

Why an Atlas and for whom?

This Atlas has developed from interest in having one source of information on the birth prevalence of birth defects¹ from as many areas around the world as one might find. Historically, such information has been difficult to find and collate, leaving a considerable gap in the international assessment of birth defect prevalence. This Atlas, now in its second edition, aims at filling such gap. To do so, we have brought together many distinguished researchers involved in birth defect monitoring worldwide, who have provided published and unpublished information to this collaborative effort.

The burden of disease: the Atlas as a minimum estimate

The information from the Atlas can be used in many ways, some of which are more appropriate than others, as discussed in the sections that follow. Primarily, these data can be used to estimate the current world 'burden of disease' related to birth defects detected at or soon after birth. Because of the difficulties in ascertaining all cases of disease, and because in many areas fetal deaths and pregnancy terminations associated with birth defects are incompletely recorded, such estimate of the burden of disease should be considered as a minimum estimate only.

Even so, we trust that the considerable burden of birth defects will become readily apparent to anyone perusing the maps and tables included in the atlas. Public health professionals, health care experts, birth defect epidemiologists, and policy makers within each country are likely to find such data useful in their professional activities.

Interpreting geographic variations: some words of caution

Early on it was decided to use maps and tables as a reasonable way to summarize birth defect information. The tables allow us to present precise and detailed data, whereas maps provide a powerful visual summary for large segments of the information. Such an approach, however, is not without risks. In particular, the juxtaposition of data and maps from different areas inevitably leads the reader to focus on comparisons between countries and regions. While there may be times when such comparison can provide useful clues, in general it should be resisted.

In particular, the temptation might be to attribute variations in birth prevalence to variations in risk factor distribution or genetic susceptibility. In fact, geographical variation may reflect to a large degree what data are being collected and how, rather than true variations in occurrence. For examples, differences in prenatal screening, diagnostic services, methods of collecting epidemiological data, and even chance can all have a major impact on the recorded prevalence of birth defects in a given area or country. Because of the recognized impact in some parts of the world of prenatal diagnosis followed by termination of the affected pregnancy, we have attempted to provide, where available, the information on the proportion of terminations of pregnancy for each birth defect, as recorded by the registries. Thus, maps and tables are only a starting point in assessing the burden of disease represented by birth defects in different countries.

¹ The terms birth defects and congenital anomalies are often used interchangeably in the text, although technically these terms are not identical. "Birth defects" is more general and all-inclusive, referring to many kinds of abnormal development that originated in the prenatal period, whether present at birth or expressed later. Some birth defects such as certain inborn errors of metabolism can be diagnosed only with specialised techniques, while others are obvious to the naked eye.

Causes of birth defects

The geographic variations shown by the maps and tables must be interpreted with caution. Nevertheless, a careful study of the data might provide researchers with initial clues that might prove useful in etiologic studies of birth defects.

The significance of finding such causes cannot be underestimated. Birth defects are a major cause of infant mortality and childhood morbidity, affecting 2-3% of all babies (Stevenson, 1993; Stellman, 1986). They are also responsible for large numbers of embryonic and fetal deaths. In addition, birth defects are among the leading causes of years of potential life lost and contribute substantially to childhood morbidity and long-term disability.

Today, the causes of birth defects are largely unknown. Although genetic and environmental causes are thought to play a role, qualifying and quantifying their contribution to occurrence of birth defects has proven difficult (Kalter, 1983a; O'Rahilly, 1992; Mortensen, 1991). Overall, it has been estimated that single gene and chromosomal conditions might account for as many as one quarter of cases of birth defects, with chromosomal conditions having an even larger contribution to anomalies seen in spontaneous abortions and stillbirths.

The role of the environment (including the maternal environment) is even less clear, reflecting our limited understanding of their contribution to the etiology of birth defects (Schardein, 1993; Kalter, 1983b; Mortensen, 1991; O'Rahilly, 1992).

Some known environmental causes of birth defects such as maternal conditions (e.g., diabetes, rubella) or certain medications (e.g., valproic acid, retinoic acid), though relatively uncommon, are important to recognize because the exposure is preventable. An important environmental cause of birth defects, certainly of neural tube defects and perhaps many other defects, is insufficient folic acid intake. The major efforts at primary prevention are now being focused on periconceptional folic acid supplementation, whether by use of specific supplements or by food fortification (MRC, 1991; Czeizel, 1992; Berry, 1999).

Such prior information on the relation between exposure and outcome, combined with data such as those in the Atlas, can provide medical and public health with an appreciation of the preventable number of cases of birth defects, including, for example, the number of cases of spina bifida that could be averted through primary prevention using folic acid.

Where to find more information

The data presented in this second edition of the World Atlas come from registries participating in the International Clearinghouse of Birth Defects and EUROCAT (European Surveillance of Congenital Anomalies). For more information, readers are referred to the most recent reports of the International Clearinghouse of Birth Defects (ICBDMS, 2001) and EUROCAT (EUROCAT Working Group 2002) and websites (see www.icbd.org and www.lshtm.ac.uk/php/eeu/eurocat or www.eurocat.ulst.ac.uk).

Concluding comments

The primary aim of this Atlas is to provide an easily accessible resource with recent information on birth defect prevalence from many areas of the world. These data, while not exhaustive, represent the outcome of careful selection from programs whose primary function is to conduct effective monitoring of birth defects.

Collectively, the information in the Atlas represents a unique resource for medical and public health professionals involved in birth defect research, monitoring and prevention,

at both a national and international level. We hope that this collaborative effort will increase the appreciation of the global burden of birth defects, and possibly contribute to finding and implementing effective measures of primary prevention worldwide.

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