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Distribution of Congenital Anomalies in Children, Risk Factors

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Abstract---In world congenital anomalies are serious medical and social problem. They doubled over last decade 20th century and played significant role in the structure pathology and mortality in the infant, perinatal and children. Now time is 1800 out of 10,000 children born with congenital anomalies. Through progress made in our country in the future reform health system strengthening and the protection of the health children, congenital abnormalities play leading role in the structure morbidity disability and mortality. Reasons development of 40-60% anomalies is unknown. Apply the term "sporadic the defect of birth", meaning unknown cause, accidental appearance and low risk re appearance have future children. For 20-25% of anomalies more likely "multi-factor" reason - complex interaction many small genetic defects and risk factors environmental environment. The rest 10-13% of anomalies associated with the influence environment. Only 12-25% of anomalies have purely genetic causes (Akhmadalieva et al., 2020). Statistics shows disproportionate a high proportion of the total number survivors
newborns premature children who suffer physical, intellectual and emotional disability.

**Keywords**—anomalies, disability, health children, pathology, problem.

**Introduction**

Many thousands of primary health care workers work in the global health system. Their number is important not only because of their large number, but also because of the safe, effective, high-quality, wide range of medical services provided by them (Tukhtarov et al., 2020; Kamilova et al., 2010). The resolution of the International Council of Nurses states that without sufficient qualified specialists in nursing, preventive and medical care, especially chronic diseases, including congenital anomalies, cannot be achieved effectively and efficiently (International Council of Nurses, ICN 2013). (Yanchenko & Frolkova, 2021; Касимова, 2021). Therefore, one of today’s issues is to improve the health of the population, especially children, and to further improve the services of midwives, including community nurses, in the prevention of non-communicable diseases among them.

Despite the progress made in further deepening health care reform, strengthening and protecting the health of the population in our country, congenital anomalies (developmental defects) occupy a leading position in the structure of morbidity, disability and mortality among children. Based on this, scientific research will focus on the prevention and early study of congenital anomalies in primary health care facilities, identification of leading risk factors for the disease, a systematic approach to disease prevention, measures to improve the quality of life and quality of life is important. In the implementation of the above, the role of the closest and most direct approach to the population, as well as patronage nurses working in collaboration with the family doctor in the institutions of the ICU is significant (Kasimova, 2012).

Scientists and medical professionals from almost all countries are conducting research aimed at diagnosing and treating congenital anomalies. Over the next 10 years, views on the pathogenesis of congenital anomalies changed completely (Rizaev & Nurmamatova, 2018; Mirzarakhimova et al., 2019). According to statistics, congenital anomalies in the Republic of Uzbekistan range from 8,5 to 16,8 per 1,000 live births. The most common congenital anomalies are congenital cleft lip and palate, which is growing from year to year in Tashkent at 1: 745, and in Karakalpakstan at 1: 510. The purpose of the study. The study of the prevalence of congenital anomalies among children and risk factors, their prevention, development of scientifically based systemic measures to improve the patronage service.
Materials and Methods

Research objectives

The dynamics of the study of the characteristics of age and gender groups in the prevalence of congenital anomalies in children. To study the living conditions and lifestyle that cause congenital anomalies and to identify leading risk factors. Study, comparative assessment of the activities of the medical (community) nurse in the monitoring of congenital malformations in children, the implementation of preventive measures. Development of a set of science-based measures to improve the system of prevention of congenital malformations among children, increase its quality and effectiveness (Masharipova & Khasanova, 2020; Mirvarisova et al., 2018; Nurmamatova, 2016).

Subject of research


The scientific novelty of the research

For the first time, a systematic approach to the prevention and prevention of congenital anomalies in primary health care (PHC) is based on medical and organizational proposals for the comprehensive rehabilitation of mothers and children; Leading risk factors influencing the prevalence and origin and development of birth defects in children living in Tashkent for the first time in the country: mother’s age, education, family ties, poor family environment, number of pregnancies, complications during childbirth and extragenetic diseases in the mother detected (Kamilova et al., 2020; Mamatkulov & Avezova, 2015; Mirzarakhimova, 2020).

- A prognostic table has been developed to help develop a set of individual and group measures, a systematic approach to the prevention, early detection, timely diagnosis and treatment of birth defects in children.
- Deficiencies in monitoring the health of expectant mothers and children by nurses have been identified, which has a negative impact on the quality of care for expectant mothers and children in primary health care.

Globally, 7.9 million babies (6 % of births worldwide) are born with serious birth defects each year. Although some birth defects can be controlled and treated, approximately 3.2 million of these children remain permanently disabled. In addition, birth defects are a leading cause of infant mortality in the United States. But where do these defects come from? While some congenital defects are inherited, others are the result of harmful environmental factors known as teratogens, while others occur as a result of complex interactions of genetic and environmental influences. However, in about half of the cases of congenital
malformations, the causes are unknown (Mirvarisova et al., 2018; Mirzarakhimova et al., 2020; Mamatqulov et al., 2021).

In recent years, epidemiological studies have shown that between 5% and 10% of the world’s population suffers from congenital anomalies (Akhmadalieva et al., 2020). Congenital anomalies now rank first among the top five causes of infant mortality worldwide. Trends in infant mortality: Infant mortality has declined globally from 93% per 1,000 live births in 1990 to 39% in 2017. All regions of the WHO have halved the under-five mortality rate over the same period. Mortality rates in children under five remain unallocated. About 73% of under-five deaths occurred in 2017 in two regions, WHO Africa (49%) and WHO Southeast Asia (24%). The highest infant mortality rate was 74 in the WHO Africa region (74 per 1,000 live births) and 8 times higher than in the WHO European region (9 per 1,000 live births) (Mirzarakhimova et al., 2022; Denisova & Akhmadalieva, 2019; Mirzarakhimova et al., 2020).

In about 50% of cases, the exact cause of the congenital anomaly could not be determined, but there are certain risk factors that may be associated with the occurrence of the malformation. Congenital anomalies can be caused by single gene defects, chromosomal diseases, multifaceted inheritance, environmental teratogens (a means of causing birth defects), and micronutrient deficiencies (Denisova & Akhmadalieva, 2019, 2019). According to the World Health Organization (WHO), 270,000 people died in the first 28 days of life in 2018 due to congenital anomalies worldwide (Denisova & Masharipova, 2019; Mamatqulov et al., 2021).

According to the March Dimes (MOD) global report on birth defects, 7.9 million births (6% of all births) worldwide are born with serious birth defects each year, and 94% of such births occur in middle- and low-income countries. According to a joint report by the WHO and the MOD meeting, birth defects account for 7% of all neonatal deaths and 3.3 million deaths of children under five. The prevalence of birth defects in India is 6-7%, which accounts for about 1.7 million birth defects every year. Common congenital defects include congenital heart disease (8–10 per 1,000 live births), congenital deafness (5.6–10 per 1,000 live births), and fallopian tubes (4–11.4 per 1,000 live births).

Some birth defects are clinically apparent at birth. A structural defect, such as spina bifida, is evident at birth, but a hemophilia functional defect (bleeding disorder) is usually not apparent until infancy or childhood (Kasimova, 2012; Salimova et al., 2017). The Ministry of Health and Family Welfare has addressed the issue of the Government of India implementing various national health programs. In 2015, child health screening and early intervention services covered 30 cases of children aged 0–18 years using a variety of approaches (Rizaev & Ch, 2021; Yanchenko & Frolkova, 2021).

The Newborn Infant Action Plan (INAP), developed in India in September 2016, combines medical care with a special focus on maternal and child health to prevent and care for a newborn with a birth defect. INAP is a response by the WHO to the Newborn Action Plan (ENAP) to prevent neonatal death and birth. According to medical statistics in Uzbekistan, this figure is as follows: Infant
(under 1 year) mortality rate in 2016 was 10.7 per 1,000 live births, of which the main cause of death - deaths from perinatal events - 5.94, respiratory diseases of the organs - 2.44, congenital anomalies - 1.11, infectious and parasitic diseases - 0.35, other diseases - 0.84.

In 2016, the number of children dying under the age of five in the country was 10.1 thousand, and the infant mortality rate (under 5) per 1,000 live births was 14.1 per thousand. The infant mortality rate (up to 5 years) per 1,000 live births in the country was 48.2 per thousand in 1991, 28.5 per thousand in 2000, and 14.8 per thousand in 2010 (Mirzarakhimov et al., 2022; Denisova & Masharipova, 2019; Mirzarakhimova et al., 2020). According to statistics, in 2018, 154.7 thousand people died in Uzbekistan. This is 3.7% less than in 2017 [4,7].

Of those who died, 60.3 percent died from diseases of the circulatory system, 9.7% from various tumors, 5.6 percent from digestive organs, 4 percent from respiratory diseases, and 20.4% from other diseases. Among them, 7,500 are said to be infants under one year of age. 60.5% of infant deaths were due to perinatal events, 16.8% to respiratory defects, 11.7% congenital anomalies and 11% to other diseases (Rizaev & Nurmamatova, 2018; Mirzarakhimova et al., 2020). The rate of congenital anomalies is observed in industrialized countries: the United States and Africa. The study of the epidemiological aspects of congenital anomalies has led to the conclusion that the incidence of this disease has been steadily increasing over the last 10 years (Kasimova, 2012). These data suggest the need for an in-depth and comprehensive study of the origin of congenital anomalies. The research was conducted in several stages according to the goals and objectives of the scientific work.

### Table 1
Prevalence of congenital anomalies (developmental defects), deformities and chromosomal disorders (0-14 and 15-18 years) in children in the Republic of Uzbekistan (per 100,000 children)

<table>
<thead>
<tr>
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<tbody>
<tr>
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<td>1532.9</td>
<td>407.6</td>
<td>1464.2</td>
<td>363.1</td>
<td>1317.7</td>
<td>333.3</td>
<td>1370</td>
<td>313.2</td>
<td>1138</td>
<td>256.7</td>
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<td>230</td>
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<td>627.4</td>
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<td>496</td>
<td>271.9</td>
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<td>257.6</td>
<td>497</td>
<td>174.8</td>
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<td>264.2</td>
<td>165.3</td>
<td>214.5</td>
<td>128.2</td>
<td>217.1</td>
<td>71.3</td>
<td>134.4</td>
<td>289.2</td>
</tr>
<tr>
<td>Qashqadaryo</td>
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<td>200.4</td>
<td>476.8</td>
<td>130.4</td>
<td>465.1</td>
<td>167.2</td>
<td>433.4</td>
<td>183.6</td>
<td>411.4</td>
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<td>599.3</td>
<td>229.9</td>
<td>617.5</td>
<td>203.6</td>
<td>615</td>
<td>266.4</td>
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<td>195.5</td>
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<td>Namangan</td>
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<td>520.1</td>
<td>294.9</td>
<td>508.1</td>
<td>254.2</td>
<td>476.6</td>
<td>234.5</td>
<td>464.1</td>
<td>335</td>
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<td>Samarkand</td>
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<td>206</td>
<td>602</td>
<td>329</td>
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<td>Surxandaryo</td>
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<td>404</td>
<td>191.5</td>
<td>231.7</td>
<td>224.7</td>
<td>298.4</td>
<td>274.3</td>
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<td>407.7</td>
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<td>394.9</td>
<td>214.2</td>
<td>392.9</td>
<td>232.4</td>
<td>397.6</td>
<td>200.2</td>
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<tr>
<td>Tashkent region</td>
<td>1166.5</td>
<td>354.6</td>
<td>949.2</td>
<td>810.2</td>
<td>960.3</td>
<td>458.5</td>
<td>1014.1</td>
<td>415.4</td>
<td>928.3</td>
<td>419.3</td>
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<td>325.5</td>
<td>885.6</td>
<td>296.8</td>
<td>894.9</td>
<td>396.9</td>
<td>819.8</td>
<td>267.6</td>
<td>1021.8</td>
<td>377.5</td>
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<tr>
<td>Khorezm</td>
<td>771.5</td>
<td>350.9</td>
<td>619.7</td>
<td>298.1</td>
<td>393.7</td>
<td>206</td>
<td>393.6</td>
<td>170.6</td>
<td>386.1</td>
<td>155.8</td>
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<tr>
<td>The Republic of Karakalpakstan</td>
<td>319.8</td>
<td>113.4</td>
<td>334.4</td>
<td>131.7</td>
<td>325.7</td>
<td>121.5</td>
<td>394.1</td>
<td>102.5</td>
<td>301.6</td>
<td>84.1</td>
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<tr>
<td>Republic of</td>
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<td>267.1</td>
<td>626</td>
<td>295.2</td>
<td>571.5</td>
<td>261.7</td>
<td>574.6</td>
<td>252.3</td>
<td>598.8</td>
<td>264.5</td>
</tr>
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</table>
Results and Discussions

We have developed a comprehensive research program and plan to conduct an in-depth study of the prevalence and risk factors for congenital anomalies in children. Given that the number of observations per calendar year is insufficient to conduct scientific research, it is recommended to study and calculate them for at least five years, given that congenital defects in the fetus may reflect elements of a random nature and be unreliable. In-depth study of the prevalence and risk factors of congenital anomalies in children has been carried out by us in dynamics for 5 years (2015-2019). A total of 7783083 congenital anomalies were studied in the country: 5230 of them in Tashkent.

The ultimate goals and objectives of this study were to select methodological approaches that studied the risk factors for congenital anomalies in children, the proper preparation of women of reproductive age for pregnancy and their status during pregnancy, and the satisfaction of patronage nursing providers in family clinics. The study was conducted in a multi-stage manner based on the districts of Tashkent.

In the first stage, to properly select the object of research, to obtain representative information about the causes of morbidity, disability and death of children from congenital anomalies, as well as to study the factors of lifestyle and living conditions that may affect children’s health, sanitary and demographic indicators of Tashkent The medical services provided to the population of the districts, as well as the condition of family clinics in the city were analyzed on the basis of data from the Tashkent City Health Department. In the example of the city of Tashkent, congenital anomalies were studied in all districts of the general census.

In the second stage, we studied the prevalence of congenital anomalies in children, the structure of their age, gender characteristics, the dynamics of disease. For this purpose, in order to obtain reliable and representative data, first of all, the number of selected complexes was determined depending on the number of general complexes. The table “Number of observation units with an error of no more than 5% of the survey results when the general complex is accurate” was used to generate a selected set from the General set (Mirzarakhimova & Nurmamatova, 2017; Sherkuzieva et al., 2017).

As a result, family clinics of 5 districts of Tashkent: Mirabad, Mirzo Ulugbek, Yunus Obod, Almazar, Yashnabad districts were selected by cluster method using the “Bird’s Nest” method and studied the health of children (0-18 years old) attached to them. So put In order to obtain information about congenital anomalies, the risk factors that cause them, we analyzed in detail each of the data obtained from the mothers of children born with congenital anomalies. To compare the birth rate with healthy and congenital anomalies, these figures were calculated separately for the city and districts of Tashkent.

It should be noted that since 2015, the Ministry of Health of the Republic of Uzbekistan and the State Statistics Service have jointly moved to the international system for determining the criteria for births of children with congenital
anomalies. However, the country uses the old system of calculation of indicators to monitor the state program to improve the health of children and women, which is convenient for comparing ongoing processes. However, to facilitate the analysis of the causes of congenital anomalies, we decided to use the criteria used in many countries around the world.

To study the morbidity in young people, we analyzed children congenital anomalies according to the internationally accepted scheme of the following age groups according to the following periods: 0-14, 15-18 age groups. Age-appropriate marriage - Parents should pay close attention not only to maintaining women's health during pregnancy, but also to the healthy growth of adolescent girls from an early age. The most favorable period for women to have children is 20-30 years. If the pregnant woman is less than 20 years old, then up to 30% of the fetus is underdeveloped, the baby is underweight, the number of stillbirths doubles, and the number of maternal deaths among young mothers triples. Apparently, the negative consequences of early marriage are many. In this, first of all, the health of the mother plays an important role. The reason is that a healthy mother gives birth to a healthy child (Denisova, 2020; Inakov et al., 2020; Kamilova et al., 2021; Rustamova & Tursunkulova, 2016).

Inbreeding - Many scientific studies show that such marriages have a negative impact on the health of the unborn child. Such a marriage is called Exugami. For example, about 82 diseases have been identified in marriages between close relatives. It can also be said that while the incidence of the disease in incestuous marriages is 7 to 8% out of 100, the rate among strangers is 100 to 5%. Concomitant diseases of the pregnant woman - Anemia, TORCh infection, Fetal viability, Thalassemia, Muscular dystrophies, Facioscapulohumeral-dystrophy FSHD (DNA), Colorectal cancer, Cardiovascular diseases have a negative impact on the child with disabilities and mental growth and development.

GRIPP or influenza during pregnancy - If the expectant mother catches a cold in the early stages of pregnancy, the fetus may develop symptoms of oxygen deficiency and the fetus may lag behind in development. Therefore, in this case, it is extremely important to apply treatment measures aimed at eliminating the infection in the body. Severe viral infections can affect the fetus in the form of abnormalities in the development of the organs that began to form during the mother's cold. Influenza can cause adverse events such as increased and turbid water surrounding the fetus, risk of miscarriage, premature birth, and deterioration of a woman's health (especially if the immune system is damaged). The highest rates of congenital anomalies in the Republic of Uzbekistan in 2014-2015 were higher in Tashkent among children aged 0-14 years (1364,6 ‰ per 100,000 live births), in Tashkent and Fergana regions (1003,7 ‰ -901,5 ‰) compared to the national average. formed the size. The lowest rates of congenital anomalies are in the mountainous regions of the country, Jizzakh - 203,5 ‰, Andijan - 331,3 ‰, Syrdarya - 324,9 ‰.

As mentioned above, the city of Tashkent is a leader in the country in terms of the prevalence of congenital anomalies. In the Tashkent region, the incidence of the disease has increased dramatically. In particular, in 2018, the incidence
increased by 40.7% compared to 2014. Although the incidence decreased slightly in 2015, by 2017 the figure had reached a new high.

Table 2
Prevalence of congenital anomalies (developmental defects), deformities and chromosomal disorders among children in 2014-2018 (average 5 years (per 100,000 children))

<table>
<thead>
<tr>
<th>Name of regions</th>
<th>Average 5 years</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>0-14 years</td>
</tr>
<tr>
<td>Tashkent city</td>
<td>1364.6</td>
</tr>
<tr>
<td>Andijan region</td>
<td>331.3</td>
</tr>
<tr>
<td>Bukhara region</td>
<td>537.6</td>
</tr>
<tr>
<td>Jizzakh region</td>
<td>203.5</td>
</tr>
<tr>
<td>Kashkadarya region</td>
<td>461.4</td>
</tr>
<tr>
<td>Navoi region</td>
<td>545.8</td>
</tr>
<tr>
<td>Namangan region</td>
<td>491.5</td>
</tr>
<tr>
<td>Samarkand region</td>
<td>655.6</td>
</tr>
<tr>
<td>Surkhandarya region</td>
<td>324.9</td>
</tr>
<tr>
<td>Syrdarya region</td>
<td>397.0</td>
</tr>
<tr>
<td>Tashkent region</td>
<td>1003.7</td>
</tr>
<tr>
<td>Fergana region</td>
<td>901.5</td>
</tr>
<tr>
<td>Khorezm region</td>
<td>512.9</td>
</tr>
<tr>
<td>The Republic of Karakalpakstn</td>
<td>335.1</td>
</tr>
<tr>
<td>Republic of Uzbekistan</td>
<td>601.5</td>
</tr>
</tbody>
</table>

As shown in the table, the incidence of congenital anomalies in the country, although much lower in the regions than in Tashkent, is growing rapidly from year to year. We have achieved the following results in comparison with the scale of our republic. As shown in the table, the incidence of congenital anomalies among children aged 15-18 years is not evenly distributed in the region and in Tashkent. In particular, in Tashkent city (334.8‰), Tashkent region (491.6‰), Fergana region (332.9‰), Namangan region (303.2‰), Samarkand region (288.3‰), Bukhara regions (282.5‰) incidence is also slightly higher in the country than in other regions. The lowest morbidity rates were observed in Jizzakh region (159.1‰), Kashkadarya region (165.9‰), and the Republic of Karakalpakstan (110.6‰).

Tashkent is the largest city in Central Asia. There, manufacturing, industry flourished. Population density, well-developed urban infrastructure, well-provided medical care, including specialized medical care, allows for early detection, early diagnosis and reliable information about the spread of diseases. The city of Tashkent is a leader in the country in terms of the prevalence of congenital anomalies. That is why we decided to conduct this study in the cities and districts of Tashkent. Prevalence of congenital anomalies (developmental defects), deformities and chromosomal disorders among children in 2015-2018
According to the analysis, congenital anomalies in Tashkent due to medical reasons, including social factors, are the highest rate of congenital anomalies in children in Tashkent over the past five years (1464,4) observed, and in recent years a dynamic decline is felt. In 2019, its rate was 100,000 live births (1103,0). It can be seen that the rate of congenital anomalies in Tashkent over the past five years decreased from 1464.4 in 2015 to 1103,0 in 2019 per 100 thousand live births, ie the rate of congenital anomalies in Tashkent decreased by 1,5 times. In order to provide a socially hygienic description of families, we conducted a survey of families with children with congenital anomalies in 300 healthy and 300 families with congenital anomalies in polyclinics in 5 districts of Tashkent: Mirabad, Mirzo Ulugbek, Yunus Obod, Almazar, Yashnabad.

The study of factors such as family lifestyle, living conditions, medical activity of parents revealed that these factors affect the birth of children with congenital anomalies. First of all, the presence of hereditary diseases in the family, children with congenital anomalies, including these families in the "high-risk families", reduces the risk of having a healthy child in this family, including the results of our research. 14% of families had children with congenital anomalies, and 17% of families had a predisposition to hereditary diseases.

Among biological factors, the interval between births is one of the factors that strongly influences the health of children. It was noted that children's health indicators were inversely proportional to the interval between births, and that the smaller the interval, the higher the risk of children being born with developmental defects. In about 70% of cases, children are born at intervals of up to 3 years. The results of the survey showed that the interval between births was 3 years or more in 40% of mothers, 2-3 years in 30% of mothers, 1-2 years in 24% and even 1 year in 6% of cases.

Given the negative impact of in-law marriage on children's health indicators, we found it necessary to study it. Our study found that children from families whose parents were related were 4.0 times more likely to be born with congenital anomalies than children from families whose parents were not related. 6.0% of children born with birth defects were found to be close relatives of their parents,
and 20.0% were found to be distant relatives. The health of the parents determines the health of the family. It was found that children in a family with a chronic illness or one of the parents with frequent acute illnesses were 2.0 times more likely to have the disease than children born to healthy parents. The data obtained showed that the birth of children with various hereditary diseases, extragenital diseases of the mother in the development of congenital anomalies (anemia, nephropathy, gynecological diseases), late pregnancy (toxicosis, gestosis) have a strong effect on the health of children.

In the course of the study of medical social factors, it became clear that the majority of women with chronic and gynecological diseases of mothers during pregnancy, which inevitably affects the health of the unborn child. 24.8% of mothers had a history of anemia at birth, 27.3% had various infectious diseases, 10.4% had kidney disease, and 5.3% had heart defects. All this has an impact on the health of the unborn child.

Conclusion

The practical results of the study are as follows

A comprehensive, systematic approach to primary, secondary and tertiary prevention of congenital anomalies in the primary care system has been developed. On the basis of this program, an algorithm of measures for early detection and prevention of congenital anomalies, as well as practical recommendations for the formation of healthy lifestyle skills among the population, improving the care of expectant mothers and children. Comprehensive assessment of the level of risk factors influencing the formation of congenital anomalies in the activities of nurses in family clinics, identification of risk groups of the population, development of a prognostic table that allows differentiated, individualized measures according to their risk level.

Scientific and practical significance of research results

The scientific significance of the results of the study is explained by the fact that the conclusions and recommendations received play an important role in the development of patronage services and contribute to the optimization and improvement of medical nursing care for expectant mothers and children. The practical significance of the results of the study is that a number of recommendations have been developed and implemented in health care to improve the care of expectant mothers and children. increase the level of satisfaction with the service. The prevention of congenital anomalies is explained by the creation of a prognostic table that allows them to be divided into leading risk groups, to conduct targeted dispensary control.

References


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