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Utility of Population-based Birth Defects Surveillance for Monitoring the Health of Infants and as a Foundation for Etiologic Research

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The National Birth Defects Prevention Network (NBDPN) is a nonprofit volunteer organization in the United States for individuals working on birth defects surveillance, research, and prevention (www.nbdpn.org) including, among others, public health officials, epidemiologists, clinicians, and parent advocates. NBDPN works to improve the quality of birth defects surveillance data, and to make these data more accessible both in publications and online. NBDPN also works to increase collaboration among surveillance programs and advance our understanding of the causes and risk factors for birth defects. In this year's annual report from the NBDPN, 40 programs reported updated birth defects surveillance data on at least some of the 47 defects monitored by this organization.

Multiple Uses of Population-based Birth Defects Surveillance Data

Population-based birth defects surveillance is a systematic collection of data on major birth defects among fetuses and infants. Major birth defects are characterized as having medical, surgical, or serious cosmetic importance and substantially contribute to infant mortality and, to a lesser extent, childhood morbidity and disability in the United States (Boyle et al., 2005; Correa et al., 2007). Thus, a primary and critical focus of surveillance has been tracking the occurrence of birth defects to monitor unexpected changes in prevalence that might indicate an exposure of concern or a change in the population that is being monitored. Surveillance is the cornerstone of the public health model and its utility is not limited to a single point in time or public health action (Lee et al., 2010). In addition to the more immediate use of surveillance data for detection of disease or other changes affecting population health, data collected over a period of time are essential for estimating the public health burden of a condition as well as evaluating response to interventions. Surveillance data that are maintained can serve as the data source for describing the natural history of a disease or condition and as a foundation for public health research (Nsubuga et al., 2006; Thacker et al., 2012).

Because specific birth defects are relatively rare, case–control studies are the primary approach used to identify risk factors for major birth defects (Rasmussen and Shaw, 2010). Population-based birth defects surveillance serves as the foundation for these research efforts by providing an unbiased case source. The Centers for Disease Control and

Prevention (CDC) coordinated the National Birth Defects Prevention Study for births from 1997 to 2011, and is currently coordinating the Birth Defects Study to Evaluate Pregnancy Exposures for births occurring from 2014 forward (Reefhuis et al., 2015; Tinker et al., 2015). This work has identified some important, potential risk factors that might contribute to the occurrence of major birth defects including poor diet quality, maternal use of opioid medications, maternal use of selective serotonin reuptake inhibitors, and maternal obesity (Waller et al., 2007; Broussard et al., 2011; Carmichael et al., 2012). Although these research studies sometimes garner attention, particularly when the findings have clinical implications, the foundation of birth defects surveillance contributing to this work is typically not highlighted. However, these birth defect studies have major advantages over clinic-based studies and self-enrolled registries. By including all women residing in a defined region who have the opportunity to participate, regardless of socioeconomic level, healthcare access, educational level, and race/ethnicity, they minimize one source of selection bias (Kukull and Ganguli, 2012). Ultimately findings that can be generalized appropriately to the population are useful to inform public health action.

Using Birth Defects Surveillance Data to Assess Key Longer Term Outcomes and Barriers to Care

Data from birth defects surveillance programs are also increasingly used to assess key longer term outcomes for those born with major birth defects. Collaboration among state-based birth defects surveillance programs ensure sufficient numbers of individual birth defects to produce reliable estimates. Recently 12 state programs contributed data to an assessment of racial/ethnic differences in survival for children with at least one of 21 different types of major birth defects (Wang et al., 2015). For 13 of the 21 types of birth defects assessed, postneonatal mortality was greater for infants born to non-Hispanic black mothers than for those born to non-Hispanic white mothers.

Surveillance data have also been used to study trends in morbidity and mortality. Using data from birth defects surveillance programs in 10 regions in the United States, researchers found improved survival of individuals with Down syndrome over a 20-year period of time from 1983 to 2003 (Kucik et al., 2013). They were able to describe a diminution of racial/ethnic disparities during infancy; however, during childhood and adolescence a greater risk for mortality was noted for non-Hispanic blacks. Identification of factors such as healthcare access that might be contributing to this disparity would necessitate linkage to additional data sources such as hospital discharge data.

Birth defects surveillance has also served as the foundation for surveys to better understand barriers to care. Using data on orofacial clefts from the North Carolina Birth Defects Monitoring Program, an existing population-based system, researchers assessed distance to care as a potential barrier (Cassell et al., 2013). They found that almost half of the respondents traveled more than an hour to receive care. Further work identified additional barriers, including having to take time off from work, long waits for appointments, and cost (Cassell et al., 2014). Work is in progress using data from the Florida Birth Defects Registry to similarly assess distance to care as a potential barrier.

Linking Birth Defects Surveillance Data to Other Data Sources

The linkage of birth defects surveillance data to other existing data sources such as hospital discharge data, billing data, and outpatient data has the opportunity to extend our knowledge of the specific use and costs of this healthcare for children with birth defects and their families. Using linked data for case infants with craniofacial malformations, identified by the Massachusetts Birth Defects Monitoring Program both mean and median number of inpatient days for these children in the first 2 years of life were assessed and found to be three times higher than for children born without these conditions (Weiss et al., 2009). Another example is a recent analysis in Florida, again from population-based surveillance, looking at healthcare use for children with orofacial clefts, and identifying factors associated with high healthcare use and costs. Among other factors, researchers found that one type of defect (i.e., cleft palate) and the presence of multiple birth defects were associated with greater inpatient days and costs (Razzaghi et al., 2015).

The March of Dimes, in collaboration with CDC National Center on Birth Defects and Developmental Disabilities, has recently funded two cooperative agreements to extend the utility of birth defects surveillance programs through additional linkage with existing data sources (March of Dimes, 2015). One cooperative agreement with the Arkansas Children's Hospital Research Institute plans to use data from the Arkansas Reproductive Health Monitoring System, a population-based birth defects surveillance system, to explore maternal characteristics, financial burden, heath care usage, survival, and long-term outcomes of individuals born with birth defects. Approximately 14,000 cases born between 2000 and 2011, as well as approximately 28,000 controls, will be identified and linked to birth and death certificates, the Arkansas Hospital Discharge Database, Arkansas Medicaid claims for mother and child, Arkansas standardized education data, and clinical data of children receiving care at Arkansas Children's Hospital. A second cooperative agreement with the University of South Florida will build on the previous data linkage work in Florida and add information such as linking with the childhood cancer registry to further evaluate the link between major birth defects and childhood cancer (Fisher et al., 2012; Botto et al., 2013). Data from the Florida Birth Defects Registry will be linked with birth and death certificates, hospital discharge data, and the Florida Cancer Data System. Approximately 3 million individuals born with a birth defect between 1998 and 2011 are expected to be identified/linked to these datasets.

Future Directions for Birth Defects Surveillance

Despite the many uses of population-based birth defects surveillance data for monitoring the health of infants and as the foundation for etiologic research, budgetary constraints preclude the development of these programs in some states. In states with programs, lack of sufficient resources can affect the geographical areas that can be monitored, number and types of data sources, timeliness of reporting, as well as the ability to creatively use these data through linkage and other methods to better understand the impact of birth defects on affected individuals and their families and communities.

Emerging technologies are impacting the practice of public health surveillance. Some, such as electronic health records and the establishment of health information exchanges, are increasingly being used by traditional public health systems (Savel et al., 2012). However, for other surveillance activities such as detection of diseases and evaluation of response to intervention, emerging data sources could potentially include the Internet and social media (Eysenbach, 2011; Velasco et al., 2014). Examples of application of these sources are most numerous in the realm of communicable disease, most prominently influenza, (Bernardo et al., 2013; Wiwanitkit, 2014) and real-time estimates of levels of influenza-like illness in a population. But due to several challenges such as data quality, these types of emerging resources have not been integrated into public health surveillance and their widespread use awaits evidence of added value when compared with traditional surveillance (Bernardo et al., 2013). Recently, researchers have shown that Internet search activity can be used in the noncommunicable disease arena to accurately predict population disease risk when compared with population-based data collected by the CDC's Behavioral Risk Factor Surveillance (Nguyen et al., 2015). The authors note the potential utility of Internet search activity to provide real-time information on population risk during population-level interventions. They also highlight the public availability of these data and their ability to generate data before traditional methods of data gathering such as population surveys. These findings pave the way for the application of this technology to birth defects surveillance.

CONCLUSIONS

Data from population-based birth defects surveillance programs will continue to help us better understand the consequences and challenges of birth defects for affected individuals and their families and communities. These programs have also been instrumental in supporting etiologic research to find modifiable risk factors for birth defects. Surveillance programs have been resourceful in the use of data, stretching the data utility through collaborations and linkages. Newer methods of surveillance are emerging and might complement traditional methodology, as programs with increasingly fewer resources strive to provide data to support a number of public health functions as well as needed information on existing and emerging issues related to birth defects for clinicians and families.

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