

Report on the Environment

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Birth Defects

Birth defects or congenital anomalies are structural or functional anomalies causing physical or mental disability, some of which can be fatal. Birth defects are the leading cause of infant mortality (deaths occurring to those under 1 year of age) in the U.S. ([Infant Mortality](#) indicator). The most recent available data indicate birth defects accounted for 4,047 (20.7 percent) of the 19,578 infant deaths reported in the U.S. in 2020 (CDC, 2022). For approximately 70 percent of all cases, the cause of birth defects is unknown (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 fetal growth or development, resulting in different types of birth defects (CDC, 2021; NICHD, 2017).

Birth defects rates among infants in the U.S. are recorded in the National Vital Statistics System (NVSS), which registers virtually all births nationwide. Birth defects data are currently collected on “standard” birth certificates from all 50 states and the District of Columbia. Standard birth certificates are revised periodically, with the most recent revisions occurring in 2003.

This indicator presents birth defects prevalence at birth from 1999 to 2020 for the five congenital anomalies consistently reported on both the 2003 (revised) and 1989 U.S. Standard Certificates of Live Birth: anencephaly, cleft lip or palate, Down syndrome, omphalocele or gastroschisis (a defect or abnormality of the anterior abdominal wall), and spina bifida or meningomyelocele. Rates for these congenital anomalies include data for all 50 U.S. states and the District of Columbia. This indicator also presents prevalence from 2010 to 2020 for the additional five congenital anomalies reported on the 2003 U.S. Standard Certificate of Live Birth: cyanotic congenital heart disease, congenital diaphragmatic hernia, hypospadias (males only), limb reduction defect, and suspected chromosomal disorder. Rates for these congenital anomalies are shown separately because they only account for the states reporting on the 2003 revised form from 2010 to 2020 (ranging from 33 states in 2010 to 50 states and the District of Columbia in 2016-2020).

What the Data Show

Exhibit 1 presents the prevalence of live births with identified specific congenital anomalies between 1999 and 2020. Rates for these five anomalies fluctuated over time since 1999. 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, particularly patterns across maternal age groups.

For instance, rates for certain types of anomalies differ widely with maternal age. For example, in 2020 as in past years, infants of the youngest mothers (under 20 years of age) have the highest rates for omphalocele or gastroschisis (101.5 per 100,000 live births); infants of mothers aged 40-54 years have the highest rates for Down syndrome (338.8 per 100,000 live births).

Exhibit 2 presents prevalence data for 2010 to 2020 for the five additional congenital anomalies presented on the 2003 birth certificate. Rates for these five anomalies fluctuated over this time period. Because the annual rates in this exhibit do not consistently represent all 50 U.S. states and the District of Columbia, trends through time should be interpreted with caution.

Rates for two types of anomalies in Exhibit 2 differ by maternal age. For all years from 2010 to 2020, mothers aged 40-54 years have the highest rates for both cyanotic congenital heart disease and suspected chromosomal disorder.

Of the 10 anomalies presented in Exhibits 1 and 2, four have been consistently reported at the highest rates across the U.S. population among live births. In 2020, the most current reporting year, rates for these four anomalies were as follows: hypospadias in male births (108.7 per 100,000), cleft lip/palate (72.9 per 100,000), cyanotic congenital heart disease (63.4 per 100,000), and Down syndrome (54.1 per 100,000).

Limitations

- Birth defects are often underreported on birth certificates (Boulet et al., 2011; Friis and Sellers, 2014). 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; Salemi et al., 2017). Research shows that the NVSS birth records can produce prevalence estimates that are lower than those based on ascertainment of congenital defects using records from population-based surveillance efforts (Canfield et al., 2014; Mai et al., 2015; 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.
- The annual rates shown in Exhibit 2 are based on the states reporting on the revised 2003 U.S. Certificate of Live Birth each year from 2010 (33 states and the District of Columbia) to 2016-2020 (50 states and the District of Columbia). This may introduce some uncertainty regarding the national representativeness of these data and trends of these five congenital anomalies through time.

Data Sources

The birth defects rate data used in this indicator for Exhibit 1 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, 2011, 2012a, 2013, 2014a, 2015a,b, 2017, 2018a,b, 2019, 2021, 2022), which are available online at <https://www.cdc.gov/nchs/products/nvsr.htm>. The birth defects rate data from 2010 to 2015 for Exhibit 2 were obtained from NCHS's User Guides for the Birth Data Files (NCHS, 2012b, 2014b,c,d, 2015c, 2016), available online at https://www.cdc.gov/nchs/data_access/vitalstatsonline.htm. The birth defects rate data from 2016 to 2020 for Exhibit 2 were obtained from National Vital Statistics Reports published by NCHS (2018a,b, 2019, 2021, 2022).

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NCHS. 2021. Births: Final data for 2019. National Vital Statistics Report 70(2). Table I-25. <https://www.cdc.gov/nchs/data/nvsr/nvsr70/nvsr70-02-tables-508.pdf> (PDF) (35 pp, 499K).

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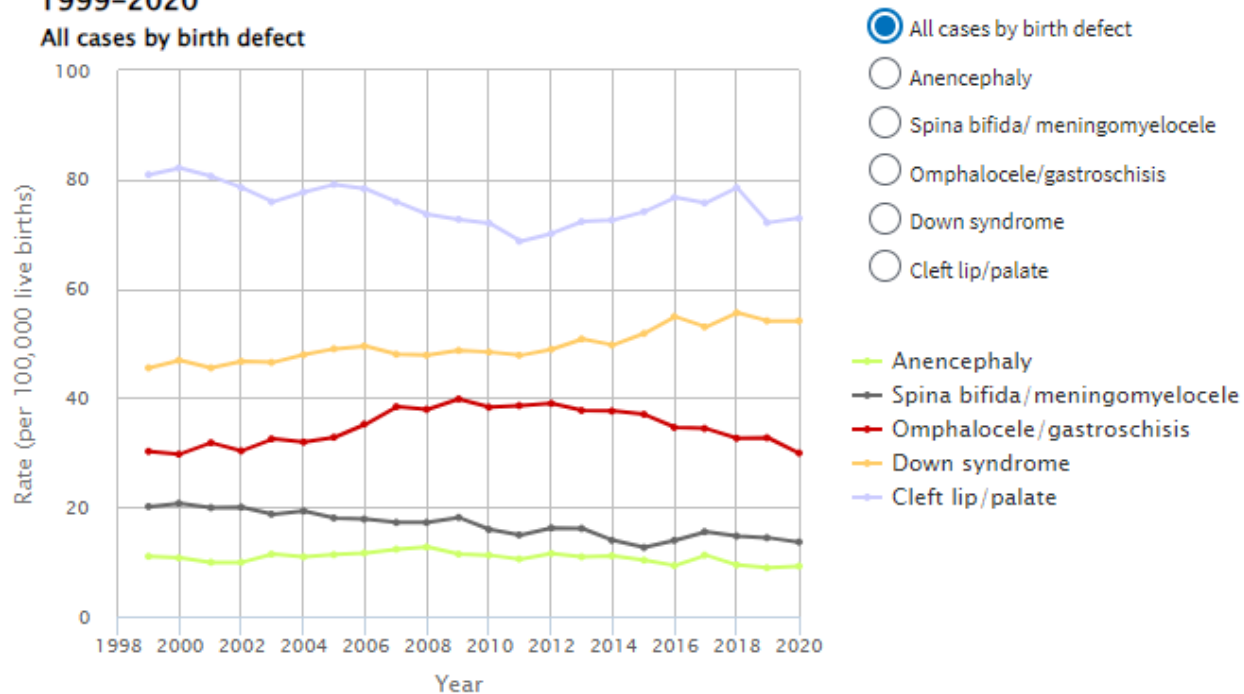
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Exhibit 1. Rate of live births in the U.S. with specific birth defects (congenital anomalies), as reported on the 1989 and 2003 Standard Certificates of Live Birth, 1999–2020

All cases by birth defect



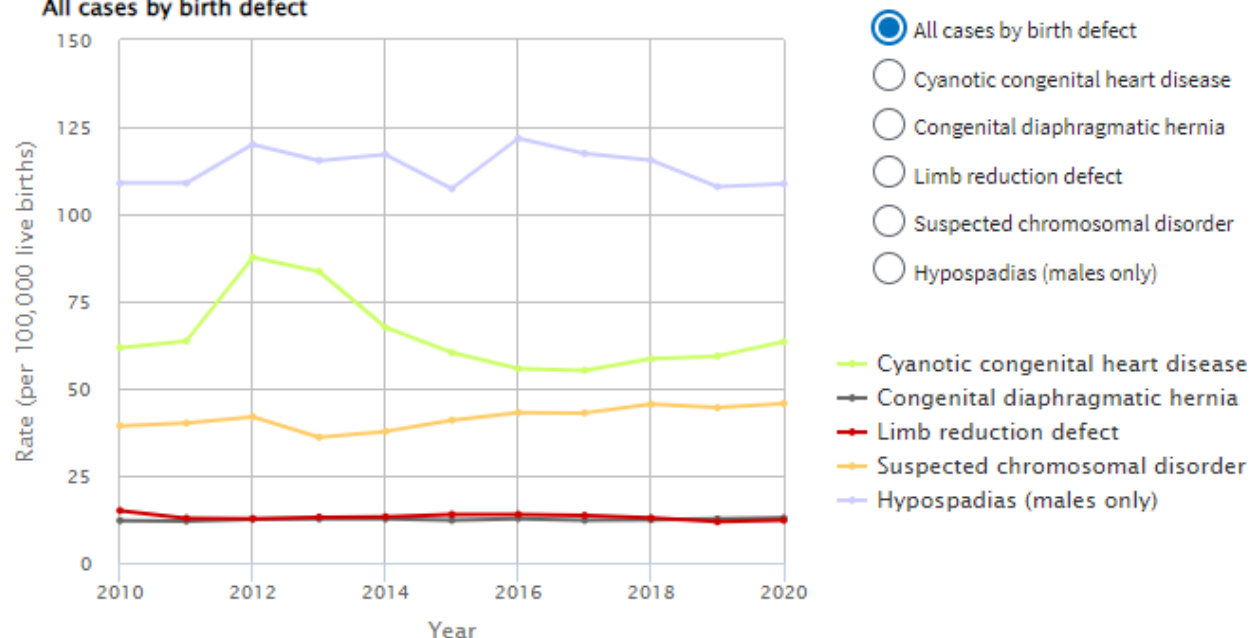
Information on the statistical significance of the trend in this exhibit is not currently available. For more information about uncertainty, variability, and statistical analysis, view the technical documentation for this indicator.

Data source: NCHS, 2001, 2002a,b, 2003, 2005, 2006, 2007, 2009, 2010a,b, 2011, 2012a, 2013, 2014a, 2015a,b, 2017, 2018a,b, 2019, 2021, 2022

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Exhibit 2. Rate of live births in the U.S. with five specific birth defects (congenital anomalies) as reported on the 2003 Standard Certificate of Live Birth, 2010–2020

All cases by birth defect



Anomalies were reported using the 2003 certificate by 33 states and the District of Columbia in 2010, with all 50 states and the District of Columbia using the 2003 certificate by 2016.

Information on the statistical significance of the trends 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, 2012b, 2014b,c,d, 2015c, 2016, 2018a,b, 2019, 2021, 2022

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