). Finally, we were concerned that families of children whose NTD status was misdiagnosed on the basis of birth certificate data might be contacted for follow-up and intervention activities. Vital statistics data do provide denominator data and important demographic characteristics. In addition, these data (obtained from birth, death, and fetal death certificates) were found to be somewhat or very useful by most of the 31 programs that perform NTD surveillance. However, although vital statistics data may have a "vital" role if used in combination with data from other sources, these data, by themselves, are inadequate for NTD surveillance.

In summary, 25 U.S. states use a statewide surveillance system to monitor the rate of NTDs. Many of these states, however, do not yet include prenatally diagnosed cases, take full advantage of available data sources, evaluate the timeliness of reports, or use the data in prevention or intervention programs. We suggest that these be areas of focus as states continue to develop and improve their systems.

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