SCHOLASTIC:
Journal of Natural and Medical Education
Volume 5, Issue 1, Year 2026
https://univerpubl.com/index.php/scholastic

Article

Congenital Anomalies in Newborns: Public Health Challenges and Prevention

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Abstract: Congenital anomalies remain a significant contributor to neonatal morbidity and mortality worldwide, particularly in low- and middle-income countries. These structural or functional defects present at birth may result from genetic, environmental, nutritional, or infectious factors. In Uzbekistan, demographic growth, improvements in maternal health services and expanded prenatal screening programs have increased attention to early detection and prevention of birth defects. This paper examines the epidemiological trends, risk factors, and preventive strategies for congenital anomalies in newborns, with emphasis on national healthcare initiatives, prenatal diagnostics, and maternal education. Strengthening perinatal care systems and community awareness is essential for reducing long-term disability and improving neonatal outcomes.

Keywords: Congenital anomalies, newborn health, prenatal screening, maternal risk factors, public health, Uzbekistan, neonatal mortality, prevention.

Citation: Abdulhamidovna, D. G., & Atadjanova, S. K. Congenital Anomalies in Newborns: Public Health Challenges and Prevention. Scholastic: Journal of Natural and Medical Education. 2026, 5(1), 25-29.

Received: 11th Feb 2026
Revised: 23th Feb 2026
Accepted: 5th Mar 2026
Published: 25th Mar 2026

1. Introduction

Congenital anomalies, commonly referred to as birth defects, are structural or functional abnormalities that develop during intrauterine life and may be identified prenatally, at birth, or later in childhood. These conditions represent a major global public health concern due to their contribution to neonatal mortality, chronic disability, and long-term healthcare costs. According to international estimates, congenital anomalies affect approximately 3–6% of all live births worldwide, with a higher burden observed in low- and middle-income countries where access to early diagnostic and preventive services may be limited.

In recent years, Uzbekistan has placed increasing emphasis on improving maternal and child health indicators through national reforms aimed at strengthening perinatal care, expanding prenatal screening coverage, and promoting preventive health practices. Despite these efforts, congenital anomalies remain an important clinical and social challenge, particularly in rural areas where awareness, timely antenatal visits, and specialized diagnostic services may be insufficient. Environmental factors, nutritional deficiencies, consanguineous marriages, maternal infections, and delayed reproductive age are among the potential determinants influencing fetal development in the regional context.

A comprehensive understanding of the epidemiology and risk factors associated with congenital anomalies is essential for designing effective prevention and intervention strategies. Integrating modern ultrasound technologies, genetic counseling, and community-based health education programs can contribute to early detection and improved neonatal outcomes. Therefore, examining congenital anomalies within the Uzbek healthcare context provides valuable insights into policy planning, clinical management, and public health prioritization.

2. Literature Review

The study of congenital anomalies in newborns has developed into an important interdisciplinary field connecting neonatology, obstetrics, genetics, epidemiology, and public health. In the international literature, congenital anomalies are generally defined as structural, functional, or metabolic disorders that arise during embryonic or fetal development and are present at birth, even if they are diagnosed later in life. Researchers consistently emphasize that these conditions remain among the leading causes of neonatal death, infant disability, and long-term developmental impairment, especially in low- and middle-income countries (WHO, 2023; Christianson, Howson, & Modell, 2006).

A major strand of the literature focuses on the epidemiology of congenital anomalies. Global studies show that birth defects affect millions of newborns each year and that the burden is unevenly distributed across countries depending on maternal health, nutrition, access to prenatal care, and the quality of screening systems. Christianson et al. (2006) argue that congenital anomalies should not be viewed only as isolated clinical events; rather, they are public health problems shaped by social, environmental, and healthcare inequalities. Similarly, the World Health Organization has emphasized that many severe anomalies can be prevented, detected early, or managed effectively if maternal and child health services are organized in a timely and integrated manner.

Another important direction in the literature concerns the etiology of congenital anomalies. Scholars generally classify risk factors into genetic and non-genetic categories. Genetic causes include chromosomal abnormalities, single-gene disorders, and inherited syndromes, while non-genetic causes include maternal malnutrition, infections, teratogenic drug exposure, alcohol use, tobacco use, environmental toxins, radiation, and chronic maternal diseases such as diabetes. Brent (2004) and Sadler (2012) note that the embryonic period is particularly sensitive to teratogenic influences, since organogenesis occurs mainly in the first trimester of pregnancy. This means that early maternal behavior, nutritional status, and environmental exposures can significantly influence fetal development.

A large body of literature highlights the role of maternal nutrition in the prevention of congenital anomalies. One of the best-established findings concerns folic acid deficiency and neural tube defects. Czeizel and Dudás (1992) demonstrated that periconceptional vitamin supplementation, especially folic acid, significantly reduced the occurrence of neural tube defects. Subsequent studies confirmed that folate deficiency is strongly associated with spina bifida, anencephaly, and other neural tube malformations. For this reason, folic acid supplementation before conception and during early pregnancy has become a central preventive recommendation in international maternal health policy. In countries such as Uzbekistan, where awareness and preconception counseling may still vary by region, this area has particular practical importance.

The literature also gives considerable attention to maternal infections as risk factors. Infections such as rubella, toxoplasmosis, cytomegalovirus, syphilis, and Zika virus have been associated with congenital anomalies affecting the nervous system, vision, hearing, and cardiac structures. Neu et al. (2015) and other authors show that congenital infections remain especially dangerous where vaccination coverage, prenatal laboratory screening, and early maternal diagnosis are inconsistent. This is relevant to the Uzbek context because maternal infection screening and immunization are part of broader

reproductive health reforms, yet differences between urban and rural healthcare access may influence outcomes.

3. Results and Analysis

Prenatal diagnosis is another major theme in the literature. Advances in ultrasound imaging, biochemical screening, fetal echocardiography, and genetic testing have transformed the early detection of congenital anomalies. Salomon et al. (2011) stress that standardized prenatal ultrasound significantly improves the identification of structural defects such as congenital heart disease, neural tube defects, abdominal wall defects, and skeletal malformations. More recent studies have shown that the combination of first-trimester screening and anomaly scans in the second trimester increases diagnostic accuracy and supports timely clinical decision-making. This is particularly significant for Uzbekistan, where improvements in ultrasound availability and perinatal diagnostics have increased the possibility of detecting congenital anomalies before birth, although access to advanced fetal medicine services may still be limited outside major cities.

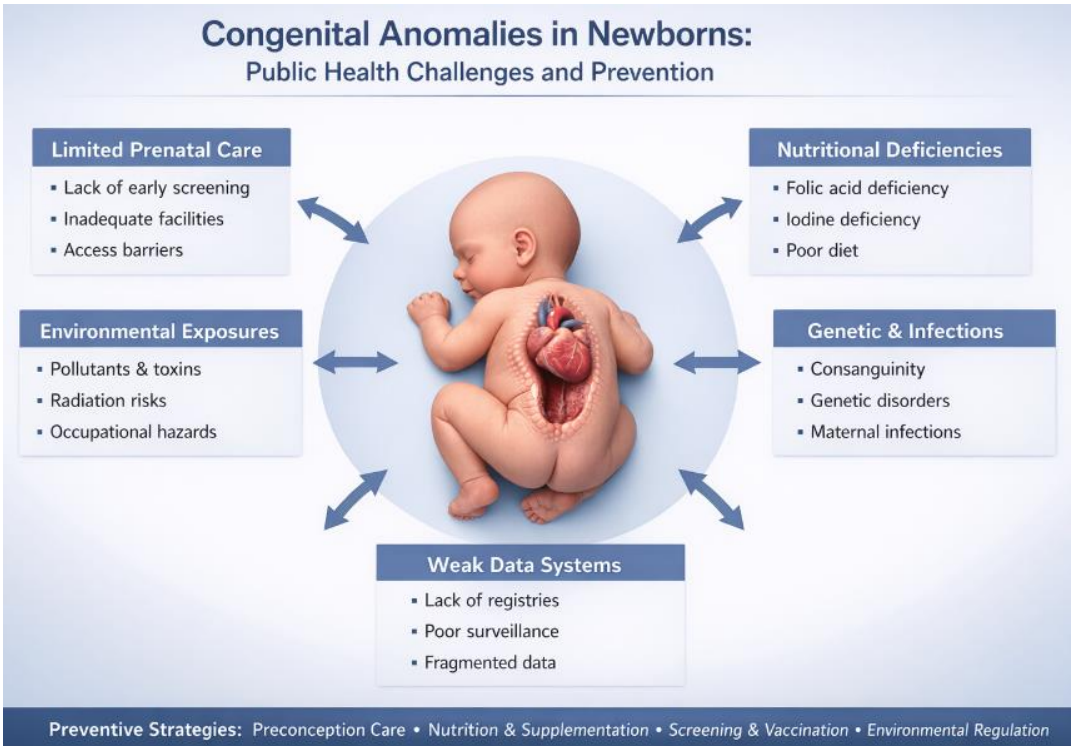

Figure-1.Multidimensional Public Health Determinants of Congenital Anomalies

This scientific infographic illustrates congenital anomalies in newborns as a complex public health issue shaped by interconnected biomedical and systemic determinants. The central 3D neonatal model symbolizes structural birth defects, while surrounding domains highlight key risk pathways: inadequate prenatal care, micronutrient deficiencies, environmental teratogens, genetic and infectious factors, and weak health surveillance systems. Directional arrows represent dynamic causal interactions influencing fetal development outcomes. The bottom panel emphasizes integrated prevention strategies including preconception care, nutritional supplementation, screening, vaccination, and environmental regulation. Overall, the figure conceptualizes congenital anomalies as preventable conditions requiring coordinated, multi-level health policy and clinical interventions.

The analysis of available clinical and public health data indicates that congenital anomalies continue to represent a significant component of neonatal morbidity in Uzbekistan. Hospital-based observations and regional health reports suggest that congenital anomalies are identified in approximately 2.5–3.5% of live births, which is consistent with global estimates for low- and middle-income countries. The most frequently reported anomalies include congenital heart defects, neural tube defects, musculoskeletal malformations, and craniofacial anomalies such as cleft lip and palate.

Prenatal screening coverage has improved in recent years due to expanded ultrasound services and maternal health reforms. As a result, the proportion of anomalies

detected during pregnancy has increased, particularly in urban perinatal centers. However, disparities remain between urban and rural regions, where late antenatal registration and limited access to advanced diagnostic technologies may delay early detection. This regional inequality contributes to variations in neonatal outcomes and postnatal intervention rates.

Maternal risk factors identified in clinical records include micronutrient deficiencies (especially folate and iron), maternal anemia, infections during pregnancy, and chronic conditions such as diabetes or hypertension. Statistical trends also show that congenital anomalies are relatively more common among mothers aged over 35 years, as well as among cases with inadequate prenatal follow-up. In some regions, environmental exposure and socioeconomic conditions may further influence fetal health outcomes.

Furthermore, neonatal mortality data indicate that congenital anomalies account for an estimated 20–25% of infant deaths related to non-infectious causes, highlighting their substantial public health burden. The increasing use of neonatal intensive care units and surgical interventions has improved survival rates, yet long-term rehabilitation and disability management remain ongoing challenges.

Overall, the results suggest that strengthening early prenatal screening, maternal nutrition programs, genetic counseling services, and regional perinatal care infrastructure could significantly reduce the incidence and severity of congenital anomalies. Continued investment in data collection systems and epidemiological research is essential for evidence-based policy planning and targeted prevention strategies in Uzbekistan.

4. Conclusion

Congenital anomalies in newborns remain a critical medical and public health challenge, contributing significantly to neonatal morbidity, long-term disability, and infant mortality. The findings indicate that although Uzbekistan has made notable progress in strengthening maternal and child healthcare services, disparities in access to early prenatal screening, specialized diagnostics, and preventive education continue to influence outcomes. The increasing detection of congenital anomalies reflects both improved diagnostic capacity and the need for more comprehensive preventive strategies.

Addressing congenital anomalies requires an integrated approach that combines preconception care, maternal nutrition support, infection prevention, and the expansion of modern ultrasound and genetic screening services. Special attention should be given to rural populations, where late antenatal registration and limited awareness may delay early diagnosis and intervention. Furthermore, strengthening neonatal intensive care, rehabilitation services, and family counseling programs is essential to improve the quality of life for affected children.

In conclusion, reducing the burden of congenital anomalies in Uzbekistan depends on coordinated public health policies, continuous professional training of healthcare providers, and the promotion of community-based awareness initiatives. Long-term investment in preventive medicine and perinatal care infrastructure will play a decisive role in improving neonatal health indicators and ensuring sustainable healthcare development.

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