

**Abstract Title:** Global burden of Birth Defects Estimation: Methods, progress and selected results

**Authors:** Kathleen Strong, Scientist, World Health Organization, Geneva Switzerland; Hannah Blencowe, Associate Professor, London School of Hygiene and Tropical Medicine, London, UK; Helen Malherbe, Director of research and epidemiology, Rare Diseases South Africa, Cape Town, South Africa; Jamie Perin, Associate Scientist, Department of International Health, Johns Hopkins Bloomberg School of Public Health, Baltimore, Maryland, USA and the Burden of Birth Defects Technical Working Group

**Background:** Globally, birth defects are associated with substantial morbidity and mortality, accounting for 8% of under-5 child deaths in 2019, with the highest mortality found in low- and middle- income countries (LMICs). Global estimates for major birth defects for all countries were first published in 2006 by the March of Dimes in collaboration with the WHO<sup>1</sup>. The 2006 estimates were made for single gene disorders, chromosomal disorders, and malformations combining demographic data with data sourced from the literature and surveillance systems. These have since been updated as part of the Modell Global Database (MGDb) but not published. Estimates are also published by the Institute for Health Metrics and Evaluation (IHME) since 2012. However, these include a different range of conditions and methods and results have not been readily accessible. We present the process that WHO and partners are using to revise estimates of the global burden of birth defects with updated birth prevalence estimates from registries and literature.

**Methods:** In 2022, WHO formed a Burden of Birth Defects Technical Working Group (BBD-TWG) composed of 27 technical experts to review the previous estimation work, identify available data and advise on optimal approaches to estimation.

First, priority conditions were defined. Next, available prevalence and case-fatality data for these conditions were identified through registry review supplemented when necessary for specific conditions, time periods or geographical contexts by a review of the literature. Data were extracted into a standard database. Finally, the MGDb is being updated to include revised demographic, birth prevalence and case-fatality data. A repository for the MGDb that is on-line and open access will document:

- data inputs and methods;
- results by country, region and globally; and
- limitations of the results.

The mortality component for birth defects is also being updated using country Vital Registration (VR) data, registry data where it is aligned to outcomes and peer-reviewed studies.

Prevalence and mortality estimates of birth defects are being incorporated into *the Born Healthy Toolkit* for use in countries. Additionally, work is on-going with the IHME team to detail inputs, methods and results for birth prevalence of birth defects in the Global Burden of Disease (GBD) 2024 and to compare them with the results of the updated MGDb results.

**Results:** Five priority conditions of immediate relevance to policy makers and clinicians, were identified by the BBD-TWG for which prevention and care strategies exist, including:

- Structural anomalies that can be seen at birth or in uteri, including NTDs, orofacial clefts, abdominal wall anomalies;
- Trisomies and syndromic conditions
- Congenital heart defects
- Congenital hypothyroidism and congenital syphilis

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<sup>1</sup> Christianson A, Howson CP, Modell B. March of Dimes. Global report on birth defect. The hidden toll of dying and disabled children. New York. 2006.

- Haemoglobinopathies

Preliminary results will be presented for selected birth defects. These results will be compared with other estimates of congenital disorder burden and similarities and differences will be discussed.

**Discussion and conclusions:** National, regional and global health estimates can play an important role in raising visibility of specific health conditions and advocating for commensurate allocation of resources for implementing policies and programmes to tackle these conditions. Birth defects as a group have been neglected in many national child health strategies and policies, especially in LMICs where they are thought to be less prevalent than in high income countries. Our work shows that this is not the case and in fact, as child under-5 survival improves, the proportion of mortality due to birth defects increases. To capitalize on this finding, we focus on including available data from registries and studies in LMICs to close the data gap. Data improvement is the next step and this is being done by the development of congenital disorder registries in LMICs, notably sub-Saharan Africa with the sub-Saharan Africa Congenital Anomalies Network (sSCAN).

We hope to generate consensus on how to take global estimates further for advocacy. As a next step, we plan to support countries in using our revised estimates for planning purposes, for example, as part of the *Born Healthy Toolkit*.