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The prevalence of selected major birth defects in the United States



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ABSTRACT

Although collectively they are fairly common, birth defects receive limited attention as a group of outcomes either clinically or from a public health perspective. This article provides an overview of the prevalence, trends and selected socio-demographic risk factors for several major birth defects, including neural tube defects, cranio-facial anomalies, congenital heart defects, trisomies 13, 18, and 21, and gastroschisis and omphalocele. Attention should focus on strengthening existing registries, creating birth defects surveillance programs in states that do not have them, and standardizing registry methods so that broadly national data to monitor these trends are available.

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Birth defects comprise a group of conditions, including physical or biochemical abnormalities, present at birth, which may involve malformations, disruptions, deformations. The term “birth defects” is often used synonymously with “congenital anomalies” or “congenital malformations,” however, these terms are to some extent artificially constrained by widely used coding systems such as the ICD-9 and ICD-10 rubrics.

Estimates of the overall prevalence of birth defects are complicated by inconsistent inclusion and exclusion criteria regarding major birth defects and minor defects or variants, as well as by differences in surveillance methods. Population-based data are not always consistently available for all conditions included in general definitions of major birth defects, rendering comparisons across registries difficult. Additionally, registries differ as to inclusion of stillbirths or terminations, as well as the duration of surveillance for live born infants. For these reasons, birth defects epidemiologists are often reluctant to provide estimates of the total prevalence of birth defects in a defined population. Having said that, the overall prevalence of birth defects among live born infants is generally regarded to be in the range of 3–5%, a figure widely cited though poorly documented. Birth defects

epidemiologists have achieved consensus as to whether to use the term “incidence” or “prevalence” in discussing occurrence of birth defects in populations.¹ Because birth defects by definition are present at birth regardless of when diagnosed, incidence does not apply and the term “prevalence” should be used.

Birth defects, taken collectively, are fairly common, and account for a disproportionate share of adverse perinatal outcomes. Infants born with birth defects are more than twice as likely to be born preterm (<37 weeks gestation),² and account for at least 20% of infant deaths³ and comprise a leading cause of death in early childhood.⁴

Birth defects receive less attention than other similarly common adverse pregnancy outcomes. Collectively, birth defects are considerably more prevalent than infant mortality, very low birth weight (<1500 g) or very preterm birth (<32 weeks gestation). The lack of attention likely results from the tendency to consider specific birth defects, or birth defects in a body system, as separate entities. This makes sense from both a clinical and a child/family systems perspective, as treatment, follow-up, and developmental issues differ depending on the nature and severity of the birth defect(s) a child might have. However, from a public health

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perspective, it heightens the challenges involved in organizing a comprehensive population-based response.

There is considerable interest in the prevalence of specific birth defects. From an epidemiologic perspective, spatial variation in prevalence may yield clues to environmental or behavioral risk factors, possibly genetic variation as well. Temporal trends can identify signals indicating emergence of new teratogens, and monitor the effects of population-based interventions on prevalence of specific birth defects. This article examines recent literature, primarily from North American sources, on the prevalence of selected birth defects, across time, registries, and by maternal characteristics and infant sex. While space does not permit an exhaustive treatment of all major birth defects, this overview should provide a sufficient introduction for the reader to explore additional topics of interest.

National prevalence estimates

The National Birth Defects Prevention Network developed national estimates for selected birth defects based on pooled data for numerous state registries for 1999–2001⁵ and 2004–2006.⁶ Additionally, a large pooled database with data from 15 state registries for 1999–2007 has supported several analyses of race/ethnic differences in prevalence and survival with selected major defects.^{7,8} These sources together with prevalence studies for specific defects were used for most of the statistics in this report. Other statistics based on birth certificates are published occasionally. While these data may be useful for exploration of trends over time, birth certificates typically under ascertain the specific birth defects reported, with overall sensitivity in the range of 20–30% compared to data from birth defects registries, with relatively poor predictive value positive.^{9,10} Clinicians and researchers should use data from population-based registries, especially those utilizing active case-finding methods, as the best sources for birth defects prevalence data.

Table 1 presents national estimates for 2004–2006 for selected major birth defects, using pooled data from 14 population-based birth defects registries. These data are drawn from the following registries: 11 with active case-finding, Arkansas, Arizona, California, Georgia, Iowa, Massachusetts, North Carolina, Oklahoma, Puerto Rico, Texas, and Utah, and three with passive ascertainment and follow-up, Colorado, Illinois, and Kentucky. For more details, refer to Parker et al.⁶ Race-specific comparisons of prevalence for selected major defects are presented in Table 2; for more details, refer to Canfield et al.⁷

In the next sections, we will examine prevalence, trends and patterns by race/ethnicity and other demographic factors for selected birth defects of general interest. This presentation is not meant to be comprehensive, but should provide basic information with adequate references to obtain more details elsewhere.

Neural tube defects

Neural tube defects result from the incomplete closure of the neural tube, at approximately 26–28 days after conception,

and include anencephaly and spina bifida. While anencephaly is lethal, spina bifida varies in prognosis with the complexity of the case presentation, whether it is an isolated condition, at which vertebrae the lesion is located and how deep the lesion is, and whether additional central nervous system complications such as hydrocephalus occur postnatally. These conditions are of particular interest because epidemiologic studies from the 1970s and two randomized controlled trials in the early 1990s demonstrated that periconceptional dietary supplementation with folic acid could significantly reduce the prevalence and recurrence of neural tube defects.¹¹

The prevalence of anencephaly and spina bifida in the United States declined by approximately 20% from 1995–1996 to 1999–2000, the period from immediately prior to mandatory fortification of wheat-based food products with folic acid by the US Food and Drug Administration effective January 1, 1998, to the first two full years thereafter. However, not only was the decline greater for spina bifida than for anencephaly, the rate of improvement varied by maternal race/ethnicity, and in recent years essentially no appreciable declines have been observed.^{12,13} The estimated prevalence of anencephaly among live births in the United States in 2004–2006 was 2.1 per 10,000 live births, and for spina bifida 3.5 per 10,000 live births.

The prevalence varies by state, by race/ethnicity, as well as by case-ascertainment methodology. Compared to infants born to non-Hispanic white mothers, prevalence of anencephaly and spina bifida was significantly higher among infants born to Hispanic mothers, while infants born to non-Hispanic black and to non-Hispanic Asian/Pacific Islander mothers had significantly lower prevalence of spina bifida (Table 2). Inclusion of data on stillbirths and prenatally diagnosed cases generally results in higher prevalence.

Cranio-facial anomalies

Cranio-facial anomalies, also referred to as oral or orofacial clefts, include cleft lip and cleft palate. Until recently, these were typically categorized as cleft palate without cleft lip, and cleft lip with or without cleft palate. Cleft lip with/without cleft palate is one of the most common birth defects, with an estimated national prevalence of 10.63 per 10,000 live births in 2004–2006, while the estimated prevalence of cleft palate alone was 6.35 per 10,000 live births (Table 1). Prevalence varies by race/ethnicity, with significantly lower prevalence among infants born to non-Hispanic black mothers (Table 2). Mortality is significantly higher during infancy and childhood among infants born to non-Hispanic black and Hispanic mothers.⁸ More recently, birth defects epidemiologists have begun classifying these conditions in three categories: cleft lip alone, cleft lip with cleft palate, and cleft palate alone.¹⁴

Congenital heart defects

Taken collectively, congenital heart defects comprise the most prevalent category of birth defects. In common with the reluctance to report an overall prevalence of birth defects

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