Birth Defects

Birth defects or congenital anomalies are structural or functional anomalies causing physical or mental disability, some of which can be fatal. Although birth defects are the leading cause of infant mortality (deaths occurring to those under 1 year of age) in the U.S., the cause is unknown for approximately 70 percent of all cases (Infant Mortality indicator) (CDC, 2011). Many different factors may be associated with the development of birth defects, such as genetic and/or chromosomal aberrations, *in utero* exposure to viruses or bacteria, uncontrolled maternal diabetes, maternal cigarette smoke, maternal use of drugs and alcohol during pregnancy, and prenatal exposure to chemicals. All of these factors may influence normal infant growth or development, resulting in different types of birth defects (NICHD, 2012).

This indicator presents birth defects prevalence at birth for five specified congenital anomalies (anencephaly, cleft lip or palate, Down syndrome, omphalocele or gastroschisis, and spina bifida or meningomyelocele) that are consistently reported on both the 2003 (revised) and 1989 U.S. Standard Certificates of Live Birth and mortality rates among infants in the U.S. as recorded in the National Vital Statistics System (NVSS), which registers virtually all births and deaths nationwide. Birth defects data are currently collected on birth certificates and death certificates from all 50 states and the District of Columbia. Reported race and ethnicity data are based on the race and ethnicity of the mother.

What the Data Show

Exhibit 1 presents the prevalence of live births with identified specific congenital anomalies between 1999 and 2010. While the rates of birth defects are rare and underreported, as stated in the Limitations, it is possible to make some general inferences based on the available data. For example, as Exhibit 1 shows, the rates for cleft lip or palate and spina bifida or meningomyelocele decreased slightly over the last decade. Rates for the other three fluctuated over time with a slight increase in rates since 1999.

Rates for certain types of anomalies differ widely with maternal age. For example, in 2010 as in past years, infants of the youngest mothers (under 20 years of age) have the highest rates for omphalocele orgastroschisis, a defect or abnormality of the anterior abdominal wall (102.1 per 100,000 live births); infants of mothers age 40-54 years have the highest rates for Down syndrome (333.9 per 100,000 live births).

Birth defects continue to be the leading cause of infant mortality, accounting for 5,319 (20.1 percent) of the 26,412 infant deaths in 2009 (Exhibit 2, Infant Mortality indicator). Between 1979 and 1998, a decline in the national birth defects mortality rate has been observed, ranging from 255.4 per 100,000 live births in 1979 to 157.6 per 100,000 live births in 1998. From 1999 to 2009, the birth defects mortality rates ranged from a high of 150.9 in 2000 to a low of 124.8 per 100,000 live births in 2009. (Data not shown.)

Birth defect mortality rates differ by sex, race, and ethnicity. From 1999 to 2009, females had lower rates for each individual year than males, but the range in rates was similar between the two sexes. Specifically, birth defect mortality rates for females ranged from a high of 146.2 (2000) to a low of 122.6 (2009) per 100,000 live births compared to a high of 155.4 (2000) and a low of 127.0 (2009) per live births for males. Asian or Pacific Islanders consistently had the lowest rates of mortality

from birth defects among the reported racial groups from 1999 to 2009. During the 11-year period from 1999-2009, Blacks or African Americans had the second highest birth defect mortality rates for seven years, while American Indians/Alaska Natives had the second highest rates for four years. Whites did not have the highest or lowest birth defects mortality rate for any of these years. For each year during 1999 to 2009, Hispanics or Latinos had higher birth defects mortality rates than non-Hispanics except for two years: 1999 and 2009. (Data not shown.)

Limitations

- In order to enable comparisons over time, this indicator represents only a subset of possible birth defects. This is necessary because of changes made to the U.S. Standard Certificate of Live Birth in 2003. Consistent with NCHS reporting in its recent "Births: Final Data" publications, only the five congenital anomalies reported on both the 2003 and 1989 U.S. Standard Certificates of Live Birth are included (i.e., anencephaly, cleft lip or palate, Down syndrome, omphalocele or gastroschisis, and spina bifida or meningomyelocele).
- Birth defects are often underreported on both birth and death certificates (Boulet et al., 2011; Friis and Sellers, 1999). Many anomalies are hard to detect at birth, which limits early ascertainment and complete reporting. While the most serious and/or apparent anomalies are more likely to be identified and reported prior to hospital discharge, studies have reported low overall sensitivity (e.g., 23-28%) of selected birth defects reported on birth certificates (Boulet et al., 2011; Honein et al., 2001). Research shows that the NVSS birth records can produce prevalence estimates that are 2 to 3 times lower than those based on ascertainment of congenital defects using records from active surveillance efforts (Parker et al., 2010).
- The congenital anomalies reported on birth certificates are rare events. Since a small change in the number of anomalies reported can result in a relatively large change in rates, caution should also be used in comparing yearly rates for a specific anomaly.
- Birth defects mortality rates are based on underlying cause of death as entered on a death certificate by a physician. Incorrect coding and low rates of autopsies that confirm the cause of death may occur. Additionally, some individuals may have had competing causes of death. When more than one cause or condition is entered by the physician, the underlying cause is determined by the sequence of conditions on the certificate, provisions of the ICD [International Classification of Diseases], and associated selection rules and modifications. Consequently, some misclassification of reported mortality might occur in individuals with competing causes of death, as well as underreporting of some birth defects as the cause of death.
- The International Classification of Diseases 9th Revision (ICD-9) codes were used to specify underlying cause of death for years 1979-1998. Beginning in 1999, cause of death is specified with the International Classification of Diseases 10th Revision (ICD-10) codes. The two revisions differ substantially, and to prevent confusion about the significance of any specific disease code, data queries are separate. The relatively large difference between birth defects mortality rates reported from 1979 through 1998 and those reported beginning in 1999 may be due to some changes in the criteria used to report birth defects mortality during the switch from ICD-9 to ICD-10.

Data Sources

The birth defects rate data used for this indicator are from National Vital Statistics Reports published

by the Centers for Disease Control and Prevention's National Center for Health Statistics (NCHS, 2001, 2002a,b, 2003, 2005, 2006, 2007, 2009, 2010a,b, 2012a,b). The birth defects mortality data were obtained from CDC's compressed mortality files (underlying cause of death), accessed via CDC WONDER (CDC, 2012), athttp://wonder.cdc.gov.

References

Boulet, S.L., M. Shin, R.S. Kirby, D. Goodman, and A. Correa. 2011. Sensitivity of birth certificate reports of birth defects in Atlanta, 1995-2005: Effects of maternal, infant, and hospital characteristics. Public Health Reports 126(2):186-94. http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3056031/?tool=pubmed.

CDC. 2012. CDC Wide-ranging OnLine Data for Epidemiologic Research (WONDER). Compressed mortality file, underlying cause of death. 1999-2009 (with ICD 10 codes) and 1979-1998 (with ICD 9 codes). Accessed December 3, 2012. http://wonder.cdc.gov/mortSQL.html.

CDC. 2011. Birth defects. Last updated February 24, 2011. Accessed December 3, 2012. http://www.cdc.gov/ncbddd/birthdefects/facts.html.

CDC. n.d. CDC WONDER: Help page for compressed mortality file. Accessed December 3, 2012. http://wonder.cdc.gov/wonder/help/cmf.html.

Friis, R.H., and T.A. Sellers. 1999. Epidemiology for public health practice. Second edition. Gaithersburg, MD: Aspen Publishers.

Honein, M.A., L.J. Paulozzi, and M.L. Watkins. 2001. Maternal smoking and birth defects: Validity of birth data for effect estimation. Public Health Reports 116(4):327-335.

NCHS (National Center for Health Statistics). 2012a. Births: Final data for 2010. National Vital Statistics Reports 61(1). See Table I-6. http://www.cdc.gov/nchs/data/nvsr/nvsr61/nvsr61_01.pdf (PDF) (72 pp, 1.7MB).

NCHS. 2012b. Births: Final data for 2009. National Vital Statistics Reports 60(1). See Table I-6. http://www.cdc.gov/nchs/data/nvsr/nvsr60/nvsr60_01_tables.pdf (PDF) (15 pp, 323K).

NCHS. 2010a. Births: Final data for 2008. National Vital Statistics Reports 59(1). See Table I-6. http://www.cdc.gov/nchs/data/nvsr/nvsr59/nvsr59_01_tables.pdf#tableI06 (PDF) (17 pp, 323K).

NCHS. 2010b. Births: Final data for 2007. National Vital Statistics Reports 58(24). See Table 25. http://www.cdc.gov/nchs/data/nvsr/nvsr58/nvsr58_24.pdf (PDF) (86 pp, 1.8MB).

NCHS. 2009. Births: Final data for 2006. National Vital Statistics Reports 57(7). See Table 25. http://www.cdc.gov/nchs/data/nvsr/nvsr57/nvsr57_07.pdf (PDF) (102 pp, 1.4MB).

NCHS. 2007. Births: Final data for 2005. National Vital Statistics Reports 56(6). See Table 25. http://www.cdc.gov/nchs/data/nvsr/nvsr56/nvsr56_06.pdf (PDF) (104 pp, 2.5MB).

NCHS. 2006. Births: Final data for 2004. National Vital Statistics Reports 55(1). See Table 25. http://www.cdc.gov/nchs/data/nvsr/nvsr55/nvsr55_01.pdf (PDF) (102 pp, 3.3MB).

NCHS. 2005. Births: Final data for 2003. National Vital Statistics Reports 54(2). See Table 49. http://www.cdc.gov/nchs/data/nvsr/nvsr54/nvsr54_02.pdf (PDF) (116 pp, 3.9MB).

NCHS. 2003. Births: Final data for 2002. National Vital Statistics Reports 52(10). See Table 49. http://www.cdc.gov/nchs/data/nvsr/nvsr52/nvsr52_10.pdf (PDF) (114 pp, 1.9MB).

NCHS. 2002a. Births: Final data for 2001. National Vital Statistics Reports 51(2). See Table 49. http://www.cdc.gov/nchs/data/nvsr/nvsr51/nvsr51_02.pdf (PDF) (103 pp, 6MB).

NCHS. 2002b. Births: Final data for 2000. National Vital Statistics Reports 50(5). See Table 49. http://www.cdc.gov/nchs/data/nvsr/nvsr50/nvsr50_05.pdf (PDF) (102 pp, 1.5MB).

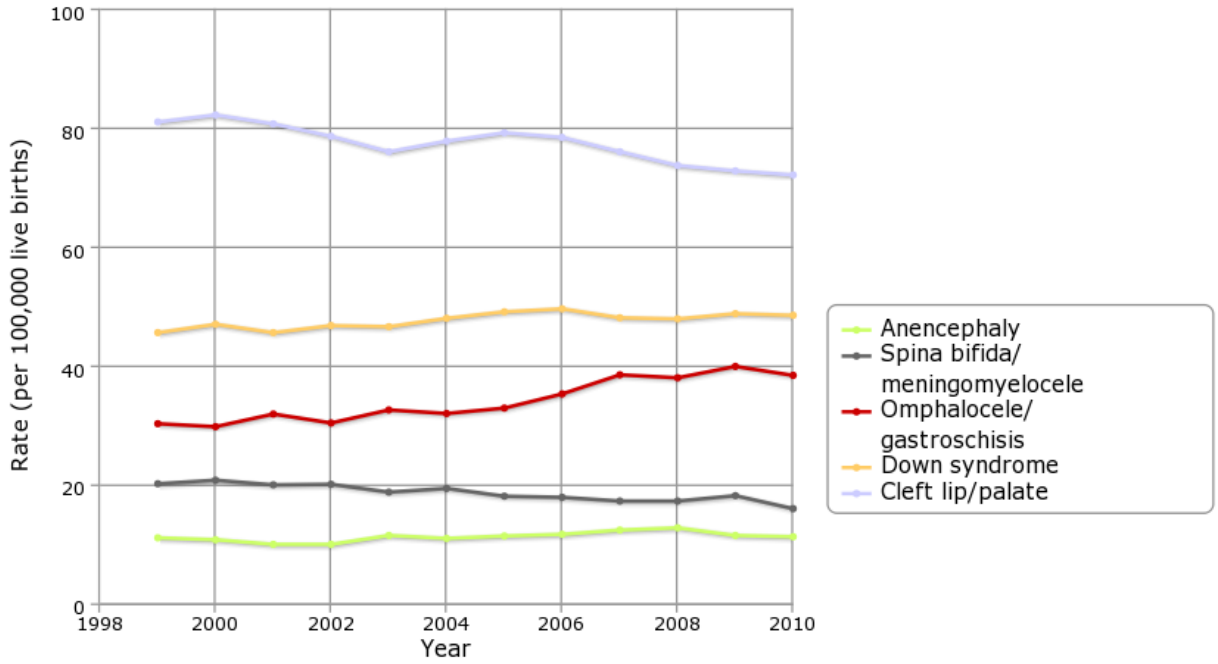
NCHS. 2001. Births: Final data for 1999. National Vital Statistics Reports 49(1). See Table 49. http://www.cdc.gov/nchs/data/nvsr/nvsr49/nvsr49_01.pdf (PDF) (100 pp, 6.2MB).

NICHD (National Institute of Child Health and Human Development). 2012. Birth defects overview. Last updated November 30, 2012. Accessed December 3, 2012. http://www.nichd.nih.gov/health/topics/birthdefects/Pages/default.aspx.

Parker, S.E., C.T. Mai, M.A. Canfield, R. Rickard, Y. Wang, R.E. Meyer, et al. 2010. Updated national birth prevalence estimates for selected birth defects in the United States, 2004-2006. Birth Defects Research (Part A) Clin Mol Teratol. 88(12):1008-1016.

Exhibit 1. Rate of live births in the U.S. with specific birth defects (congenital anomalies), 1999-2010

All cases by birth defect



Information on the statistical significance of the trend in this exhibit is not currently available. For more information about uncertainty, variability, and statistical analysis, view the technical documentation for this indicator.

Data source: NCHS, 2001, 2002a,b, 2003, 2005, 2006, 2007, 2009, 2010a,b, 2012a,b

Visit http://www.epa.gov/roe to see the full exhibit.