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Ascertaining the Burden of Birth Defects

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In this issue of the *American Journal of Preventive Medicine*, Cui et al.[1] examined the congenital anomaly (CA)–specific under-5 mortality rate in China from 1996 to 2013 and found a substantial reduction in this rate from 407.7 per 100,000 live births in 1996 to 217.4 per 100,000 live births in 2013. In addition, they also found that the proportion of total under-5 mortality due to CAs (also referred to as birth defects) was increasing but were unable to include CA-specific data on stillbirths and terminations of pregnancy.

The Millennium Development Goal (MDG)-4, established in 2000, aimed to reduce under-5 mortality rate by two thirds by 2015.[2] Globally, the under-5 mortality rate has decreased by 53%, from an estimated rate of 90 deaths per 1,000 live births in 1990 to 43 deaths per 1,000 live births in 2015.[2] However, the MDG-4 goal of a two-thirds reduction in under-5 mortality rate was not achieved despite great progress made during the past few decades.[2] In January 2016, the MDGs were replaced by a new framework, the Sustainable Development Goals (SDGs), within which a new target to end preventable deaths of newborns and children under age 5 years was established with all countries aiming to reduce the neonatal mortality rate to 12 per 1,000 live births or fewer and the under-5 mortality rate to 25 per 1,000 live births or fewer by 2030.[3] In order to achieve this SDG, a major cause of neonatal and under-5 mortality, birth defects, must be addressed. More global efforts need to be undertaken in the prevention of birth defects to realize a reduction in neonatal and under-5 mortality.

WHO estimated that 276,000 neonatal deaths globally were attributable to birth defects, with neural tube birth defects (NTDs) being one of the most common and serious of these defects.[4] Since 1998, the U.S. and 80 other countries have mandated the fortification of staple food with folic acid to prevent the occurrence of NTDs, which affect the development of the brain and spine and result in mortality or lifelong disability.[5] The Food Fortification Initiative is an important advocate of such fortication.

In 2009, the Sixty-third World Health Assembly adopted a birth defects resolution in an effort to decrease the number of birth defects worldwide.[6] This resolution encouraged countries to develop population-based surveillance programs that accurately capture birth defects, including live births, stillbirths, and elective terminations of pregnancy for fetal

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anomaly (ETOPFAs).[6] As Cui and colleagues[1] commented in their article, if they had been able to assess the causes of deaths in stillbirths and induced abortions, the proportion of deaths from CAs would have been even higher than what they found in their study. Obeid et al.[7] estimated that more than two thirds of anencephaly-affected and more than half of spina bifida–affected pregnancies were terminated following prenatal diagnosis in their study of more than 9 million pregnancies in the European Surveillance of Congenital Anomalies surveillance region from 2000 to 2010. The authors also estimated that in Germany in 2009, counting only the number of live-born infants with spina bifida (the most common form of NTDs) would result in missing approximately 60% of NTD-affected pregnancies.[7] Similarly, a systematic review by Johnson and colleagues[8] found that the overall frequency of pregnancy termination following prenatal diagnosis was 83% for anencephaly (range, 59%–100%) and 63% for spina bifida (range, 31%–97%). In addition, Morris et al.[9] estimated in their study that the proportion of NTD-affected pregnancies resulting in a termination of pregnancy was 81% in the United Kingdom. Another study by Svensson and colleagues[10] estimated that the prevalence of birth defects among second-trimester terminated pregnancies was 14%. Incidentally, 14% of pregnancies with birth defects were ended by a second-trimester termination and the groups of birth defects with the highest proportion of second-trimester terminations included NTDs. If birth defects surveillance is limited to live births, the failure to ascertain all birth defects would likely lead to an underestimation of their true burden and contribution to child mortality. In addition, the recognition of physiological and psychological consequences that occur after stillbirths and ETOPFAs would be reduced. Inclusion of stillbirths and ETOPFAs would enable surveillance systems to better estimate the true burden of birth defects and to study the causes of these defects.

Capturing ETOPFAs and stillbirths is challenging because of several factors. For example, in countries where abortion is not permitted, women might seek an ETOPFA from other sources. In countries where pregnancy termination is available, information concerning the indication for ETOPFA might not be reported. Similarly, information about stillbirths and recognized birth defects might not be reported or otherwise readily available. Therefore, efforts to focus on developing a systematic approach to collecting and reviewing of data from ETOPFAs and stillbirths would be beneficial. Countries with existing birth defects surveillance programs may consider expanding their surveillance capabilities to include stillbirths because the necessary infrastructure and methodology are in place already.[11] Recognizing that many countries either do not have a birth defects surveillance system or a population-based surveillance system, WHO, CDC, and the International Clearinghouse for Birth Defects Surveillance and Research have recently released a manual titled “Birth Defects Surveillance: A Manual for Programme Managers” (www.cdc.gov/ncbddd/birthdefectscount/documents/bd-surveillance-manual.pdf), which is intended to serve as a tool for the development, implementation, and ongoing improvement of birth defects surveillance programs, particularly for countries with limited resources. CDC, the International Clearinghouse for Birth Defects Surveillance and Research, and WHO are also committed to providing technical assistance to countries to help develop or strengthen their birth defects surveillance systems.

As stated in the MDG Report 2015, "only by counting the uncounted can we reach the unreached."[2] If we want to accurately estimate the true burden of birth defects, we must develop better surveillance systems that include ETOPFAs and stillbirths.

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