

Foreword

Preventing Birth Defects: Implications and Prospects

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Efforts to lower maternal and neonatal mortality rates and enhance the quality of maternal and child care are pivotal, particularly in low- and middle-income countries (LMICs). These initiatives are critical for the realization of the Sustainable Development Goals by 2030. In 2019, congenital anomalies ranked as the tenth leading cause of global health loss (1). These conditions, also known as birth defects (BDs), are structural changes present at birth that can impact virtually any body part (e.g., heart, brain, foot). They are a significant contributor to early miscarriage, stillbirth, and fetal death. Individuals who survive these conditions often face disabilities, developmental delays, and a lifetime need for specialized medical care and health services. Therefore, comprehensive efforts to prevent BDs could substantially improve population health.

A sizable prospective cohort study conducted in China in the early 1990s found that daily periconceptional intake of 0.4 mg of folic acid could significantly reduce the risk of neuraltube defects (NTDs) (2), congenital limb reduction defects (3), and various other abnormalities. A nationwide folic acid supplementation program was not implemented until over a decade later, in 2009. Following the initiation of this program, there were marked increases in the rate of periconceptional folic acid supplementation and plasma folate concentration (4–5). Correspondingly, the prevalence of both NTDs and non-isolated NTDs with concurrent malformations dropped noticeably (6–7).

China implemented the universal two-child policy in 2016 and extended it to a three-child policy in 2021, anticipated to have a notable effect on maternal and infant health. In 2015, the Beijing Municipal Commission of Health and Family Planning innovated secondary prevention of BDs, establishing an integrated service management model of prenatal screening and diagnostic testing. Currently, non-invasive prenatal testing (NIPT), a superior accuracy high-throughput sequencing technology, has been instituted in 110 prenatal screening institutions and 10 prenatal diagnostic testing institutions in Beijing. The improved socioeconomic status and health literacy among the reproductive population prompted scientists to examine the influence of population policies and novel technology use on BDs epidemiology. This special issue contains four Preplanned Studies that address this question. Xia et al. reported an increased prevalence of chromosomal abnormalities (CAs) and their subtypes in Beijing over the last decade, based on hospital-based BDs surveillance data (8). Wang et al. revealed a significant decrease of both total BDs and NTDs in northern China over the past two decades (9). Wu et al. discovered, in Beijing, an increase in noncritical congenital defects (non-CCHDs) while the survival rate for critical congenital heart defects (CCHDs) remained stagnant despite advanced detection (10). Finally, Zhang et al. affirmed that strong compliance considerably mitigated the risk for NTDs (11).

Recent recommendations reaffirm that individuals intending to, or who may become pregnant, should consume a daily supplement containing 0.4 to 0.8 mg (400 to 800 µg) of folic acid, a widely supported prophylactic measure to prevent NTDs in their offspring. This recommendation, based on substantial evidence, bolsters the effectiveness of folic acid supplementation in disease prevention (12). In 2009, the Ministry of Health (previous) of the People's Republic of China launched the major public health project of “Folic Acid Supplementation to Prevent Neural Tube Defects”, providing free folic acid supplements for women with registered residence and family planning in rural areas. Since 2019, extensive national efforts have been made to include folic acid supplementation in the National Basic Public Health Service Project in China. It is heartening to note that such efforts persist and were incorporated into the National Basic Public Service Standards as of July 30, 2023, providing an optimal platform for preventing NTDs and other BDs. A sustained educational effort remains vital to ensure compliance with supplementation guidelines, including earlier initiation and appropriate supplementation durations (5). Concurrent advancements in diagnostic techniques and tools underscore the need for ongoing epidemiological research into folic acid-resistant NTDs, congenital heart defects (CHDs), and comorbid malformations (7). It is also crucial to explore the etiology including genomics (13), exposomics (14), and epigenomics in future studies.

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