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ANALYSIS OF THE PREVALENCE OF CONGENITAL DEVELOPMENTAL DEFECTS IN NEWBORNS IN MANGISTAU REGION

Indira R. Seitmagambetova[1], https://orcid.org/0009-0008-8978-285X
Gulbarshyn D. Mukasheva[1], https://orcid.org/0000-0003-3490-5628
Zhanar K. Kurmangali[2], https://orcid.org/0000-0001-9380-3432
Dariya M. Shabdarbayeva[1], https://orcid.org/0000–0001–9463–1935
Zhanargyl K. Smailova[1], https://orcid.org/0000-0002-4513-4614
Saule B. Maukayeva[1], https://orcid.org/0000-0002-2679-6399
Nazym K. Kudaibergenova[1], https://orcid.org/0000-0002-6165-7677
Yernar K. Kairkhanov[1], https://orcid.org/0000-0001-7289-3272
Saule K. Kozhanova[1], http://orcid.org/0000-0003-3807-9765
Dinara A. Mukanova[1], http://orcid.org/0000-0001-5186-2346
Aisulu S. Zhunuspekova[1], https://orcid.org/0000-0002-2413-317X

[1] NCJSC «Semey Medical University», Semey, Republic of Kazakhstan;
[2] University Medical Center, Astana, Republic of Kazakhstan.

Abstract

Relevance of the study: Congenital malformations (CM) rank among the primary contributors to infant death, perinatal complications, and long-term disability in childhood. According to the World Health Organization, about 6% of babies worldwide are born each year with congenital anomalies, making it a significant issue for healthcare systems and society at large. The relevance of this study is due not only to the high frequency of CM but also to their significant impact on the life expectancy and quality of life of affected children, as well as the financial burden on healthcare systems. Studying CM in environmentally disadvantaged regions such as Mangystau Region of the Republic of Kazakhstan is especially important, where industrial and ecological factors may contribute to the growing prevalence of congenital anomalies.

Aim: To assess the prevalence and structure of congenital malformations among newborns in the Mangystau Region from 2020 to 2024.

Materials and Methods: A retrospective analysis of statistical data on CM was conducted based on reports from regional medical institutions. Trends were assessed by year, by antenatal screening stages, and by organ systems. Standard descriptive statistical methods were used. No personal data were analyzed.

Results: A rise in registered cases of CM was observed from 86 in 2020 to 214 in 2024. Analysis by antenatal screening stages revealed a marked increase in CM detection during the second and third trimesters, particularly in the second screening where 100 cases were recorded in 2024 compared to 58 in 2020. This may reflect advances in prenatal monitoring technologies and increased physician awareness of potential fetal anomalies. By organ system, the highest number of cases in 2024 was reported in the nervous system (39), cardiovascular system (41), and multiple anomalies (47 cases).

Conclusion: The findings highlight the need to strengthen preventive programs, improve the accessibility and quality of prenatal screening, and conduct further research into the impact of environmental factors on CM prevalence in the region.

Keywords: *congenital malformations, newborns, screening, prevalence, Mangystau Region.*

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Резюме

АНАЛИЗ РАСПРОСТРАНЕННОСТИ ВРОЖДЕННЫХ ПОРОКОВ РАЗВИТИЯ У НОВОРОЖДЕННЫХ В МАНГИСТАУСКОЙ ОБЛАСТИ

Индира Р. Сейтмаганбетова[1], https://orcid.org/0009-0008-8978-285X
Гүлбаршын Д. Мукашева[1], https://orcid.org/0000-0003-3490-5628
Жанар К. Курмангали[2], https://orcid.org/0000-0001-9380-3432
Дария М. Шабдарбаева[1], https://orcid.org/0000–0001–9463–1935
Жанаргуль К. Смаилова[1], http://orcid.org/0000-0002-4513-4614

Сауле Б. Маукаева[1], https://orcid.org/0000-0002-2679-6399
Назым К. Кудайбергенова[1], https://orcid.org/0000-0002-6165-7677
Ернар К. Каирханов[1], https://orcid.org/0000-0001-7289-3272
Сауле К. Кожанова[1], http://orcid.org/0000-0003-3807-9765
Динара А. Муканова[1], http://orcid.org/0000-0001-5186-2346
Айсулу С. Жунуспекова[1], https://orcid.org/0000-0002-2413-317X
[1] НАО «Медицинский университет Семей», г. Семей, Республика Казахстан;
[2] University Medical Center, г. Астана, Республика Казахстан.

Актуальность исследования: Врожденные пороки развития (ВПР) являются одной из ведущих причин младенческой смертности, перинатальной заболеваемости и инвалидности детей. По данным ВОЗ, ежегодно в мире регистрируется около 6% случаев рождения детей с врождёнными аномалиями, что составляет существенную медико-социальную проблему. Актуальность исследования обусловлена не только высокой частотой ВПР, но и значительным влиянием этих патологий на продолжительность и качество жизни детей, а также на затраты системы здравоохранения. Особое значение приобретает изучение ВПР в экологически неблагоприятных регионах, таких как Мангистауская область Республики Казахстан, где воздействие промышленных и экологических факторов может способствовать росту распространенности врождённых аномалий.

Цель: Оценить распространенность и структуру врожденных пороков развития среди новорождённых в Мангистауской области за период с 2020 по 2024 годы.

Материалы и методы: Проведен ретроспективный анализ статистических данных о ВПР, полученных из медицинских учреждений региона. Оценивалась динамика по годам, по этапам антенатального скрининга, а также по системам органов. Использовались стандартные методы описательной статистики. Персональные данные не анализировались.

Результаты: Отмечен рост зарегистрированных случаев ВПР с 86 в 2020 году до 214 в 2024 году. Анализ по этапам антенатального скрининга показал значительное увеличение выявляемости ВПР на II и III триместрах, особенно во втором скрининге, где в 2024 году зафиксировано 100 случаев против 58 в 2020 году. Это может указывать на совершенствование технологий пренатального мониторинга и повышение настороженности врачей к потенциальным аномалиям развития плода. В разрезе органных систем наибольшее количество ВПР было зарегистрировано в 2024 году в нервной системе (39 случаев), сердечно-сосудистой системе (41 случай) и в категории множественных ВПР (47 случаев).

Выводы: Результаты свидетельствуют о необходимости усиления профилактических программ, повышения доступности и качества пренатального скрининга, а также проведения дополнительных исследований влияния экологических факторов на частоту ВПР в регионе.

Ключевые слова: *врожденные пороки развития, новорождённые, скрининг, распространенность, Мангистауская область.*

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Түйіндеме

МАҢҒЫСТАУ ОБЛЫСЫНДА ЖАҢА ТУҒАН НӘРЕСТЕЛЕРДЕ ТУА БІТКЕН ДАМУ АҚАУЛАРЫНЫҢ ТАРАЛУЫН ТАЛДАУ

Индира Р. Сейтмаганбетова[1], https://orcid.org/0009-0008-8978-285X
Гүлбаршын Д. Мукашева[1], https://orcid.org/0000-0003-3490-5628
Жанар К. Курмангали[2], https://orcid.org/0000-0001-9380-3432
Дария М. Шабдарбаева[1], https://orcid.org/0000–0001–9463–1935
Жанаргуль К. Смаилова[1], http://orcid.org/0000-0002-4513-4614
Сауле Б. Маукаева[1], https://orcid.org/0000-0002-2679-6399
Назым К. Кудайбергенова[1], https://orcid.org/0000-0002-6165-7677
Ернар К. Каирханов[1], https://orcid.org/0000-0001-7289-3272
Сауле К. Кожанова[1], http://orcid.org/0000-0003-3807-9765
Динара А. Муканова[1], http://orcid.org/0000-0001-5186-2346
Айсулу С. Жунуспекова[1], https://orcid.org/0000-0002-2413-317X
[1] «Семей медицина университеті» КеАҚ, Семей қ., Қазақстан Республикасы;
[2] University Medical Center, Астана қ., Қазақстан Республикасы.

Зерттеудің өзектілігі: Туа біткен даму ақаулары (ТБА) нәрестелер арасындағы өлім-жітімнің, перинаталдық сырқаттанушылықтың және бала жастағы мүгедектіктің негізгі себептерінің бірі болып табылады. ДДСҰ деректеріне сәйкес, жыл сайын дүниежүзінде шамамен 6% нәресте туа біткен ақаулармен дүниеге келеді, бұл маңызды медициналық-әлеуметтік мәселе болып отыр. Зерттеудің өзектілігі тек ТБА жиілігінің жоғары болуына ғана емес, сонымен қатар бұл патологиялардың балалардың өмір сүру ұзақтығы мен сапасына, сондай-ақ денсаулық сақтау жүйесіне түсетін салмаққа айтарлықтай әсер етуімен де байланысты. Әсіресе, Маңғыстау облысы сияқты экологиялық жағынан қолайсыз аймақтарда ТБА-ны зерттеу өзекті, себебі өнеркәсіптік және қоршаған орта факторлары бұл патологиялардың таралуына ықпал етуі мүмкін.

Зерттеудің мақсаты: 2020–2024 жылдар аралығында Маңғыстау облысында жаңа туған нәрестелер арасындағы туа біткен даму ақауларының таралуы мен құрылымын бағалау.

Материалдар мен әдістер: Аймақтың медициналық мекемелерінен алынған ТБА-ға қатысты статистикалық деректерге ретроспективті талдау жүргізілді. Жылдар бойынша, антенаталдық скрининг кезеңдері мен ағза жүйелері бойынша динамика бағаланды. Сипаттамалық статистиканың стандартты әдістері қолданылды. Жеке деректер қарастырылған жоқ.

Нәтижелер: 2020 жылы тіркелген 86 жағдайдан 2024 жылы 214 жағдайға дейін ТБА санының өскені байқалды. Антенаталдық скрининг кезеңдеріне жасалған талдау екінші және үшінші триместрлерде ТБА-ны анықтаудың едәуір артқанын көрсетті, әсіресе 2024 жылы екінші скринингте 100 жағдай тіркелген (2020 жылы – 58 жағдай). Бұл пренаталдық бақылау технологияларының жетілдірілуіне және дәрігерлердің ұрықтың даму ақауларына деген қырағылығының артуына байланысты болуы мүмкін. Орган жүйелері бойынша 2024 жылы жүйке жүйесі (39 жағдай), жүрек-қантамыр жүйесі (41 жағдай) және көп мүшелі ТБА (47 жағдай) ең жиі тіркелген.

Қорытынды: Зерттеу нәтижелері профилактикалық бағдарламаларды күшейтудің, пренаталдық скринингтің қолжетімділігі мен сапасын арттырудың, сондай-ақ экологиялық факторлардың ТБА жиілігіне әсерін зерттеудің маңыздылығын көрсетеді.

Түйінді сөздер: *туа біткен даму ақаулары, жаңа туған нәрестелер, скрининг, таралуы, Маңғыстау облысы.*

Дәйексөз үшін:

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Introduction

Congenital malformations (CMs) are structural or functional abnormalities that develop during the prenatal period and have a significant impact on a child's health. These defects can affect various organs and systems, leading to serious developmental challenges and often requiring long-term medical intervention [2]. The causes of CMs are diverse and can be either genetic or environmental. Environmental factors include harmful exposures such as infections, poor nutrition (especially folic acid deficiency), and other external influences [10]. Genetic factors include gene mutations and chromosomal abnormalities such as Down syndrome [2,10].

The prevalence of CMs varies by region, ranging from 2% to 6% of newborns [23]. In developed countries, this rate is usually lower due to better access to high-quality prenatal care and diagnostics. In the United States, congenital anomalies are diagnosed in roughly 3% of newborns, whereas in certain Latin American countries, this figure can rise to 5% [8]. In lower-income regions, such as parts of Africa and South Asia, the prevalence is often even higher. This is mainly due to limited access to quality healthcare, poor folic acid intake, and adverse environmental conditions [14]. Congenital malformations also have serious social and economic implications, placing considerable strain on both healthcare systems and social services.

Genetic and chromosomal abnormalities are among the leading causes of congenital malformations. However, environmental factors also play a significant role, including maternal infections during pregnancy (such as rubella, toxoplasmosis, and cytomegalovirus), exposure to harmful substances like alcohol, drugs, or certain medications, and chronic health conditions in the mother, such as diabetes and hypertension [16,19].

Importantly, a lack of folic acid during pregnancy is strongly associated with an increased risk of neural tube defects in the developing fetus [13]. Some of the most commonly observed congenital anomalies include heart defects, cleft lip and palate, neural tube defects, and limb abnormalities [13,16].

In countries with advanced healthcare systems, preventing congenital anomalies remains a public health priority, as they continue to be a major cause of infant mortality and long-term disability. Modern diagnostic and treatment methods can help detect and manage many anomalies, but the mortality rate remains high in regions with less developed healthcare infrastructure [8,14]. Genetic counseling is one of the most effective preventive tools, helping identify at-risk groups and reduce the birth of children with severe abnormalities [15]. Screening programs and health promotion also play a vital role in reducing CM rates [19].

According to official statistics, CM rates vary depending on region and population health. For example, in the Moscow region, the rate is about 3% of all births, while in some parts of Russia such as Krasnodar Krai, an increase in CM cases has been reported, likely linked to worsening environmental conditions [3]. In countries like Kazakhstan,

the CM rate is 22.9 per 1,000 live births, with regional variation from 13.3 to 44.4 per 1,000 [11]. These figures confirm that environmental conditions and healthcare quality directly influence CM prevalence. In Kazakhstan, the highest rates are found in the South Kazakhstan and Pavlodar regions, associated with poor environmental conditions and greater healthcare system strain [4].

Mangystau Region is one of Kazakhstan's industrial areas and has a high rate of congenital malformations. Research shows a significant prevalence of congenital ear defects in the region, which may be linked to environmental pollution and industrial exposure. In cities like Aktau and other populated areas, one in every ten registered CMs involves malformations of the ear [5].

Congenital anomalies affecting hearing and vision are particularly common, which may be due to a combination of genetic and environmental factors [4].

Thus, the aim of this study is to assess the prevalence of congenital malformations among newborns in the Mangystau Region and to identify the most common types of CMs.

Materials and Methods

This study is based on a retrospective analysis of data on CMs in newborns from 2020 to 2024. Data Collection

The data were sourced from official statistical reports provided by medical institutions that documented cases of congenital malformations (CMs) across various organ systems, including the nervous, cardiovascular, respiratory, and others. Data collection followed a standardized protocol and was conducted at all levels of primary healthcare.

The analysis included both annual and categorical frequency data for different types of CMs. No personal information was used in the study, as all data were aggregated and anonymized.

Ethical Considerations

Since the study relied exclusively on anonymized data, informed consent was not required. Ethical approval was obtained from the Ethics Committee of Semey Medical University. The research was conducted in full compliance with the ethical principles set out in the Declaration of Helsinki.

Statistical Analysis

Descriptive and trend analyses were carried out using Microsoft Excel and SPSS software. These tools were used to calculate CM incidence rates and examine patterns over the five-year study period. The results are presented in tables and charts that show how the rates have changed over time.

Results

Figure 1 shows how the number of congenital malformation (CM) cases changed from 2020 to 2024. Overall, there's a clear upward trend, which could be due to several reasons — better diagnostic tools, wider use of screening, or shifts in the general health and environmental situation.

In 2020, there were 86 recorded cases, the lowest over the five-year period. By 2021, the number jumped to 148, possibly because more people were accessing healthcare and screening methods had improved. The increase continued in 2022, reaching 175 cases. In 2023, there was a slight dip to 152, which might reflect changes in how data were collected or reported, or temporary shifts in screening efforts. Then in 2024, the number rose sharply to 214 cases.

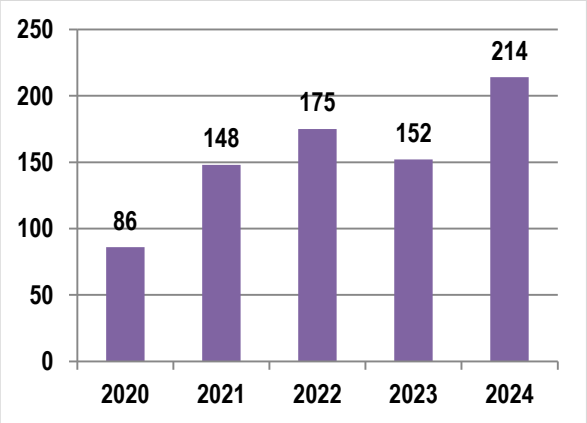

Figure 1. Dynamics of the detection of CM by year (2020–2024).

Figure 2 presents data on the number of both terminated and completed pregnancies from 2020 to 2024. Over this five-year period, the number of terminated pregnancies gradually rose from 68 cases in 2020 to 117 in 2024. At the same time, the number of continued pregnancies (resulting in a live birth) also increased, particularly in 2024, when it reached 97 cases, significantly higher than the 18 cases recorded in 2020.

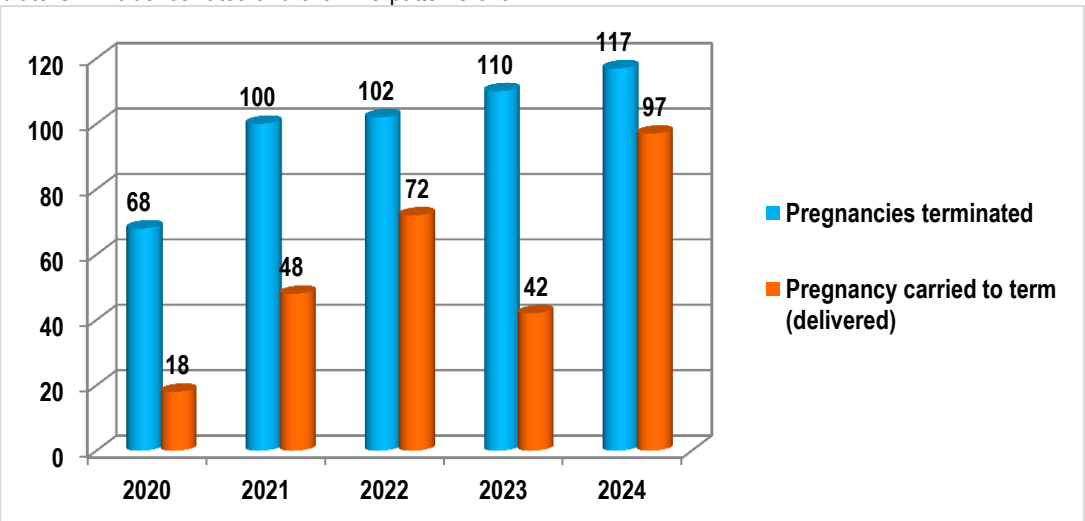

Figure 2. Dynamics of pregnancy termination and continuation by year (2020–2024).

For example, in 2021 there were 100 recorded pregnancy terminations and 48 continued pregnancies, reflecting a general rise in medical interventions during pregnancy. However, in 2023, a slight decrease in continued pregnancies was observed (42 cases), alongside an increase in terminations (110 cases).

Figure 3 illustrates the number of congenital malformations (CMs) identified at various stages of prenatal screening (first, second, and third) from 2020 to 2024. In 2020, 23 cases were detected during the first screening, 58 during the second, and 5 during the third. In 2021, detection rates increased across all stages, with a particularly sharp rise during the second screening, which identified 89 cases — a significant jump compared to the previous year.

By 2022, most cases (128) were identified during the first screening, while the second screening saw a notable decrease, detecting only 24 cases. In 2023, more cases were picked up at the third screening, 52 in total, suggesting better detection of issues later in pregnancy. Then in 2024, the number of cases found at the first screening dropped to 66, but the second screening saw a big rise to 100.

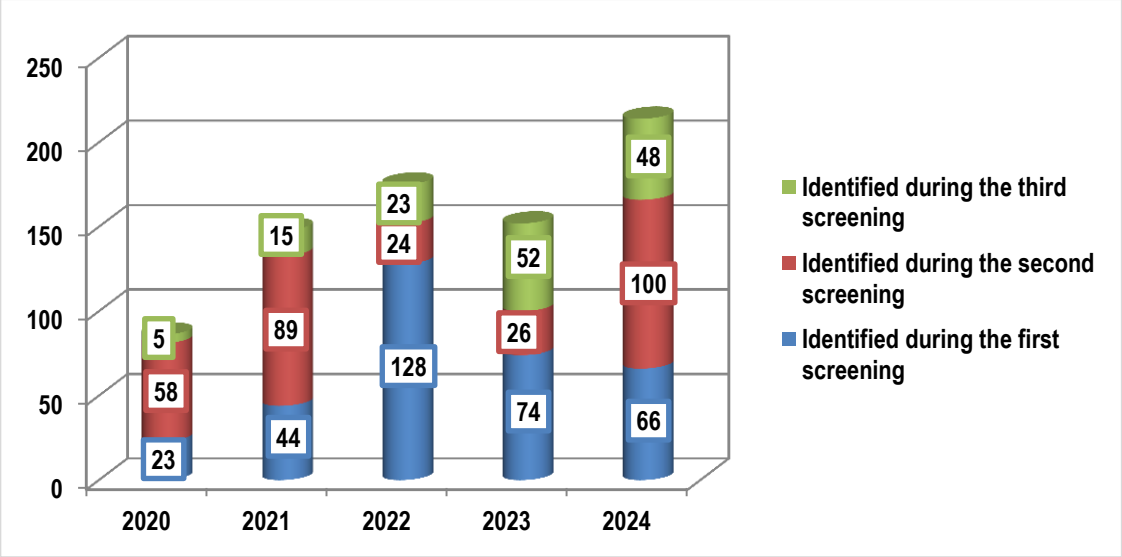

Figure 3. Dynamics of the detection of congenital malformations at different screening stages by year (2020–2024).

Table 1 presents the number of congenital malformation (CM) cases detected in newborns between 2020 and 2024, categorized by affected organ systems. Across the five-year period, nervous system anomalies remained the most frequently reported, showing minor year-to-year variations. The highest number of such cases was recorded in 2024, reaching 39, up from 28 cases in 2020. A significant increase was observed in cardiovascular defects, rising from 11 cases in 2020 to 41 in 2024. A similar trend was noted in genitourinary system anomalies, with cases increasing from 2 to 19 over the same period.

Particular attention is drawn to the rise in multiple congenital anomalies. Starting from 12 cases in 2020, the number nearly quadrupled to 47 by 2024. This may indicate both a true increase in prevalence and improvements in diagnostic accuracy. The category “Other CMs”, which includes congenital anomalies not classified under the main groups, also shows a growth trend, from 8 cases in 2020–2021 to a peak of 41 in 2023, and 34 in 2024.

Table 1.

Detection Trends of Congenital Malformations by Organ System (2020–2024).

Type of Malformation	2020	2021	2022	2023	2024
Nervous system	28	37	35	34	39
Cardiovascular system	11	15	30	27	41
Bronchopulmonary system	3	4	4	4	2
Facial structures	3	13	6	3	6
Gastrointestinal tract (GIT)	5	7	3	1	4
Genitourinary system	2	8	9	5	19
Musculoskeletal system	8	23	5	2	13
Diaphragm	3	4	0	0	1
Anterior abdominal wall	3	5	8	3	8
Multiple congenital anomalies	12	24	45	32	47
Other CMs	8	8	30	41	34

Discussion

CMs result from a combination of genetic and environmental factors. Inherited conditions, gene mutations, and chromosomal abnormalities like Down syndrome can lead to developmental issues [2,10]. Genetic predisposition is often linked to common defects such as heart malformations and cleft lip and palate.

In addition, exogenous factors such infections, environmental toxins, and exposure to teratogens during pregnancy also significantly increase the risk of CMs.

Infections such as rubella, toxoplasmosis, and cytomegalovirus can interfere with fetal development, leading to conditions like neural tube defects, heart abnormalities, and cleft lip or cleft palate [16,19]. Environmental pollution, including emissions of harmful substances into the air and contamination of water resources, has a negative impact on the health of pregnant women and fetal development. or instance, research shows that regions with higher levels of environmental pollution, such as East Kazakhstan, tend to report more cases of congenital anomalies, especially heart defects and malformations of the central nervous system [9,17].

Particular attention should be paid to cardiovascular malformations, which rank among the most prevalent types of CMs. According to statistical data, congenital heart defects are diagnosed in 0.7% to 1.7% of newborns in Kazakhstan, indicating a relatively high prevalence. These types of defects are a major contributor to infant mortality, making up about 47.5% to 47.8% of all deaths caused by congenital anomalies. This underscores the urgent need to prioritize the issue in pediatric healthcare [6]. Early diagnosis and timely treatment are crucial for reducing both infant mortality and the risk of long-term disability.

In addition, external factors such as folic acid deficiency during pregnancy — play a major role in the development of neural tube defects. Numerous studies have confirmed that insufficient folic acid significantly raises the risk of conditions like spina bifida and anencephaly [13]. This underscores the importance of maintaining a balanced diet and ensuring adequate folic acid intake during the preconception period [13].

Another key factor influencing the prevalence of congenital malformations (CMs) is access to medical care. In many low-resource settings, particularly in parts of Africa and South Asia, limited access to quality healthcare often results in late diagnoses. This delay contributes to higher rates of disability and infant mortality associated with congenital anomalies. On the other hand, countries with well-developed healthcare systems like the United States and the United Kingdom tend to have much lower rates of such outcomes, thanks to advanced diagnostic technologies and better medical care [19].

In Kazakhstan, despite ongoing efforts to improve early detection, including initiatives under the “Densaulyk” program aimed at prevention at multiple stages, significant challenges remain. One of the key components of effective prevention is medical and genetic counseling, which plays an important role in identifying at-risk groups and reducing the likelihood of severe congenital conditions. This is particularly important in countries with high CM prevalence, such as Kazakhstan, where environmental and social factors significantly influence the distribution of these conditions [19].

Studies from various regions around the world have identified several factors influencing the prevalence of congenital malformations. For example, a study by *Abdolahi H. et al.* found that in rural areas of Tabriz, nervous system anomalies were the most frequently observed congenital defects. These were largely associated with environmental exposure, particularly in regions located near industrial facilities such as petrochemical plants and thermal power stations [20].

Similarly, in the Republic of Tatarstan, research by *Ginter E.K. et al.* demonstrated significant differences in the prevalence of monogenic hereditary diseases were linked to varying levels of inbreeding across different districts, highlighting the role of genetic factors in disease distribution. These findings may also be relevant to understanding the genetic aspects of congenital anomaly prevalence in the Mangystau region, where unique genetic characteristics of the population may be present [21].

Additionally, research conducted in Gorno-Altaysk and other regions of Russia (Krikunova N.I. et al., 2004) demonstrates that both environmental and genetic predispositions can influence the prevalence of developmental defects. For instance, the high frequency of limb deformities observed in some areas of the Altai Republic may be related to environmental conditions similar to those in the ecologically disadvantaged regions of Mangystau [7].

In Kazakhstan, a similar study (*Sadykova A. et al.,* 2021) shows that despite an overall decline in the incidence of congenital anomalies in recent years, high rates of morbidity persist in southern regions, including areas with increased environmental risk. Studies conducted in the Pre-Aral region (*Nadeev* A.P. *et al.,* 2018) also confirm the influence of environmental factors on newborn health, with a higher frequency of congenital malformations observed in areas with polluted environments [12, 22].

A study in the East Kazakhstan region (*Abylgazinova* A.Zh. *et al*., 2016) showed that although the incidence of congenital anomalies decreased after the closure of the Semipalatinsk nuclear testing site, higher morbidity rates remained in areas close to the former testing zones. This highlights the urgent need for more in-depth research into the long-term ecological impacts of nuclear contamination and its potential effects on newborn health in affected areas [1].

Limitations:

This study has some limitations. Data quality may differ between regions due to varying diagnostic and reporting standards. It also didn’t explore genetic and environmental factors in depth, limiting our understanding of their separate roles. Being based on retrospective data, the study may include gaps or biases. Finally, the results might not apply to regions with different socio-economic or environmental backgrounds. More comprehensive, standardized research is needed.

Conclusions:

The findings stress the need for a broad, integrated approach to understanding congenital malformations. In areas like Mangystau, both environmental and genetic factors, along with healthcare access and diagnostic capacity, must be considered. Addressing these conditions effectively requires early detection, better healthcare access, and reducing environmental risks.

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Ethical Standards

Ethical approval was obtained from the Ethics Committee of Semey Medical University. The research was conducted in full compliance with the ethical principles set out in the Declaration of Helsinki.

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Information about the authors:

Seitmagambetova Indira – master of Public Health, NCJSC «Semey Medical University», Semey, Kazakhstan, phone+7 701 555 2566, email indira.s1901@gmail.com, https://orcid.org/0009-0008-8978-285X

Mukasheva Gulbarshyn Darynkyzy - Senior Teacher of the Department of Epidemiology and Biostatistics, NCJSC «Semey Medical University», Semey, Kazakhstan; E-mail: gulbarshyn_1_12@mail.ru; phone +7 775 220 07 45, https://orcid.org/0000- 0003-3490-5628

Kurmangali Zhanar– associate professor of Department of Internal Medicine of University Medical Center, Astana, Republic of Kazakhstan, email zhanar.k.kurmangali@gmail.com, https://orcid.org/0000-0001-9380-3432

Shabdarbayeva Dariya Muratovna – Doctor of Medical Sciences, Professor, Vice Rector for Science, NCJSC «Semey Medical University», Semey, Kazakhstan, phone 8 707 365 82 71, e-mail: dariya_kz@bk.ru, https://orcid.org/0000-0001-9463-1935

Smailova Zhanargyl Kaiyrgaliyevna - Candidate of Medical Sciences, Associate Professor, Vice Rector for Academic and Educational Work, NCJSC «Semey Medical University», phone 8 707 365 82 71, e-mail: zhanargul.smailova@smu.edu.kz; https://orcid.org/0000-0002-4513-4614

Maukayeva Saule Boranbayevna - Candidate of Medical Sciences, Professor of the Department of Infectious Diseases, Dermatovenerology and Immunology, NCJSC «Semey Medical University», Semey, Kazakhstan; phone: 8 705 529 66 75, e-mail: solly66@mail.ru, https://orcid.org/0000-0002-2679-6399

Kudaibergenova Nazym Konyrovna - Candidate of Medical Sciences, Associate Professor of the Department of Infectious Diseases, Dermatovenerology and Immunology, NCJSC «Semey Medical University», Semey, Kazakhstan, phone: 8 705 188 0836, e-mail: nazym.kudaibergenova@smu.edu.kz, https://orcid.org/0000-0002-2679-6399

Kairkhanov Yernar Karimhanovich - Doctor of Medical Sciences, Professor, Director of Pavlodar branch, NCJSC «Semey Medical University», Pavlodar, Kazakhstan; E-mail: Kairkhanov67@mail.ru; phone +7 701 458 72 18, https://orcid.org/0000-0001-7289-3272

Kozhanova Saule Keneskhanovna - Candidate of Medical Sciences, Associate Professor, Head of the Department of Anatomy, Histology and Topographic Anatomy named after Doctor of Medical Sciences, prof. N.A. Khlopov, NCJSC «Semey Medical University», Semey, Kazakhstan, phone: 8 707 721 55 68, e-mail: saule.kozhanova@smu.edu.kz, http://orcid.org/0000-0003-3807-9765

Mukanova Dinara Adletovna - Candidate of Medical Sciences, Associate Professor, Head of the Department of Simulation and Educational Technologies, NCJSC «Semey Medical University», Semey, Kazakhstan, phone: 8 701 491 98 29, e-mail: dinara.mukanova@smu.edu.kz, http://orcid.org/0000-0001-5186-2346

Zhunuspekova Aisulu Sarsengazievna - PhD, Assistant of the Department of Therapy, NCJSC «Semey Medical University», Semey, Kazakhstan, phone: 8 702 863 99 53, e-mail: aisulu.zhunuspekova@mail.ru, https://orcid.org/0000-0002-2413-317X

Corresponding Author:

Mukasheva Gulbarshyn Darynkyzy - PhD, Senior Lecturer of the Department of Epidemiology and Biostatistics, NCJSC «Semey Medical University», Semey, Republic of Kazakhstan.

Postal Address: Republic of Kazakhstan, 071400, Semey, Abay St., 103.

E-mail: gulbarshyn_1_12@mail.ru

Phone: +7 775 220 0745