

## BIRTH DEFECTS

Birth defects, also known as congenital anomalies, occur in approximately 3 percent of all live births<sup>1</sup> and are the second leading cause of infant mortality behind prematurity, accounting for 20.9 percent of all infant deaths in the United States in 2011 (see page on infant mortality). Birth defects are conditions present at birth that most often occur during the first trimester of pregnancy and cause structural changes in one or more parts of the body.<sup>2</sup> Although most birth defects have unknown causes, they are thought to be caused by a combination of genetic, behavioral, and environmental factors. Some factors that have been linked to birth defects include tobacco, alcohol, and illicit drug use during pregnancy; obesity and uncontrolled diabetes; use of certain medications during pregnancy; a maternal age of more than 34 years; and a family history of birth defects.<sup>2</sup>

Congenital heart defects are the most common type of birth defect in the United States, affecting nearly 1 percent of—or about 40,000—births per year.<sup>3</sup> Atrioventricular septal defects, in which there is a hole

in the wall of the heart chambers and valves, are a common type of congenital heart defect, with about 2,000 cases annually (table 1). Among the chromosomal abnormalities, trisomy 21, or Down syndrome, is the most common, with about 6,000 annual cases. Orofacial defects, including cleft lip and cleft palate, are another common type of birth defect, with approximately 7,000 cases annually.

In 2011, congenital heart defects and chromosomal abnormalities were the leading categories of infant death due to birth defects, accounting for 23.6 and 19.5 percent of deaths attributable to birth defects, respectively. Central nervous system defects, also known as neural tube defects, were the third leading category at 13.6 percent.

Infant mortality rates due to birth defects vary by several demographic characteristics, including maternal age, race/ethnicity, educational attainment, and rural/urban residence. For example, infant mortality due to birth defects generally increased with rurality, ranging from 11.21 per 10,000 live births among residents of large fringe metro

**Table 1. National Prevalence Estimates of Selected Major Birth Defects,\* 2004–2006**

	Estimated Annual Number of Cases	Prevalence per 10,000 Live Births
<b>Congenital heart defects**</b>		
Atrioventricular septal defect	1,966	4.71
Common truncus	301	0.72
Hypoplastic left heart syndrome	960	2.30
Tetralogy of Fallot	1,657	3.97
Transposition of great arteries	1,252	3.00
<b>Chromosomal abnormalities†</b>		
Trisomy 13	528	1.26
Trisomy 18	1,109	2.66
Trisomy 21 (Down syndrome)	6,037	14.47
<b>Orofacial defects**</b>		
Cleft palate without cleft lip	2,651	6.35
Cleft lip with or without cleft palate	4,437	10.63
<b>Central nervous system defects**</b>		
Anencephaly	859	2.06
Encephalocele	341	0.82
Spina bifida without anencephaly	1,460	3.50

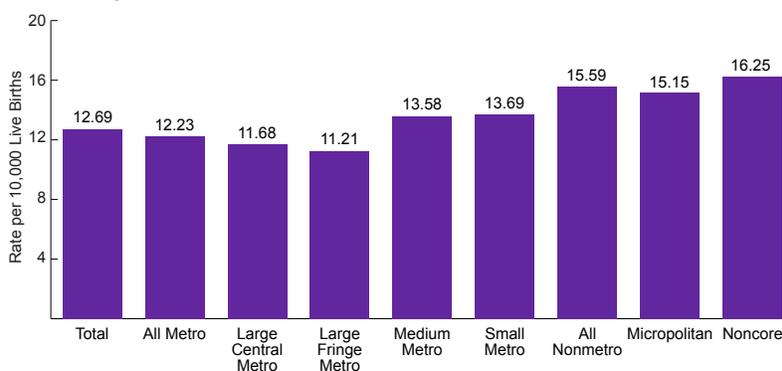
\*Fourteen programs contributed data: Arkansas, Arizona, California (eight-county Central Valley), Colorado, Georgia (five-county metropolitan Atlanta), Illinois, Iowa, Kentucky, Massachusetts, North Carolina, Oklahoma, Puerto Rico, Texas, and Utah. The number of live births represented by these 14 programs from 2004 to 2006 was 4,038,506. \*\*Estimates are adjusted for maternal race/ethnicity. †Estimates are adjusted for maternal age.

counties (suburban) to 16.25 per 10,000 live births among residents of noncore or completely rural counties (figure 1). Demographic differences in mortality rates attributable to birth defects may be due to differential exposures and the prevalence of birth defects, as well as differential access to screening and risk-appropriate care.

Certain birth defects can be prevented by maintaining a healthy weight before and during pregnancy, controlling diabetes, abstaining from substance use, talking to a doctor about which medications are safe to take during pregnancy, getting appropriate vaccinations to avoid infection, and taking a daily prenatal vitamin prior to and during

pregnancy.<sup>2</sup> In particular, taking folic acid before becoming pregnant has been shown to reduce the risk for neural tube defects by 50–70 percent.<sup>4</sup> Screening tests that can identify some birth defects can be administered during both the first and second trimesters of pregnancy and may include blood tests, ultrasounds, and/or testing of the placenta or amniotic fluid.<sup>2</sup> Screening healthy newborns using pulse oximetry can be a useful, cost-effective way to identify babies born with critical congenital heart defects before they are discharged from the birth hospital.<sup>5</sup>

**Figure 1. Infant Mortality Rates due to Birth Defects per 10,000 Live Births,\* by Urban/Rural Residence,\*\* 2011**



\*Infant deaths at less than 1 year of age with an underlying cause-of-death ICD-10 code of Q00–Q99. \*\*Based on: Ingram DD, Franco SJ. 2013 NCHS urban–rural classification scheme for counties. *National Vital Health Statistics*. 2014;2(166). Available at: [http://www.cdc.gov/nchs/data\\_access/urban\\_rural.htm](http://www.cdc.gov/nchs/data_access/urban_rural.htm). Accessed October 7, 2014.

#### Data Sources

Table 1. Parker SE, Mai CT, Canfield MA, et al. Updated national birth prevalence estimates for selected birth defects in the United States, 2004–2006. *Birth Defects Research, Part A: Clinical and Molecular Teratology*. December 2010;88(12):1008–1016.

Figure 1. U.S. Department of Health and Human Services, Centers for Disease Control and Prevention, National Center for Health Statistics. Underlying cause of death 1999–2011. CDC WONDER Online Database. 2014. Available at: <http://wonder.cdc.gov/ucd-icd10.html>. Accessed October 1, 2014.

#### Endnotes

1. U.S. Department of Health and Human Services, Centers for Disease Control and Prevention. Update on overall prevalence of major birth defects—Atlanta, Georgia, 1978–2005. *Morbidity and Mortality Weekly Report*. 2008;57(1):1–5.
2. U.S. Department of Health and Human Services, Centers for Disease Control and Prevention. Birth Defects. Available at: <http://www.cdc.gov/ncbddd/birthdefects/index.html>. Accessed October 3, 2014.
3. Reller MD, Strickland MJ, Riehle-Colarusso T, Mahle WT, Correa A. Prevalence of congenital heart defects in Atlanta, 1998–2005. *Journal of Pediatrics*. 2008;153:807–813.
4. Recommendations for the use of folic acid to reduce the number of cases of spina bifida and other neural tube defects. *MMWR Recommendations and Reports: Morbidity and Mortality Weekly Report*. September 11, 1992;41(RR-14):1–7.
5. Peterson, C, Grosse SD, Oster ME, Olney RS, Cassell CH. Cost-effectiveness of routine screening for critical congenital heart disease in U.S. newborns. *Pediatrics*. September 2013;132(3):e595–e603.

#### Suggested Citation

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