International Journal of Innovative Research and Scientific Studies, 8(5) 2025, pages: 983-995

IJIRSS
International Journal of Innovative Research & Scientific Studies

ISSN: 2617-6548

URL: www.ijirss.com

Epidemiological pattern and regional characteristics of congenital malformations in the Turkistan region

Ardak Ayazbekov[1], Ainash Oshibayeva[1], Secil Ozkan[2], Gulzhaukhar Taskinova[1], Makpal Taubekova[1*]

[1]*Khoja Akhmet Yassawi International Kazakh-Turkish University, Kazakhstan.*
[2]*Gazi University Medical Faculty department of public health Teknikokullar, Turkey.*

Corresponding author: Makpal Taubekova (*Email: makpal.taubekova@ayu.edu.kz*)

Abstract

This article explores the epidemiological pattern and regional characteristics of congenital malformations (CMs) in the Turkistan region. The study is based on data collected from regional perinatal centers and healthcare institutions from 2020 to 2024. During this period, 53,169 pregnancies were registered, and congenital malformations were diagnosed in 848 cases (1.6%). Year-by-year analysis revealed a consistent increase in CM prevalence: 0.8% in 2020, rising to 2.4% in 2024. The dynamics of absolute and relative growth rates were assessed, and regression modeling was applied to forecast future trends. Projections indicate that the number of CM cases could reach 310 in 2025 and 357 in 2026. The findings underscore the need to monitor contributing factors, enhance preventive measures, and improve screening programs. The resulting data serve as a critical foundation for planning and optimizing perinatal care in the Turkistan region.

Keywords: Congenital malformations, Turkistan region, Epidemiology, Pregnancy, Screening, Forecast, regression.

DOI: 10.53894/ijirss.v8i5.8922
Funding: This study received no specific financial support.
History: Received: 17 June 2025 / **Revised:** 21 July 2025 / **Accepted:** 23 July 2025 / **Published:** 29 July 2025

Competing Interests: The authors declare that they have no competing interests.
Authors' Contributions: All authors contributed equally to the conception and design of the study. All authors have read and agreed to the published version of the manuscript.
Transparency: The authors confirm that the manuscript is an honest, accurate, and transparent account of the study; that no vital features of the study have been omitted; and that any discrepancies from the study as planned have been explained. This study followed all ethical practices during writing.
Institutional Review Board Statement: The Ethical Committee of the Khoja Akhmet Yassawi International Kazakh-Turkish University, Astana, Kazakhstan has granted approval for this study
Publisher: Innovative Research Publishing

1. Introduction

1.1. Relevance of the Research Problem

Congenital malformations (CMs) are recognized as one of the most pressing issues in modern biomedical science and public health. According to the World Health Organization, approximately 4–6% of newborns worldwide are diagnosed each year with various congenital anomalies. A portion of these cases result in neonatal mortality, while others lead to permanent disabilities that significantly reduce quality of life [1]. Due to their profound medical and social impact,

congenital malformations are considered a priority in the health policies and programs of many countries.

In scientific literature, the etiology of congenital malformations (CMs) is understood to follow a multifactorial model. Alongside genetic disorders, a complex interplay of social, environmental, and medical factors plays a significant role in their development. In particular, environmental agents that affect a woman's body during pregnancy, including chemical, physical, and biological factors as well as maternal somatic and infectious diseases, significantly increase the risk of CMs [2]. Although the precise mechanisms by which these factors influence fetal morphogenesis remain incompletely understood, their clinical relevance is well established.

International practice relies on epidemiological registries such as EUROCAT (European Surveillance of Congenital Anomalies) and ICBDSR (International Clearinghouse for Birth Defects Surveillance and Research), which provide data to describe regional differences in the structure and frequency of CMs, as well as temporal trends [3] Figure 1.

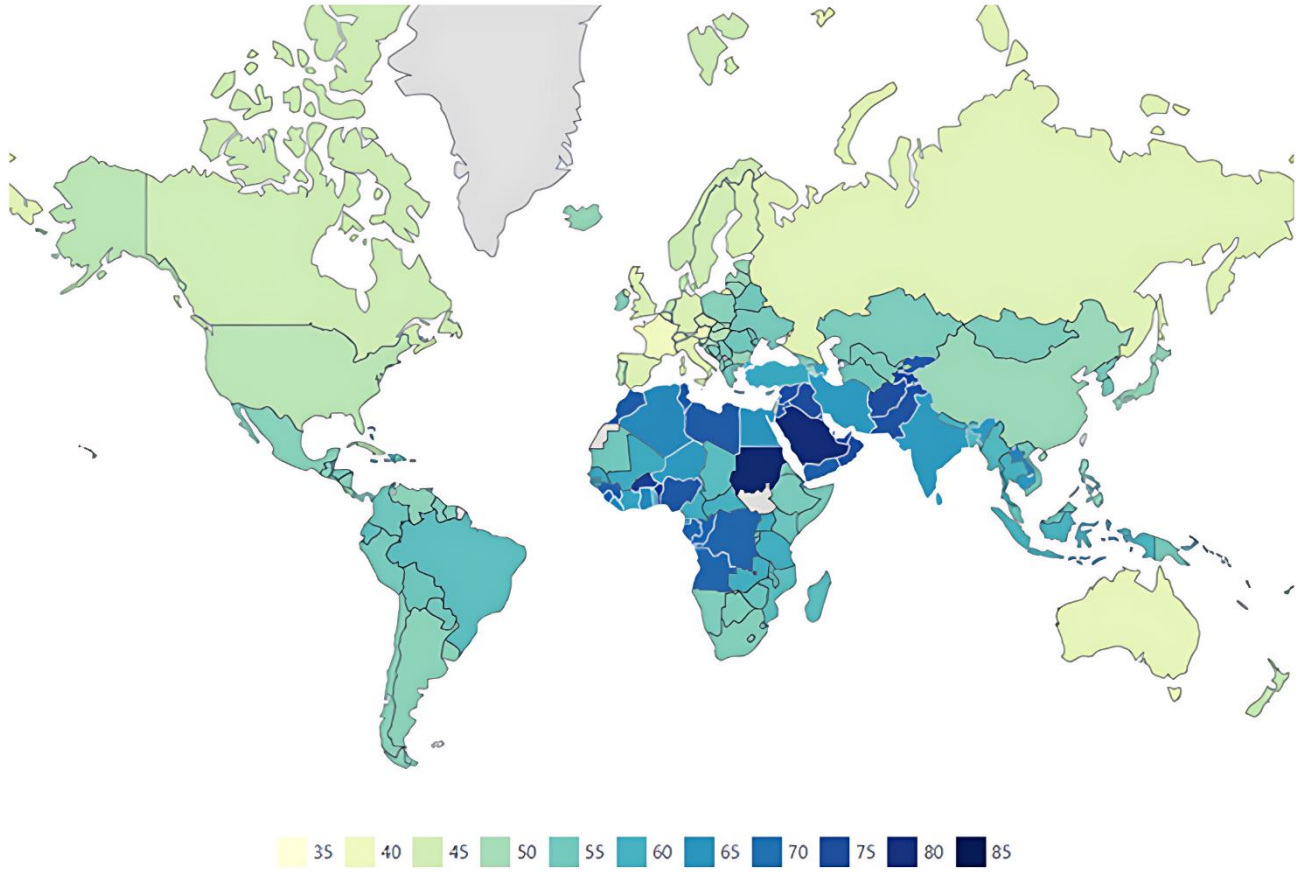

Figure 1.
Prevalence of Congenital Malformations per 1,000 Live Births.

These data indicate that the prevalence of congenital malformations is influenced by the population's ethnic, socio-economic, and environmental characteristics. Additionally, the quality and accessibility of medical care significantly affect both the detection rate and registration of such cases.

In the Republic of Kazakhstan, the overall prevalence of congenital malformations has remained relatively stable over the past decade. However, regional disparities remain insufficiently studied. According to national statistics, the average rate is 54.1 cases of congenital malformations per 1,000 live births [4] Table 1. This figure is relatively high compared to international data and suggests the need to reassess the effectiveness of CM prevention and diagnostic strategies in the country.

Table 1.
Countries with Average Levels of Congenital Malformation Rates.

Country	Congenital Malformation Rate per 1.000 Live Births	Average Annual Number of Children Born Without Malformations (in thousands)	Congenital Heart Defect Rate per 1.000 Live Births
Kazakhstan	54.1	14.2	7.9
China	51.2	964	7.9
Brazil	57.2	192.3	7.9
Argentina	52.5	38	7.9
Mexico	52.9	121.5	7.9
Ukraine	54.5	21.8	7.9

Each year, approximately 14,200 children in Kazakhstan are born without congenital malformations. These figures reflect the complexity of the country's environmental conditions and indicate that preventive measures remain underdeveloped.

The Turkistan region is one of Kazakhstan's most distinct areas in terms of socio-demographic and environmental characteristics [5]. High population density, a predominance of rural settlements, uneven medical infrastructure, and increasing anthropogenic environmental pressure may significantly contribute to the risk of congenital malformations. Additionally, factors such as reproductive behavior, low levels of preconception preparedness, and insufficient intake of folic acid and other essential micronutrients also pose considerable risks.

This scientific study aims to assess the epidemiological characteristics of congenital malformations (CMs) recorded in the Turkistan region between 2020 and 2024. The relevance of this work stems from the need to develop effective programs and interventions for the prevention and early diagnosis of CMs at the regional level, based on evidence-based data.

2. Literature Review

The limited availability of epidemiological data on congenital malformations (CMs) in Kazakhstan, particularly at the regional level, highlights the importance of further research in this field.

In our previously published study [6], we analyzed retrospective data from the No. 3 Regional Perinatal Center in Turkistan city for the period 2020–2022. The incidence of congenital malformations among live births was found to be 0.84%, and over these three years, this rate increased by 1.8 times (47.1%). This regional growth trend is concerning and underscores the need for a comprehensive evaluation of region-specific risk factors.

A similar dynamic has been observed in studies conducted in the Zhambyl region. Kemelbekov [7] and colleagues reported that, between 2014 and 2015, 176 cases of congenital heart defects (CHDs) were recorded among 15,443 live births [7]. The incidence of CHDs was 1.09% in 2014 and 1.26% in 2015. The most common types included ventricular septal defect (42.6%), patent ductus arteriosus (21%), transposition of the great arteries, and pulmonary valve atresia (each accounting for 5.7%). These findings confirm the dominant role of heart defects in the structure of CMs and their direct impact on neonatal mortality and disability.

A study conducted by Kozhakhmetova [8] emphasized that congenital malformations remain a critical issue in global public health [8].

The authors argue that congenital malformations are among the leading causes of perinatal mortality and that these conditions impose not only clinical but also significant economic and social burdens. The article demonstrates that both the frequency and severity of congenital anomalies in modern society are not declining but rather showing a steady increase. This trend reflects the multifactorial nature of congenital disorders, including hereditary predisposition, the growing prevalence of immune and metabolic disturbances, and the complex interaction between maternal health, lifestyle, and environmental factors. According to the DOHaD (Developmental Origins of Health and Disease) concept, environmental influences and maternal lifestyle during fetal development are closely linked to the child's future health outcomes and disease risks, Kenzhebayeva [9].

Kenzhebayeva [9] and colleagues conducted a study between 2014 and 2016 analyzing 213 medical records of newborns from perinatal centers in the Karaganda region and the cities of Petropavl and Balkhash to identify key risk factors associated with congenital heart defects (CHDs) [10]. The findings of this study indicated that both prenatal and preconception risk factors play a significant role in the development of CHDs. Specifically, maternal somatic illnesses before and during pregnancy (such as diabetes mellitus and arterial hypertension), frequent viral infections, spontaneous miscarriages, pregnancy losses, as well as the exacerbation of maternal conditions and exposure to infectious agents during gestation, all contribute to an increased risk of congenital heart anomalies.

Imangalieva [10] and colleagues studied the prevalence of congenital ear malformations (such as microtia and atresia) in the Republic of Kazakhstan. These anomalies involve the underdevelopment or complete absence of the external, middle, and sometimes inner ear structures, and are most commonly unilateral (typically on the right side), occurring 2 to 2.5 times more frequently in boys. The highest number of cases was recorded in the cities of Aktobe, Aktau, and Almaty, as well as in their surrounding regions [11]. Although these conditions are considered rare, the authors emphasize the importance of referring affected children at an early age to specialists in reconstructive surgery, audiology, and social adaptation.

Elikbayev and Tutaeva [11] examined the epidemiology of congenital malformations among preterm infants in Kazakhstan. The article addresses the issue of reducing perinatal mortality and identifying preterm newborns with congenital anomalies as a high-risk group [12]. The authors note that morbidity and mortality rates are elevated in preterm infants due to their low birth weight and degree of physiological immaturity. The study lists common conditions among this group, including neurological, somatic, respiratory, and gastrointestinal disorders, increased susceptibility to infections, and delayed psychomotor development. In Kazakhstan, the annual incidence of congenital malformations is estimated at 2,500–3,000 cases, with the most frequently affected systems being the cardiovascular, central nervous, and multiple organ systems. These malformations remain one of the leading causes of child mortality.

A. Sadykova analyzed the epidemiological situation of congenital malformations in Kazakhstan. The primary aim of the study was to assess the prevalence and regional characteristics of congenital anomalies between 2015 and 2019, as well as to identify the main causes of perinatal, neonatal, and infant mortality [13]. The research was based on data from 18 regions provided by the Republican Center for Health Development of the Ministry of Health of Kazakhstan. The study found that, by 2019, the overall prevalence of congenital anomalies had decreased by 30–40% compared to 2015.

However, anomalies of the respiratory system had increased by 14%. In 2015, the most frequently registered congenital malformations involved the cardiovascular system, with a rate of 18.3 cases per 10,000 live births.

Assykbaeva et al. [13] examined the issue of genetic monitoring of congenital malformations in Kazakhstan. The authors aim to enhance the prevention, early diagnosis, and treatment of congenital anomalies through the investigation of genetic causes [14]. The research revealed that the prevalence of congenital malformations varies across different regions, with the most commonly affected systems being the cardiovascular, musculoskeletal, and nervous systems. In certain cases, these pathologies were found to be associated with consanguineous marriages, environmental exposures, or folic acid deficiency.

Rakhypbekov et al. [14] conducted a study aimed at describing the prevalence of congenital anomalies and cardiovascular system malformations among children aged 0 to 14 in the Republic of Kazakhstan and the East Kazakhstan region [15]. The analysis was based on official statistical data from the Ministry of Health of Kazakhstan for the period 2007–2012. The author examined indicators corresponding to ICD-10 codes (Q00–Q99, Q20–Q28), which cover congenital malformations and heart defects. The study found that the highest rates of congenital anomalies were recorded in the Zhambyl, South Kazakhstan, North Kazakhstan, Akmola, and Mangystau regions. Although the overall incidence in East Kazakhstan was comparatively lower than in other regions, certain high-radiation areas, such as Kurchatov, Ridder, Oskemen, and Zharma, showed a higher prevalence of anomalies. This may be linked to the long-term effects of the nearby nuclear test site. Additionally, cardiovascular anomalies were more frequently reported in regions such as Akmola, Almaty, South Kazakhstan, Pavlodar, and Mangystau. While the rate in East Kazakhstan was nearly twice as low as the national average, some specific areas – such as Kurchatov exhibited significantly elevated levels.

A. Imangaliyeva examined the issue of quality of life in children aged 2 to 7 years with microtia and external auditory canal atresia in the Republic of Kazakhstan [16]. While these conditions are not life-threatening, they have a significant impact on the child's emotional and psychological well-being, speech development, social engagement, and overall personal growth. The study found that children with microtia and atresia had higher overall quality-of-life scores in the physical functioning domain. However, parents of children who had previously undergone surgery reported lower scores across all four domains. Notably, children aged 5–7 showed significantly lower scores in emotional, social, and role functioning scales compared to those aged 2–4.

Syssoyev et al. [16] conducted a study on the epidemiological status of congenital heart defects (CHDs) in Kazakhstan over the period from 2014 to 2021 [2]. A total of 68,371 patients were analyzed. Of these, 61,285 (89.6%) had a single type of heart defect, and in 40,767 cases (59.6%), the diagnosis was made before the age of one. During the study period, 5,225 patients (7.6%) died. The authors reported that the incidence of congenital heart defects (CHDs) decreased from 64.6 to 47.3 per 100,000 males and from 68.7 to 42.5 per 100,000 females between 2014 and 2020. However, during the same period, overall mortality from all causes increased from 3.3 to 4.7 per 100,000 in males and from 2.7 to 3.7 in females.

According to survival analysis, among children diagnosed with congenital heart defects (CHDs) before the age of one, the risk of mortality was higher in males (HR 1.17), in those with multiple heart defects (HR 1.70), and in cases without surgical intervention (HR 0.57). For those diagnosed after the age of one, mortality risk was also associated with male sex (HR 1.65), presence of multiple defect types (HR 1.55), and absence of surgical treatment (HR 1.82).

One of the large-scale studies on the causes of congenital malformations was conducted by Källén [17]. The study analyzed a total of 289,365 birth cases, among which 7,020 (2.4%) fetuses or newborns were diagnosed with one or more congenital anomalies. According to the results, the etiology was identified in 26.6% of the cases. The identified causes included Mendelian genetic disorders (e.g., postaxial polydactyly type B), chromosomal abnormalities, vascular defects, complications related to monozygotic twinning, and harmful environmental exposures (both ecological and pharmaceutical factors). However, the majority of cases (approximately 73.4%) were classified as having unknown etiology.

Corsello and Giuffrè [18] estimate the global birth prevalence of congenital anomalies to be around 2–3%, and emphasize that these conditions present a complex and resource-intensive challenge for modern public health systems [18]. The authors highlight the role of genetic, environmental, and multifactorial influences in the development of congenital anomalies. Furthermore, advancements in the clinical, etiological, and pathogenetic classification systems of congenital malformations have significantly contributed to improved diagnostics and early interventions. As opportunities for genetic counseling and prenatal diagnosis expand, the early detection of these pathologies allows for timely and effective treatment and rehabilitation measures.

Retrospective epidemiological studies conducted in Kazakhstan over the past decade have shown a steady increase in the prevalence of congenital heart defects (CHDs). According to a comprehensive analysis by Sermanizova et al. [19] the incidence of CHDs among children under one year of age increased from 4.4 to 8.9 per 1,000 live births between 2003 and 2012. Among children under five, this figure rose from 2.7 to 6.3 per 1,000 [19]. Regionally, South Kazakhstan which includes the Turkistan region is classified as a high-risk area for congenital heart defects. The rates in this region are comparable to those in the northern parts of the country, likely due to a combination of ecological, social, and healthcare accessibility factors.

Among congenital malformations diagnosed during the neonatal period in Kazakhstan, anorectal malformations are among the most frequently encountered and are often associated with other organ system defects. A clinical study conducted by Kaskarbayev et al. [20] demonstrated the effectiveness of early diagnosis and immediate surgical treatment at a high-level perinatal center [20]. In the presented clinical case, rectal agenesis was identified alongside a congenital anomaly of the urinary system, and complex therapy was provided at a level III perinatal center in Karaganda. Under the newly implemented organizational model, the newborn received urgent surgical care without the need for transfer, along with comprehensive neonatal support, including respiratory and enteral care, antibiotic therapy, and breastfeeding. This

approach proved effective in preventing life-threatening complications.

One of the significant studies in the field of Kazakhstani neonatology was conducted by Tusupkaliyev et al. [21] and colleagues in 2015. The research focused on a comprehensive assessment of the impact of intrauterine infections and maternal age on the development of congenital malformations (CMs) [21]. The study was based on an analysis of the medical histories of 60 deceased newborns. The authors identified cardiovascular system defects as the most commonly occurring CMs. Specifically:

Ventricular septal defect (VSD) – 40.6%

Atrial septal defect (ASD) – 12.5%

Complex anomalies such as aortic stenosis and transposition, though rare were noted to be particularly severe.

A key finding of the study was the high frequency of cytomegalovirus (CMV) and mixed infections among children with congenital heart defects. CMV-specific antibodies were detected in 22 newborns (56.4%), and mixed infections involving CMV and herpes virus were observed in 16 cases (41%). In all cases, PCR (polymerase chain reaction) results were positive, confirming the infectious etiology.

Kemelbekov [22] conducted an analysis of the prevalence and surgical treatment indicators for congenital cardiovascular anomalies among children in the Turkistan region [22]. The study was based on medical records of 201 children aged 0 to 18 years who were treated at the cardiothoracic surgery unit of the Regional Children's Clinical Hospital between 2019 and 2020. The average age was 8.0 ± 2 years, with boys accounting for 63% of the cases. The most frequently diagnosed defects were ventricular septal defect (VSD – 11.4%), patent ductus arteriosus (PDA – 16.9%), atrial septal defect (ASD – 2.5%), and double membranous septal defect (DMPP-2 – 29.8%). The authors noted that congenital heart defects often go undetected during the neonatal period due to the lack of distinctive clinical symptoms.

Igissinov et al. [23] studied the prevalence of congenital cardiovascular anomalies (CCAs) in the Republic of Kazakhstan. During the study period, a total of 71,102 CCA cases were registered nationwide. The average annual prevalence rate was 50.9 ± 3.2 cases per 100,000 population [23]. The overall trend in the age-adjusted CCA rate across the country was positive, indicating a general increase (T = +6.3%). However, regional differences were observed: while a decline was recorded in Almaty city (T = –2.1%) and Kyzylorda region (T = –4.6%), a marked upward trend was noted in South Kazakhstan (T = +16.8%), Akmola (T = +16.9%), and Pavlodar (T = +21.9%) regions.

In our previously published study Mirzarakhimova [24], we introduced the concept of a “sporadic congenital anomaly,” referring to defects of unknown origin that occur randomly and are unlikely to recur in future pregnancies. The study found that 20–25% of congenital malformations develop due to multifactorial causes – specifically, the interaction between genetic alterations and environmental exposures – while in 10–13% of cases, the anomalies were directly attributed to environmental factors. Furthermore, clearly defined genetic causes were identified in only 12–25% of cases. This study underscored the need for deeper investigation into the etiology of congenital malformations.

Abdrakhmanov et al. [25] investigated the prevalence and structural characteristics of congenital heart defects (CHDs) among children in the Aktobe region [25]. Their study found that out of 192,273 children, 848 were under dispensary observation with a congenital heart disease (CHD) diagnosis. This corresponds to 4.4 cases per 1,000 live births in the region. The structure of diagnosed heart defects was dominated by the so-called "big six": ventricular septal defect (36.6%), atrial septal defect (22.7%), patent ductus arteriosus (13.9%), pulmonary artery stenosis (15.2%), Tetralogy of Fallot (3%), and aortic coarctation (1.3%). In 53% of the 152 newly identified CHD cases, the diagnosis was made after the age of one, indicating a low rate of early detection in the region. Additionally, some districts reported CHD prevalence rates below the regional average, suggesting possible underdiagnosis.

In the study by Oliván-Gonzalvo and Gracia-Balaguer [26] congenital heart defects (CHDs) were diagnosed in 106 children (8.6%) [26]. This rate was 14.8 times higher than the average prevalence of 0.58% among live births reported in the EUROCAT study. The most frequently observed heart anomalies were ventricular septal defect (VSD, 3.65%) and atrial septal defect (ASD, 2.92%).

In a study conducted by Aldasheva [27] the primary risk factors contributing to the development of congenital malformations (CMs) were thoroughly analyzed [27]. The study included a total of 848 pregnant women and their newborns. Women who gave birth to infants with congenital malformations (CMs) formed the main study group, while the control group consisted of 252 women and their infants without diagnosed anomalies. The results showed that certain socio-biological characteristics of the mother had a direct influence on the development of congenital malformations. Notably, increased maternal age, complicated obstetric history (e.g., miscarriage, stillbirth, infertility, early pregnancy loss), infectious diseases during pregnancy, chronic conditions, toxicosis, hypoxia, and pregnancy complications were all associated with a higher risk of CMs.

The conducted literature review made it possible to identify the multifactorial etiology, regional distribution patterns, and key risk factors associated with congenital malformations (CMs) [28]. In regions such as Turkistan, which are characterized by distinct social, ecological, and demographic conditions, notable variations in the prevalence and structure of congenital malformations (CMs) are observed. Studies have shown that cardiovascular system defects hold a leading position in the structure of congenital anomalies and have a direct impact on neonatal mortality and disability. Additionally, maternal somatic and infectious diseases, environmental adversity, folic acid deficiency, and social factors related to reproductive health significantly increase the risk of developing CMs [29]. Due to the limited epidemiological data on CMs in Kazakhstan, it is essential to strengthen regional epidemiological surveillance systems, implement genetic monitoring, and ensure early identification of high-risk groups.

3. Research Methodology

3.1. Research Foundation

This study utilized data from all recorded pregnancy cases in regional perinatal centers and medical institutions in the Turkistan region between 2020 and 2024. Specifically, the analysis included 53,169 pregnancy cases, among which 848 cases of congenital malformations (CMs) were identified. Primary information was collected from regional maternity hospitals, obstetric departments of general hospitals, outpatient clinics, and statistical reports of the regional health department. Additionally, data sources included antenatal medical records of pregnant women, birth histories from maternity hospitals, and medical records of newborns.

3.2. Research Methods

A retrospective analysis method was employed, and the data were processed using standard epidemiological approaches. Statistical processing included calculations of relative frequency, growth rate, and absolute and relative increases. The chi-square (χ^2) test was used to assess statistically significant differences in the annual dynamics of congenital malformations (CMs). A linear regression model was applied to forecast the number of CMs for the coming years. Using the regression equation, approximate CM figures for 2025 and 2026 were projected. Through graphical analysis and interpretation of statistical data, the current epidemiological situation and developmental trends of CMs in the region were identified.

4. Research Findings

Within the scope of the study, all pregnancy cases registered between 2020 and 2024 at the No. 3 Regional Perinatal Center in the Turkistan Region were analyzed from an epidemiological perspective. Primary data were collected from regional perinatal centers, obstetric departments of hospitals, and outpatient and polyclinic institutions. Data sources included antenatal medical records of pregnant women, birth histories from maternity hospitals, medical records of newborns, and regional statistical reports.

Over the five-year period, a total of 53,169 pregnancy cases were registered across the region. During this time, 848 cases of congenital malformations (CMs) were identified, accounting for 1.6% of all pregnancies. Year-by-year analysis revealed a gradual upward trend in the incidence of CMs Figure 2:

In 2020, 79 cases of CMs were recorded out of 9,916 pregnancies (0.8%).

In 2021, 108 cases (0.9%), showed a slight increase.

In 2022, 184 cases (1.7%), marking a significant rise.

In 2023, 220 cases (2.0%), nearing the highest observed level.

In 2024, 257 cases (2.4%), represented the highest figure over the five-year span.

A chi-square (χ^2) test comparing the number of congenital malformations and total pregnancy cases for 2020 and 2024 was conducted to determine whether the differences between these indicators were statistically significant. The table presents the observed and expected values, along with the proportions calculated using Pearson's χ^2 formula Table 2.
Table 2 Results of the Chi-Square (χ^2) Analysis.

Table 2.
Chi-Square (χ^2) Analysis of Congenital Malformation Cases in 2020 and 2024.

	2020	**2024**	**Marginal row totals**
Congenital malformations	79 (162.8) [43,14]	257 (173.2) [40.55]	336
Total pregnancies	9916 (9832.2) [0.71]	10376 (10459.8) [0.67]	20292
Marginal column totals	9995	10633	20628 (Grand total)

In 2020, the observed number of CMs was 79, with an expected value of 162.8, contributing [43.14] to the chi-square statistic.

In 2024, the observed number of CMs was 257, with an expected value of 173.2, contributing [40.55].

The contribution from the total number of pregnancies in 2020 was [0.71], and in 2024 was [0.67].

The total Pearson chi-square (χ^2) statistic amounted to 85.07 (43.14 + 40.55 + 0.71 + 0.67).

This value is considered very high for 1 degree of freedom (df = 1) and indicates a statistically significant difference at the $p < 0.0001$ level.

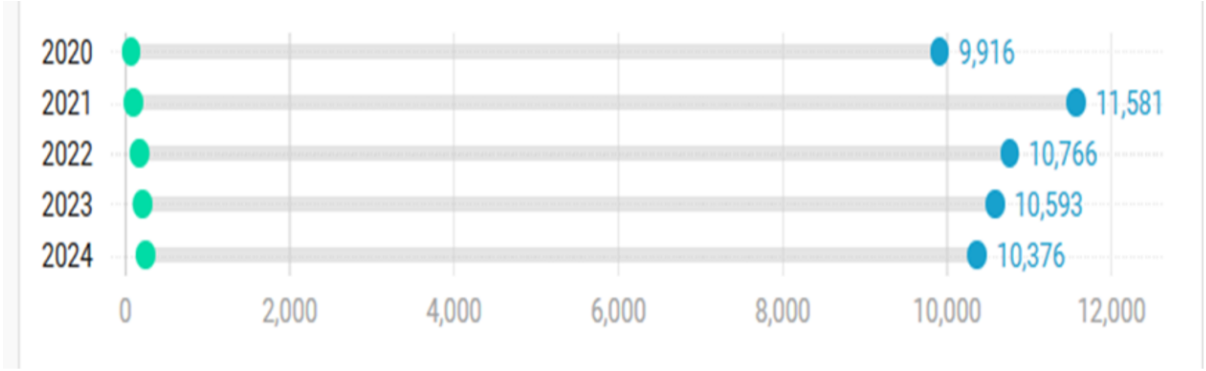

Figure 2.
Prevalence of Congenital Malformations (CMs) from 2020 to 2024

These data indicate a steady upward trend in the prevalence of CMs and reflect a worsening epidemiological situation in the region. Additionally, the increase in identified CM cases may also be attributed to improvements in diagnostic accuracy and the expansion of prenatal screening during pregnancy.

A consistent upward trend was observed in the prevalence dynamics of congenital malformations (CMs) in the Turkistan Region between 2020 and 2024 Table 3. Over the five-year period, the incidence of CMs increased by a factor of 3.2 from 79 cases in 2020 to 257 cases in 2024. To assess the nature of this growth, several epidemiological indicators were calculated: absolute increase, growth rate, and relative increase compared to 2020.

Table 3.
Dynamics of Congenital Malformation Prevalence in Turkistan Region (2020–2024).

Year	Absolute Number (CMs)	Absolute Increase	Growth Rate (%)	Growth Rate (Compared to 2020. %)	Growth Level (%)	Growth Level (Compared to 2020. %)	Absolute Value of 1%
2020	79	-	-	100.0	-	0.0	-
2021	108	29	136.7	136.7	36.7	36.7	0.79
2022	184	76	170.4	232.9	70.4	132.9	1.08
2023	220	36	119.6	278.5	19.6	178.5	1.83
2024	257	37	117.0	325.3	17.0	223.5	0.61
Total	848	-	-	-	-	-	-

An analysis of annual indicators revealed that the prevalence of congenital malformations (CMs) increased steadily, not only in absolute numbers but also across key epidemiological metrics. Notably:

The highest absolute increase was recorded in 2022 (+76 cases);

In 2024, the relative increase compared to 2020 reached 325.3%, marking the highest five-year growth rate;

The consistent growth rate indicates not only a deterioration in the region's epidemiological situation but also indirectly reflects improvements in diagnostic accuracy and case registration.

To further analyze annual changes, the absolute value of 1% and the average dynamic rate were calculated.

According to the average dynamic rate, from 2020 to 2024, the annual CM growth rate amounted to 169.6%. This figure demonstrates a rapid increase in the prevalence of congenital malformations in the region and suggests that the trend is likely to continue in the coming years Figure 3.

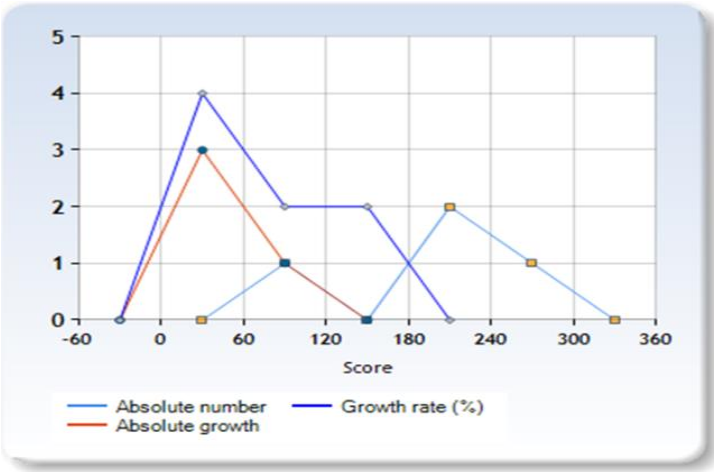

Figure 3.
CM Indicators from 2021 to 2024.

The formula for calculating the absolute value of 1% is as follows:
Absolute value of 1% = (Absolute increase / Growth level) × 100%
Let's perform the calculations for each year:
2021:
Absolute increase: 29
Growth level: 36.7%
Absolute value of 1% = (29 / 36.7) × 100% ≈ 0.79.

$$\left(\frac{29}{36.7}\right)x100\% = 0.79\%$$

2022:
Absolute increase: 76
Growth level: 70.4%
Absolute value of 1% = (76 / 70.4) × 100% ≈ 1.08.

$$\left(\frac{76}{70.4}\right)x100\% = 1.08\%$$

2023:
Absolute increase: 36
Growth level: 19.6%
Absolute value of 1% = (36 / 19.6) × 100% ≈ 1.83.

$$\left(\frac{36}{19.6}\right)x100\% = 1.83\%$$

2024:
Absolute increase: 37
Growth level: 17.0%
Absolute value of 1% = (37 / 17.0) × 100% ≈ 2.18.

$$\left(\frac{37}{17.0}\right)x100\% = 2.18\%$$

The absolute value of 1% CM growth is a metric that expresses the annual growth rate in concrete numerical terms. It allows for a quantitative assessment of the relative increase each year and helps clarify dynamic changes over time.

These indicators show that the growth rate of congenital malformations (CMs) peaked in 2023. Although growth slowed in 2024, it remained at a high level. This surge was especially evident between 2022 and 2023, when a sharp increase in CM prevalence occurred.

The absolute value of 1% serves as a crucial metric for evaluating the pace of CM spread. It reflects the effectiveness of the healthcare system and perinatal monitoring in specific years, identifies periods of epidemiological deterioration, and highlights when enhanced control measures are most needed.

Based on the data presented above, it was determined that the dynamics of congenital malformation (CM) prevalence in the Turkistan Region between 2020 and 2024 are characterized by a clear upward trend. The underlying causes of this increase are likely multifactorial, including improved prenatal diagnostics, greater public engagement with healthcare services, and the potential impact of harmful environmental factors Table 4.

Table 4 Dynamics of the prevalence of congenital malformations (CMF) in the Turkestan region from 2020 to 2024.

Table 4.
Dynamics of Congenital Malformation Prevalence in Turkistan Region (2020–2024).

Mean	308
Median	310
Standard deviation	50.02999
Skewness	-0.1796
Kurtosis	NaN
Lowest score	257
Highest score	357
Distribution range	100
Total number of scores	3
Number of distinct scores	3
Lowest class value	250
Highest class value	369
Number of classes	3
Class range	40

Based on the statistical indicators presented in the table and the results of the regression modeling, it is evident that the number of congenital malformation (CM) cases in the Turkistan Region steadily increased between 2020 and 2024. In 2024, a total of 257 CM cases were recorded, one of the highest figures within the observed period.

Descriptive statistics:

Mean – 308 (including projected values), indicating a rising trend.

Median – 310, suggesting a balanced distribution of expected future cases.

Standard deviation – 50.03, meaning the number of CM cases may vary by up to 50 cases from year to year.

Minimum value – 257; Maximum – 357, reflecting a fairly wide predicted range.

Skewness – –0.1796, indicating a slightly left-skewed distribution, meaning lower values occurred a bit more frequently.

However, to forecast how this trend may evolve in the coming years, a regression modeling method was applied. The linear regression equation obtained is:

$y = 46.80x – 94.460.00$

where:

x is the year,

y is the projected number of CM cases for that year.

According to this equation:

In 2025, the projected number of CM cases is 310

In 2026, the projection rises to 357 Figure 4.

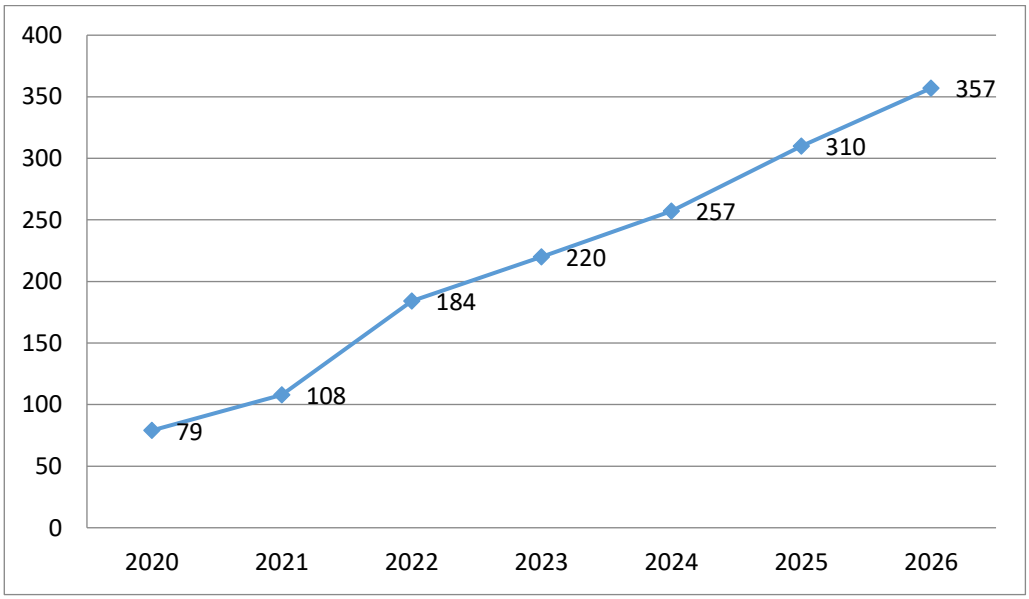

Figure 4.
Regression Model of CM Cases in Turkistan Region (2020–2026).

These results indicate that the number of congenital malformation (CM) cases in the Turkistan Region is increasing by approximately 46–47 new cases each year. If this trend continues, the regional healthcare system must be prepared to

handle this growing burden in the coming years.

The predictive model should be regarded as a vital tool for planning the management and prevention of CM cases. The data emphasize the need for the timely implementation of integrated measures, such as reallocating medical resources, expanding screening programs, and improving environmental health.

A comprehensive analysis of the structural characteristics of congenital malformations (CMs) recorded in the Turkistan Region from 2020 to 2024 revealed not only a consistent annual increase in their prevalence but also significant changes in their systemic distribution.

From a structural standpoint, the distribution of CMs across different organ systems evolved as follows Figure 5:

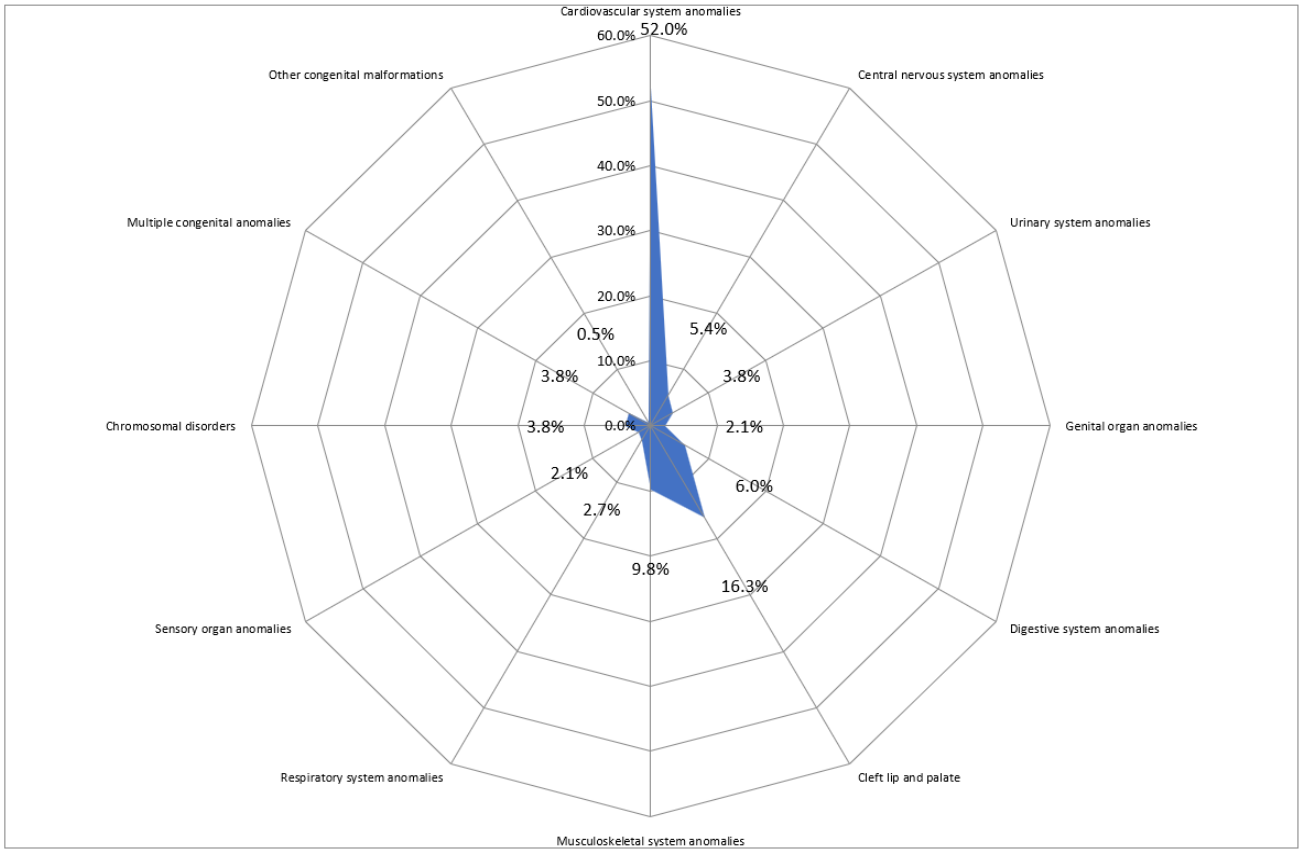

Figure 5.
Types of Congenital Malformations in 2024.

Cardiovascular system anomalies (ICD codes Q20–Q28) emerged as the most frequently occurring type of pathology. Their share increased significantly from 22.2% in 2020 to 52% in 2024. This upward trend is likely linked to improvements in medical screening systems, widespread use of echocardiography equipment, and enhanced specialist training.

Although gastrointestinal anomalies increased to 15% in 2023, their share sharply declined to 6% in 2024. This fluctuation may be explained by changes in maternal nutrition, the quality of medical care provided during pregnancy, and inconsistencies in diagnostic criteria.

Although musculoskeletal system anomalies had shown a steady decline over the past five years, in 2024 their share suddenly increased, reaching 9.8% (compared to 6.3% in 2020 and 4% in 2023). This surge may be associated with vitamin deficiencies, folic acid deficiency, maternal somatic health issues, and environmental exposure.

In general, when analyzing the structure of congenital anomalies by organ systems:

Cardiovascular system anomalies (Q20–Q28) were the most prevalent, accounting for 35.2%.

Digestive system anomalies made up 13.6%.

Cleft lip and palate accounted for 13%.

Chromosomal disorders represented 7.6%.

Musculoskeletal system anomalies comprised 6.1%.

Urinary system, respiratory system, and central nervous system anomalies had relatively lower proportions.

Other congenital anomalies (Q80–Q89) accounted for only 0.6%.

Table 5.
Distribution of Congenital Malformations by Organ Systems in Turkistan Region (2020–2024).

Year	Cardiovascular System Disorders	Central Nervous System Disorders	Respiratory System Disorders	Musculoskeletal System Disorders	Combined Congenital Anomalies (CCAs)	Row Totals
2020	22 (27.16) [0.98]	6 (4.47) [0.52]	4 (2.75) [0.57]	6 (5.16) [0.14]	6 (4.47) [0.52]	44
2021	27 (28.39) [0.07]	5 (4.67) [0.02]	3 (2.88) [0.01]	6 (5.39) [0.07]	5 (4.67) [0.02]	46
2022	27 (26.54) [0.01]	5 (4.37) [0.09]	2 (2.69) [0.04]	5 (5.04) [0.00]	3 (3.47) [0.43]	43
2023	30 (32.09) [0.14]	5 (5.28) [0.01]	4 (3.25) [0.17]	6 (6.09) [0.72]	9 (5.28) [2.62]	52
2024	52 (43.82) [1.53]	5 (7.21) [0.68]	2 (4.44) [1.34]	9 (8.32) [0.06]	3 (7.21) [2.46]	71
Column Totals	158	26	16	30	26	256 (Grand Total)

Among congenital anomalies (CA), the frequency of circulatory system defects and multiple anomalies has increased significantly from a statistical standpoint. The most notable changes were observed in 2024, with high χ^2 (chi-square) values. These findings indicate substantial shifts in the structure of congenital anomalies in the Turkistan Region. In particular, the proportion of cardiovascular defects has increased, while some other system anomalies have remained stable or declined. To interpret these changes, it is important to consider several factors simultaneously: advances in diagnostic technologies, improvements in healthcare quality, socioeconomic conditions, environmental influences, and the population's health literacy.

5. Discussion and Conclusion

The results of the study indicate that the incidence of congenital anomalies (CAs) in the Turkistan Region showed a steady increase between 2020 and 2024. In 2020, 79 cases were recorded, while by 2024, this number had risen to 257 cases, representing a 3.2-fold increase. The average annual growth was 46.8 cases.

This trend may reflect, on the one hand, improvements in the quality of medical surveillance, as advancements in diagnostic methods and the accuracy of data reporting have progressed. On the other hand, worsening environmental and socioeconomic factors may also have contributed to this upward trend.

The highest increase was observed in 2022, while 2023–2024 showed a relative stabilization trend. This phenomenon may be attributed to the impact of preventive measures, although it still requires a sustained and comprehensive approach. For example, the fact that the incidence of CAs reached 2.4% in 2024 indicates that this is not merely a medical issue but rather a strategic priority in public health.

Overall, the dynamics of CAs in the region follow a similar trend to other regions across the country. Previous studies have also linked the rise in congenital heart defects (CHDs) and other anomalies to environmental pollution, socioeconomic vulnerability, and the impact of infectious diseases on pregnant women. Similar contributing factors may be actively influencing the situation in the Turkistan Region as well.

According to the results of the regression modeling, if the current trend continues, the number of congenital anomalies (CAs) in the Turkistan Region may reach 310 cases in 2025 and 357 cases in 2026. These projections underscore the urgent need to systematically strengthen preventive measures. Specifically, this includes:

- Expanding access to prenatal screening programs;
- Improving the system of genetic counseling;
- Enhancing environmental monitoring and strengthening the local healthcare infrastructure;
- Implementing comprehensive maternal health prevention programs.

Additionally, the increase in CA cases is closely linked to socioeconomic status, education levels, and accessibility of medical care. These indicators should become a key focus of future scientific studies.

6. Recommendations

The results of this study demonstrate a consistent increase in the incidence of congenital anomalies (CAs) in the Turkistan Region between 2020 and 2024. Over five years, the total number of CA cases reached 848, accounting for 1.6% of all registered pregnancies. Compared to 79 cases in 2020, 257 cases were recorded in 2024, indicating a 3.2-fold increase in frequency. This rise may reflect the growing impact of environmental and socio-economic factors in the region, as well as the expansion of diagnostic capabilities.

Linear regression projections for the coming years also suggest a continued upward trend in CA incidence. This highlights the urgent need to strengthen preventive measures, improve the quality of prenatal screening, enhance environmental conditions, and intensify medical monitoring of pregnant women.

The findings not only describe the epidemiological pattern of congenital anomalies in the Turkistan Region but also emphasize the public health relevance of the issue. The data can serve as a foundation for developing effective preventive strategies and organizing region-specific healthcare interventions.

Given the steady increase in the incidence of congenital anomalies (CAs) in the Turkestan region from 2020 to 2024, the following comprehensive measures are strongly recommended:

- Expansion of prenatal screening programs. It is essential to increase the coverage of pregnant women with early-stage screening examinations to enable the timely detection of congenital anomalies.
- Development of genetic counseling services. Mandatory prenatal genetic counseling for families with a history of

birth defects should be introduced to reduce the risk of recurrence.

- Strengthening environmental monitoring. Particular attention should be paid to monitoring the quality of air, water, and soil in areas with a high incidence of congenital anomalies, with the involvement of environmental health authorities.
- Improvement of public awareness. Educational campaigns should be implemented to inform the population about the importance of folic acid intake, the risks of alcohol and tobacco use, and infections during pregnancy.
- Enhancing medical staff training. Medical professionals, especially in rural areas, should receive ongoing training in the diagnosis and management of high-risk pregnancies and congenital anomalies.
- Establishment of a regional congenital anomalies registry. Creating a centralized database will facilitate continuous tracking and analysis of CA trends and support the development of targeted public health interventions.

References

[1] World Health Organization, "Congenital disorders," World Health Organization, 2025. https://www.who.int/health-topics/congenital-anomalies#tab=tab_1

[2] M. H. Toufaily, M. N. Westgate, A. E. Lin, and L. B. Holmes, "Causes of congenital malformations," *Birth defects research*, vol. 110, no. 2, pp. 87-91, 2018. https://doi.org/10.1002/bdr2.1105

[3] European Surveillance of Congenital Anomalies (EUROCAT), "European surveillance of congenital anomalies (EUROCAT)," 2025. https://catalogues.ema.europa.eu/network/16478

[4] National Bureau of Statistics, "About the birth rate in 2024," National Bureau of Statistics, 2024. https://stat.gov.kz/news/2024-zhyly-tuu-k-rsetkishi-turaly/

[5] G. Dosymova, "Demographic problems of the population of Turkestan region," *Uluslararası Türk Dünyası Araştırmaları Dergisi,* vol. 4, pp. 77-86, 2021.

[6] A. K. Ayazbekov, "Problematic issues of prevention and diagnosis of congenital malformations," *Clinical medicine and Pharmacology*, pp. 72-75, 2023.

[7] K. Kemelbekov, "Structure of incidence of congenital diseases of the cardiovascular system in children in the Turkestan regionsouth," *International Journal of Scientific Pediatrics*, vol. 8, pp. 24-25, 2022. https://doi.org/10.56121/2181-2926-2022-8-21-22

[8] U. A. Kozhakhmetova, "Actual problems of the development of congenital defects in infants," *Repository of the Marat Ospanov West Kazakhstan Medical University*, vol. 60, pp. 23-28, 2018.

[9] K. A. Kenzhebayeva, "Factor of risk of development of broken hearts in a number of regions of the Republic of Kazakhstan," *Medicine and Ecology*, vol. 2, pp. 49-54, 2018.

[10] A. A. Imangalieva, "Participation in the development of the ukha in the Republic of Kazakhstan," *Bulletin of the Kazakh National Medical University*, vol. 1, pp. 322-324, 2020.

[11] G. M. Elikbayev and A. A. Tutaeva, "Epidemiology of ill-fated children with chronic diseases of development," *Medicine and Ecology*, vol. 1, pp. 16-23, 2016.

[12] A. Sadykova, "Epidemiology of congenital malformations in Kazakhstan," *Archives of the Balkan Medical Union*, vol. 56, pp. 298-308, 2021.

[13] L. Assykbaeva, N. Zharylkap, and A. Kaldybekova, "Genetic monitoring of congenital malformations in the republic of Kazakhstan," *World of Science and Education*, no. 15 март MH, pp. 30-34, 2025.

[14] T. K. Rakhypbekov *et al.*, "Comparative analysis of the prevalence of congenital heart defects in the east Kazakhstan region," *Shimane Journal of Medical Science*, vol. 34, no. 1, pp. 13-19, 2017.

[15] A. Imangaliyeva, "Quality life of children with congenital malformations of the ear," *Science & Healthcare*, vol. 23, pp. 88–96, 2021. https://doi.org/10.34689/SH.2021.23.6.010

[16] D. Syssoyev *et al.*, "Epidemiology of congenital heart disease in kazakhstan: Data from the unified national electronic healthcare system 2014-2021," *Journal of Clinical Medicine of Kazakhstan*, vol. 21, no. 3, pp. 49-55, 2024.

[17] B. Källén, *Congenital malformations," drugs during pregnancy: Methodological aspects*. New York: Nova Biomedical Books, 2016, pp. 37-46.

[18] G. Corsello and M. Giuffrè, "Congenital malformations," *The Journal of Maternal-Fetal & Neonatal Medicine*, vol. 25, no. sup1, pp. 25-29, 2012.

[19] G. Sermanizova, T. Seisembekov, and Z. Nakipov, "Epidemiological characteristics of congenital heart diseases in Kazakhstan," *Science and World*, pp. 31-33, 2013.

[20] V. M. Kaskarbayev, M. Z. Sarsenbek, A. Tolegen, L. G. Panibratets, Z. K. Elubayev, and S. T. Kizatova, "The results of a new organizational model introduced for the care of newborns with congenital malformations: A clinical case," *Science and World*, vol. 25, pp. 279–283, 2023. https://doi.org/10.34689/SH.2023.25.3.036

[21] B. Tusupkaliyev, A. Zhumalina, M. Zharlykasinova, and Z. S. Kuttymuratova, "Congenital malformations in newborns," *Medical Journal of Western Kazakhstan*, vol. 4, pp. 42-45, 2015.

[22] K. S. Kemelbekov, "Prevalence and mortality rates of congenital heart defects in infants in Zhambyl region," *Vestnik Kazakh National Medical University*, vol. 2, pp. 428-431, 2016.

[23] N. Igissinov, B. Duisenbayeva, and R. Rakhimzhanova, "Assessment of the Incidence of Congenital Anomalies of the Circulatory System in the Republic of Kazakhstan," *World Applied Sciences Journal*, vol. 23, no. 3, pp. 418-424, 2013.

[24] K. Mirzarakhimova, "Congenital anomalies in children revalence and risk factors," *Magazine "Medicine and Innovations"*, vol. 1, pp. 38-45, 2021.

[25] K. Abdrakhmanov, N. Kozhin, S. Dosmagambetov, S. Kim, and O. Dmytruk, "Prevalence and structure of congenital heart defects in children of the Aktobe oblast," *West Kazakhstan Medical Journal*, no. 2 (46), pp. 4-6, 2015.

[26] G. Oliván-Gonzalvo and J. Gracia-Balaguer, "Prevalence of congenital heart defects in assigned children for intercountry adoption," *European Journal of Preventive Cardiology*, vol. 29, no. 3, pp. e97-e99, 2022. http://dx.doi.org/10.1093/eurjpc/zwab022

[27] N. M. Aldasheva, "Assessment of risk factors for congenital malformations," *Pediatrics Journal named after GN Speransky*, vol. 89, pp. 43-46, 2010.

[28] I. Y. Sagatov, "Surgical treatment of the rare congenital heart malformation: quadricuspid aortic valve," *New Armenian Medical Journal*, vol. 12, pp. 5-6, 2018.
[29] M. S. Globa, "Forensic assessment of congenital vascular brain pathology in young adults," *Med Law*, vol. 36, pp. 7–9, 2017.