BIRTH DEFECTS

Birth defects occur in approximately 3 percent of all live births and are the leading cause of infant mortality, accounting for approximately 20 percent of all infant deaths in the United States in 2009.[25,26] Birth defects are conditions present at birth that cause structural changes in one or more parts of the body.[27] Birth defects develop before a baby is born and generally occur during the first trimester of pregnancy.

Although the causes of most birth defects are unknown, birth defects 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; uncontrolled medical conditions such as diabetes and obesity; use of some medications during pregnancy; maternal age younger than 20 and over 35 years; a family history of birth defects; and inadequate levels of folic acid (a B vitamin used by our bodies to make new cells) before and during pregnancy.[28] Taking folic acid before becoming pregnant has been shown to reduce the risk for neural tube defects by 50-70 percent.[28]

Congenital heart defects are the most common type of birth defect in the United States, affecting nearly 1% of—or about 40,000—births per year.[29] Trisomy 21, or Down syndrome, is a common birth defect with an estimated 6,000 cases identified annually.[26] Orofacial clefts, including cleft lip and cleft palate, are another common type of birth defect with approximately 7,000 cases identified annually.[26]

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. Screening healthy newborns using pulse oximetry can be a useful and cost-effective way to identify babies born with critical congenital heart defects before they are discharged from the birth hospital.[30] Although the survival of babies and children with birth defects has been improving, racial and socioeconomic disparities in the survival of these individuals with very special health care needs still persist.[31,32]

Selected Birth Defects, 2004-2006*

Source (I.10): National Birth Defects Prevention Network

Birth Defects	Cases per Births	Estimated Annual Number of Cases	Prevalence per 10,000 Live Births
Chromosomal anomalies**			
Trisomy 21 (Down syndrome)	1 in 691	6037	14.47
Trisomy 18	1 in 3,762	1109	2.66
Trisomy 13	1 in 7,906	528	1.26
Neural tube defects***			
Spina bifida without anencephaly	1 in 2,858	1460	3.50
Anencephaly	1 in 4,859	859	2.06
Encephalocele	1 in 12,235	341	0.82
Orofacial defects			
Cleft lip with and without cleft palate	1 in 940	4437	10.63
Cleft palate without cleft lip	1 in 1,574	2651	6.35
Cardiovascular defects			
Atrioventricular septal defect	1 in 2,122	1966	4.71
Tetralogy of Fallot	1 in 2,518	1657	3.97
Transposition of great arteries	1 in 3,333	1252	3.00
Hypoplastic left heart syndrome	1 in 4,344	960	2.30
Common truncus	1 in 13,876	301	0.72

**14 programs contributed data: Arkansas, Arizona, California [8-county Central Valley], Colorado, Georgia [5-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 2004-2006 was 4,038,506. **Estimates adjusted for maternal age. ***Estimates adjusted for maternal race/ethnicity.*