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PREVENTION OF CONGENITAL MALFORMATIONS IN CHILDREN IN UZBEKISTAN: RISK FACTORS ANALYSIS AND RECOMMENDATIONS

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Abstract

The article examines the current problem of birth of children with congenital developmental defects in the Republic of Uzbekistan. Despite significant achievements in the field of maternal and child health protection, the establishment of perinatal centers, screening laboratories, and genetic counseling centers, the birth rate of children with congenital anomalies remains high, especially in ecologically unfavorable regions and areas with high rates of marriages between relatives. The work analyzes the main causes and risk factors for congenital malformations, including genetic characteristics, adverse environmental factors, vitamin and microelement deficiencies, and insufficient medical monitoring during pregnancy preparation.

Special attention was paid to the state programs being implemented in Uzbekistan aimed at preventing congenital pathologies, such as the "Healthy Mother - Healthy Child" program, developing the perinatal screening system, and providing folia to pregnant women.

Congenital anomalies represent not only a medical but also a socio-demographic problem, affecting the health level of the population, birth rates, disability structure, and family quality of life. An increased frequency of births of children with developmental defects is observed, first of all, in ecologically unfavorable regions, as well as in those areas where kinship marriages are common. Among the main risk factors, genetic predisposition, exposure to unfavorable environmental conditions, lack of vitamins and microelements, and lack of systematic preparation of women for pregnancy are highlighted.

In recent years, Uzbekistan has been implementing state programs aimed at preventing congenital pathologies, particularly the "Healthy Mother - Healthy Child" program, which provides for a set of preventive measures, early diagnosis and screening of pregnant women, providing women with folic acid, and developing the reproductive network.

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Introduction

Concept and classification of congenital malformations

Congenital malformations are persistent morphological or functional deviations resulting from intrauterine fetal developmental disorders under the influence of genetic, exogenous, or combined factors. These disorders can manifest as anatomical defects, functional disorders, metabolic disorders, or chromosomal abnormalities.

According to the World Health Organization (WHO) classification, congenital anomalies are divided into:

Chromosomal (e.g., Down syndrome, Edwards syndrome);
Genetically determined (monogenic) - inherited;
Multifactorial (multifactorial) - caused by a combination of hereditary and external factors;
Teratogenic (exogenous) - occurring under the influence of unfavorable environmental factors (infections, toxins, radiation, drugs, etc.).
Such defects can be incompatible with life or lead to disability, reducing the quality of life of the child and family.

Modern concepts of risk causes and factors

The occurrence of congenital defects is caused by the action of a complex of factors.
Genetic factors include chromosomal aberrations, genetic mutations, and hereditary diseases that often manifest in related marriages.
Environmental factors are associated with environmental pollution by heavy metals, pesticides, radiation, as well as a deficiency of microelements and vitamins (especially folic acid).
Social factors - the low level of medical culture of the population, lack of prenatal monitoring, early and related marriages, and harmful habits.
The period before and during pregnancy, when the woman's body is most sensitive to external factors, is of particular importance. Nutritional disorders, stress, chronic diseases, infections, especially viral ones (rubella, cytomegalovirus, toxoplasmosis), can significantly increase the risk of developing TPR.

Analysis of previous research in Uzbekistan

A number of domestic studies indicate that the frequency of congenital malformations in Uzbekistan varies depending on the ecological situation and socio-economic conditions of the region.
The highest prevalence is observed in the Fergana Valley, Karakalpakstan, and some districts of the Syrdarya region, where there are environmental problems and a high frequency of marriages between relatives.

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Scientists note that about 30-40% of congenital malformations can be prevented through effective primary prevention - improving pregnant women's nutrition, taking folic acid, abandoning harmful habits, and ensuring accessibility of prenatal monitoring.

In recent years, genetic research aimed at identifying hereditary diseases and informing the population about the risks of related marriages has been actively developing.

Thus, the analysis of available data shows the need for a comprehensive approach, including medical, social, and educational measures aimed at preventing congenital anomalies and forming a healthy generation in the Republic of Uzbekistan.

Analysis of the incidence of hereditary diseases in children in the Namangan region for 4 years (2022-2025)

Disease Name	2022	2023	2024	2025
Anencephaly	1	0	0	0
Spina bifida	14	14	16	12
Hydrocephalus	4	3	11	2
Microphthalmia	5	3	11	10
Cleft palate and lip	26	27	39	35
Esophageal atresia	6	5	2	6
Anal atresia	2	4	6	7
Reduction defect of the upper limb	22	18	20	26
Polydactyly	0	0	0	3
Renal agenesis	1	1	2	1
Diaphragmatic hernia	2	0	0	0
Umbilical hernia	1	0	0	0
Gastroschisis	1	1	1	0
Down syndrome	56	58	58	27
Multiple congenital anomalies	22	21	18	20
Other defects	29	40	41	30

For four years (2022-2025), an analysis of the incidence of congenital hereditary pathologies in children was conducted in the Namangan region. The most frequently registered defects were Down syndrome and multiple congenital anomalies. The number of Down syndrome cases remained consistently high in 2022-2024 (around 56-58 cases per year), however, a significant decrease to 27 cases is observed in 2025.

Other common defects include the hare's lip and wolf's palate (cleft palate and lip), which gradually increased from 26 cases in 2022 to 39 cases in 2024, with a slight decrease to 35 cases in 2025. Upper limb defects (reduction defects) also show a tendency towards growth, increasing from 22 to 26 cases. There is an increase in the number of such pathologies as microophthalmia and hydrocephalus: if in 2022 the number of cases of microophthalmia was 5, then by 2024 it increased to 11, and

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hydrocephalus increased from 4 to 11 cases. Similarly, the number of polydactyly cases increased in 2025 compared to previous years, although overall, these pathologies remain rare.

Some rare defects, such as diaphragmatic and umbilical hernias, were extremely rare and were recorded mainly during the first two years of observation.

Summarized data indicate that, despite the ongoing preventive measures, a sufficiently high level of congenital hereditary diseases persists in the Namangan region. Such groups as Down syndrome, congenital anomalies of the facial skull, and limb defects, requiring increased medical supervision and prevention, deserve special attention.

Preventive measures and recommendations

In modern healthcare, the prevention of congenital malformations is a priority task, as it allows reducing the burden on the healthcare system, reducing socio-economic costs, and ensuring the birth of a healthy generation. Based on the analysis of Namangan region data and international experience, several key areas of prevention and recommendations can be highlighted.

Development of proposals to reduce the risk of congenital malformations

The primary measure is to reduce the impact of factors contributing to the development of congenital anomalies. The main recommendations include:

Early screening programs: regular examination of women of reproductive age before and during pregnancy allows for the identification of risk factors (chronic diseases, infections, genetic predispositions). Screening should include ultrasound examination, laboratory tests, and specialist consultations.

Provision of necessary vitamins and microelements: it has been proven that a deficiency of folic acid, iodine, and some vitamins increases the risk of developing nerve tube defects, cardiovascular and other anomalies in the fetus. National programs should ensure access to folic acid for women of childbearing age.

Control of chronic diseases: correcting hypertension, diabetes, endocrine disorders in expectant mothers reduces the likelihood of pregnancy complications and birth defects.

Conclusion

Congenital malformations remain a significant medical and social problem in Uzbekistan, affecting the demographic situation, socio-economic development, and quality of life of the population. Analysis of Namangan region data for 2022-2025 shows that the most common pathologies are Down syndrome, multiple congenital anomalies, as well as facial skull and upper limb defects. Despite the introduction of modern medical technologies, the work of perinatal and screening centers, the incidence of congenital malformations remains quite high.

Effective prevention requires a comprehensive approach, including the early detection of risk factors, genetic counseling, improving the medical and social literacy of the population, and improving state policy in the field of reproductive health. The implementation of these measures will reduce the birth rate of children with congenital anomalies, ensure the formation of a healthy generation, and strengthen the system of maternal and child health protection in the country.

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Thus, the prevention of birth defects and informing the population are key areas for ensuring maternal and child health and require constant attention from the state, medical institutions, and society as a whole.

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