Received: 11 February 2022 / Accepted: 22 June 2022 / Published online: 30 June 2022

DOI 10.34689/SH.2022.24.3.025

UDC 617.7

EPIDEMIOLOGICAL CHARACTERISTICS OF CONGENITAL CATARACT. LITERATURE REVIEW

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Abstract

Introduction: Congenital cataract is the one of the leading cause of preventable childhood blindness worldwide. According to the literature, congenital cataracts account for 10 to 19.5% among causes of low vision and blindness. Conducting epidemiological studies is the first step towards planning resource allocation of the healthcare sector and reducing the burden of avoidable blindness due to congenital cataracts.

Aim: To study the epidemiological characteristics of congenital cataracts, as well as reflect the statistical data of the Almaty branch of the Republican State Enterprise "Republican e-health center" of the Ministry of Health of the Republic of Kazakhstan on the incidence of congenital cataracts in children in Almaty and the number of children 0-17 years who are registered with the diagnosis "Cataract" in the Republic of Kazakhstan.

Search strategy: The literature search was carried out in the electronic databases MEDLINE, PubMed, EMBASE, Web of Science, Google Scholar and e-library using keywords (congenital cataract; children; prevalence; incidence). The search depth was 25 years (from 1996 to 2021). Relevant works reflecting the characteristics of the problem were accepted for the analysis in the review. Of all selected articles, 71 sources were included for the subsequent analysis, which met the inclusion criteria and excluded duplication or repetition of information.

Results:

1. The prevalence and incidence of congenital cataracts varies greatly throughout the world.
2. Many epidemiological studies indicate more frequent occurrence of this pathology among boys.
3. Bilateral congenital cataract is seen more frequently than unilateral form, the three most common morphological types are total, nuclear and posterior subcapsular.
4. According to the data of the Almaty branch of the Republican State Enterprise "Republican e-health center" of the Ministry of Health of the Republic of Kazakhstan, there is an increase in the incidence of congenital cataracts in children in Almaty, as well as an increase in the number of children 0-17 years who are registered with the diagnosis "Cataract" in the Republic of Kazakhstan.

Keywords: *congenital cataract, children, prevalence, incidence.*

Резюме

ЭПИДЕМИОЛОГИЧЕСКАЯ ХАРАКТЕРИСТИКА ВРОЖДЕННОЙ КАТАРАКТЫ. ОБЗОР ЛИТЕРАТУРЫ

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Введение: Врожденная катаракта является одной из основных причин инвалидности по зрению с детства. По данным литературына долю врожденных катаракт приходится от 10 до 19,5%среди причин слепоты и слабовидения Проведение эпидемиологических исследований является первым шагом к планированию распределения ресурсов сектора здравоохранения и уменьшению бремени предотвратимой слепоты вследствие врожденной катаракты.

Цель исследования: изучить эпидемиологические характеристики врожденной катаракты, а также отразить статистические данные Алматинского городского филиала РГП на ПХВ «Республиканский центр электронного здравоохранения» МЗ РК по заболеваемости врожденной катарактой у детей по городу Алматы и количеству детей 0-17 лет, состоящих на диспансерном учете с диагнозом «Катаракта» в Республике Казахстан.

Стратегия поиска: поиск литературы был осуществлен в электронных базах MEDLINE, PubMed, EMBASE, Web of Science, Google Scholar и e-library, по ключевым словам (врожденная катаракта; дети; распространенность;

заболеваемость). Глубина поиска составила 25 лет (с 1996 по 2021 годы). Релевантные работы, отражающие характеристики проблемы, были приняты для описания в обзоре. Из всех отобранных статей для последующего анализа было включено 71 источник, которые отвечали критериям включения и исключали дублирование или повтор информации.

Результаты и выводы:

1. Распространенность и заболеваемость врожденной катарактой значительно варьирует в мире.
2. Многие эпидемиологические исследования указывают на более частую встречаемость данной патологии среди мальчиков.
3. Чаще встречается двусторонняя форма врожденной катаракты, а среди клинико-морфологических форм: диффузная, ядерная и задняя субкапсулярная формы.
4. По данным Алматинского городского филиала РГП на ПХВ «Республиканский центр электронного здравоохранения» МЗ РК отмечается рост заболеваемости детей врожденной катарактой по городу Алматы, а также увеличение количества детей 0-17 лет, состоящих на диспансерном учете с диагнозом «Катаракта» в Республике Казахстан.

Ключевые слова:*врожденная катаракта,дети,распространенность, заболеваемость.*

Түйіндеме

ТУА БІТКЕН КАТАРАКТАНЫҢ ЭПИДЕМИОЛОГИЯЛЫҚ СИПАТТАМАСЫ. ӘДЕБИ ШОЛУ

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Кіріспе: туа біткен катаракта бала кезінен бастап көру қабілетінің бұзылуына негізгі себептерінің бірі болып табылады. Әдебиет бойынша, соқырлық және нашар көрудің себептерінің ішінде туа біткен катаракта 10-нан 19,5%-ға дейін құрайды. Эпидемиологиялық зерттеулер жүргізу денсаулық сақтау секторы үшін ресурстарды бөлуді жоспарлауға және туа біткен катаракта салдарынан болатын соқырлықты азайтуға бағыттылған алғашқы қадам болып табылады.

Мақсаты: туа біткен катарактаның эпидемиологиялық сипаттамасын зерттеу, сондай-ақ Алматы қаласының балалар арасындағы туа біткен катаракта ауыршандығы және Қазақстан Республикасында «Катаракта» диагнозымен диспансерлық есепте тұрған 0-17 жасар балаларының саны бойынша Қазақстан Республикасы Денсаулық сақтау министрлігінің «Республикалық электрондық денсаулық сақтау орталығы» ШЖҚРМК Алматы қалалық филиалының статистикалық мәліметтерін көрсету.

Іздеу стратегиясы: әдибиеттерді іздеу MEDLINE, PubMed, EMBASE, Web of Science, Google Scholar және e-library электронды базаларында түйінді сөздер (туа біткен катаракта; балалар; таралу; аурушандық) бойынша жүргізілді. Іздеу терең дігі 25 жыл (1996 жылдан 2021 жылға дейін) құрады. Біздің зерттеуіміздің мақсаттарына келетін 71 ғылыми жарияланымдар анықталды.

Нәтижелері:

1. Туа біткен катарактаның таралуы және аурушандығы әлемде әр түрлі болып табылады.
2. Көптеген эпидемиологиялық зерттеулер бұл патологияның ұлдар арасында жиі кездесетінін анықтайды.
3. Көбінесе туа біткен катарактаның екі жақты түрі, ал клиникалық-морфологиялық түрлерінен диффузды, ядролық және артқы субкапсулдық түрлері жиі кездеседі.
4. Қазақстан Республикасы Денсаулық сақтау министрлігінің «Республикалық электрондық денсаулық сақтау орталығы» ШЖҚРМК Алматы қалалық филиалының статистикалық мәліметтері бойынша Алматы қаласының балалар арасындағы туа біткен катаракта ауыршандықтың және Қазақстан Республикасында «Катаракта» диагнозымен диспансерлық есепте тұрған 0-17 жасар балаларының санның тұрақты өсуі байқалады.

Түйіндісөздер: *туа біткен катаракта; балалар; таралу; аурушандық.*

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Introduction

Congenital cataract is one of the main causes of childhood blindness worldwide[60]. According to the literature, congenital cataract accounts for10 to 19.5%among the causes of blindness and low vision [22]. The fight against congenital cataract is considered a high priority for the World Health Organization's VISION 2020 program - The Right to Sight [9]. This is due to the huge burden of blindness on the emotional, social, economic condition of the child, family and society [9]. The decrease in the quality of life in children with cataracts is comparable to the level in children with severe congenital heart defects and liver transplantation [7, 19, 63].

A.Zh. Aubakirova established that one of the main socially significant congenital pathologies leading to blindness and low vision in the Republic of Kazakhstan is congenital cataract[2].

Congenital cataract is opacity of the lens of different sizes and intensity, observed in a child at birth or at an older age [22]. In infants, the vision system develops intensively after birth and an obstacle in the form of clouding of the lens disrupts this process during the sensitive period of development of the visual system, leading to irreversible visual impairment [24, 26].

Cataracts in children can be classified according to the age of onset, etiology, and morphology [35, 36]. This pathology can be unilateral or bilateral, idiopathic, hereditary, or may be the result of metabolic disorders and complex syndromes [16, 31, 41, 48, 49, 64, 65]. Idiopathic congenital cataracts account for up to two-thirds of all cases[67], making primary prevention much more difficult.

The main treatment for congenital cataracts is surgical intervention aimed at removing of the lens with opacity, followed by prompt optical correction [50, 62]. The effectiveness of the treatment of children with congenital cataracts largely depends on the early detection of the disease and the timely referral of patients for surgical treatment [5, 59].

Conducting epidemiological studies is the first step towards planning the allocation of health sector resources and reducing the burden of preventable blindness due to congenital cataracts [28, 60].

Purpose of the study: to review the epidemiological characteristics of congenital cataracts, as well as to reflect the statistical data of the Almaty city branch of the Republican State Enterprise on the right of economic management "Republican e-health center" of the Ministry of Health of the Republic of Kazakhstan on the incidence of congenital cataracts in children in Almaty and the number of children 0-17 years old who are registered with a dispensary with a diagnosis "Cataract" in the Republic of Kazakhstan.

Search strategy. Literature search was carried out in electronic databases MEDLINE, PubMed, EMBASE, Web of Science, Google Scholar and e-library, using keywords (congenital cataract; children; prevalence; incidence). The search depth was 25 years (from 1996 to 2021).

Inclusion criteria: full-text articles published in English and Russian.

Exclusion criteria: duplication or repetition of information, media articles.

Relevant papers reflecting the characteristics of the problem were accepted for description in the review. Literature lists of relevant studies were also reviewed for additional sources. After the abstracts of the articles were evaluated, the corresponding articles with full-text available were added to the Mendeley library and checked for duplication.

Of all the selected articles for further analysis, 71 sources were included that met the inclusion criteria and excluded duplication or repetition of information.

Results and discussion

Prevalence and incidenceof congenital cataract

The prevalence of congenital cataracts varies considerably around the world[67]. Discrepancies between studies may be due to differences in study methodology, and likely to differing prevalences of hereditary, environmental, socioeconomic risk factors.

Studies from developed countries, such as Sweden, Great Britain, Denmark, are based on data from national registers and medical records systems, which allows conducting high-quality studies on the prevalence of diseases among the population [12, 33]. In contrast, studies from developing countries such as India and China are mostly regional or single center cross-sectional studies [32, 58].

X. Wu et al. conducted a systematic review and meta-analysis to estimate the global prevalence of congenital cataract [67]. The study included 17 population-based studies conducted between 1959 and 2010, involving 8,302,708 children from various regions of the world, including Asia, Europe, Australia, Africa, and the United States. The prevalence of congenital cataracts varied between 2.2 per 10,000 pediatric population and 13.6 per 10,000 in the included studies. The pooled prevalence was 4.24 cases per 10,000 population, with the highest prevalence in Asia and an upward trend since 2000. It should be noted that the Asian region was represented only by studies from India and the People's Republic of China, due to the lack of published data from other countries in the region[67].

S. Sheeladevi et al. also conducted a systematic review of existing studies on the prevalence and incidence of congenital and acquired cataracts in children worldwide[53]. The authors assessed these indicators according to the country's income level, calculated using the World Bank Atlas. The overall prevalence of congenital cataract in children, based on 13 studies from different regions of the world, ranged from 0.63 to 9.74 per 10,000 children (median 1.71 per 10,000). The prevalence in low-income countries ranged from 0.42 to 2.05 per 10,000 children, while in high-income countries it ranged from 0.63 to 13.6 per 10,000 children. The incidence of congenital cataract in world ranged from 1.8 to 13.6 per 10,000 child population per year [53].

A study conducted in Sweden over a 16-year period showed that the incidence of congenital cataract was 3.6 per 10,000 child population. For the entire period of the study, the incidence rates remained at the same level [1].

According to *J.M. Holmes et al.*, the prevalence of visually significant congenital cataract in the white population of the United States of America is 3.0 - 4.5 per 10,000 births. The authors analyzed the medical records of

all children (0–17 years) over a 20-year period (from 1978 to 1997) enrolled in the Rochester Epidemiology Project. There were 10 cases of visually significant congenital cataract during the entire study period, as well as 5 possible cases of congenital cataract in children aged 2 to 8 years [13].

A study from United Kingdom found that the incidence of congenital cataract in the first year of life was 2.49 per 10,000 children (95% CI: 2.10-2.89 per 10,000), increasing to 3.46 per 10,000 child population (95% CI: 3.02–3.90 per 10,000) by age of 15 [45]. This trend highlights the fact that congenital cataracts may not always be easily recognized at birth.

V.L Krasilnikova analyzed the structure of eye pathology among the pediatric population of the Republic of Belarus. In 2011, 987 cases of congenital pathology of the eye were detected, in 2010 - 1090 cases. Congenital cataract was diagnosed in 495 children, which is about half of the cases of anomalies in the development of the organ of vision in children [21].

The prevalence of congenital cataracts varies greatly even within individual countries. For example, the prevalence of this pathology in the People's Republic of China varies significantly depending on the region: the highest rates are in the west of the country (22.7 per 10,000 children)[42], the lowest in the southeast (0.7 per 10,000 children) [68]. The authors note that Western China is inferior in socio-economic development comparing to other regions of the country, which may be the reason for such high prevalence rates [42].

In the Russian Federation, the incidence of cataract in children aged 0-14 years is 29.7 per 100 000 of the pediatric population and ranks second in the incidence structure after myopia. This indicator is characterized by significant variability depending on the region. Low rates are recorded in such regions as Kaluga, Ivanovo, Yaroslavl, Murmansk regions, the Republic of Karelia. A high incidence of cataracts is noted in the regions: Belgorod, Kurgan, Smolensk, Volgograd, in the republics of Kabardino-Balkaria, Mordovia, Zabaykalsky and Khabarovsk regions. The highest incidence (71.4 per 100 000 children) is observed in Dagestan[17].

Low vision and blindness due to congenital cataracts can be prevented through early diagnosis, timely surgical intervention and subsequent rehabilitation. Thus, after the introduction of vision screening programs for newborns and preschool children in Northern Ireland, the incidence (per 1,000,000 population under the age of 16 years) of blindness due to congenital cataracts decreased more than twice from 5.89 (95% CI: 2.82-10.83) in 1984-1987 to 2.63 (95% CI: 0.72-6.74) in 2008-2011 [55].

Other epidemiological characteristics

Many studies have examined the prevalence of congenital cataracts among boys and girls. In Canada, *Z. Lim et al.* found that the proportion of boys was 56.2% of all cataract cases in children [30]. Similar results were obtained in a study from Nigeria, where *B.A. Olusanya et al.* determined that the proportion of males with congenital cataract was 54.9% [40]. Studies conducted in China also indicate a higher prevalence of congenital cataract among boys (60.4-63.6%) [32, 69, 71]. Similar data were obtained in studies from India, Korea, and the United Kingdom[39, 46, 52]. However, studies from Sweden and Denmark showed an approximately equal ratio of boys and girls among cases of cataracts in children (1.07) [33]. The same conclusion was reached by *S. Sheeladevi et al.* during a systematic review of the prevalence of cataracts in children in the world [53]. *O. Fakhoury et al.* in a study conducted in France found that the proportion of females slightly prevailed over males among cases of congenital cataract in children (53%) [6].

Regarding etiology, a recent systematic review and meta-analysis reported that the pooled proportions of hereditary, nonhereditary and idiopathic congenital cataract were 22.3%, 11.5% and 62.2%, respectively[67]. Lim et al. found the proportion of inherited cataracts to be 11.7%[30]. Danish epidemiologic study of 1027 cases of congenital/infantile cataract reported higher proportion of hereditary cataract (23%) [11], which was similar to the 18.8% reported by *Wirth et al*. in Australia [66]. This difference might be explained by the different prevalence of hereditary risk factors among populations.

Advances in genetic mapping and DNA sequencing technologies allowed to determine that in many cases the development of congenital cataracts was associated with mutations in genes encoding lens crystallins, connexins, aquaporin, cytoskeletal structural proteins, and other key regulators of lens development [3, 37]. To date, more than 100 genes are associated with congenital cataract[54].The severity of congenital cataracts associated with different or even identical mutations of the same gene can vary from dense opacity to almost transparent[29]. The majority of inherited congenital cataracts (75%) being autosomal dominant[49], but some cataracts are inherited according to autosomal recessive, X-linked or even mitochondrial DNA inheritance[29].

Congenital cataract can also be associated with other ocular and/or systemic diseases. The most common eye diseases associated with congenital cataracts are congenital aniridia[70], microcornea (congenital microcornea cataract syndrome)[27], microphthalmia[20], primary persistent hyperplastic vitreous syndrome [43], Marfan's syndrome [18], and Marchesani[61]. Systemic diseases most often associated with congenital cataracts are congenital heart and nervous system diseases, as well as Hallermann-Streiff-François syndrome [41], Wolfram syndrome [38], facial dysmorphic neuropathy and congenital cataract syndrome [27], Nancy Horan's syndrome (NHS) [65] and Low's syndrome [8, 51].

Ocular manifestations of congenital cataracts may result from inherited metabolic disorders, including galactosemia, Wilson's disease, hypocalcemia, hypo/hyperglycemia, and Lowe's syndrome[10, 25, 47]. Some congenital cataracts can be caused by intrauterine infections such as rubella virus, herpes simplex virus, toxoplasma gondii, cytomegalovirus, syphilis, and varicella virus[4, 15, 57], and therefore pregnant women should be screened for infections (toxoplasma, rubella virus, cytomegalovirus, HSV and others) [34].In addition, malnutrition during pregnancy [23], radiological exposure [56], drugs such as linezolid [14], and intrauterine hypoxia can cause cataracts in children.

Most of the published studies indicate the predominance of bilateral forms of congenital cataract over unilateral ones. Thus, the proportion of bilateral forms

ranges from 63.3 to 86% in studies from France, India, Sweden, Denmark, Great Britain [6, 33, 44, 52]. The possible explanation for this finding may be the fact that thementioned studies reported surgical cases of this condition. In a systematic review and meta-analysis of the prevalence and epidemiological characteristics of congenital cataract, *X. Wu et al.* found that the bilateral form accounts for 54% of all cases of congenital cataract in children [67].

The morphology of cataract may be a predictor of visual prognosis. Lamellar, posterior polar, and posterior lenticonus cataracts are often associated with favorable visual outcomes. The worse results often occur in younger children with greater density of mature cataract or opacity in the visual axis[69]. According to the literature, the most common clinical and morphological forms of congenital cataractsare diffuse (31.2%), nuclear (27.2%) and posterior subcapsular forms (26.8%) [67].Chinese study found that congenital cataract was total in 84.4% of pediatric patients with cataract[69]. In contrast, Holmes et al. found that infantile cataract was total in 2 (13.3%) cases in a defined US population[13].This finding would suggest late detection of cataract in developing countries because many types of cataract slowly become total in untreated cases[67].

Statistical data of the Almaty city branch of the RSE on the REM "Republican e-health center" of the Ministry of Health

The incidence of congenital cataract among children in Almaty was studied for the period of 2015-2019 according to the statistical data of the Almaty city branch of the Republican State Enterprise on the right of economic management "Republican e-health center" of the Ministry of Health of the Republic of Kazakhstan. During the study period, there is an irregular increase in the incidence. The overall incidence of congenital cataracts in children per 100 000 of the population showed that the maximum absolute increase was detected in 2016 (Figure 1).

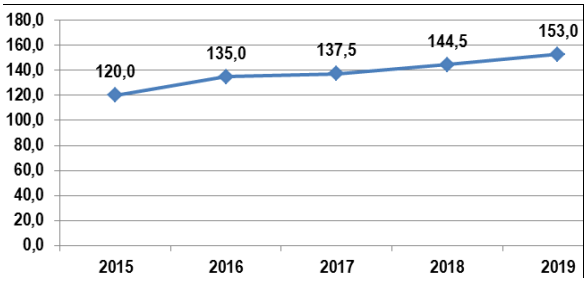

Figure 1. Incidence of congenital cataract among children per 100,000 population in Almaty.

For 2015-2019 the number of children aged 0-17 years old, registered with a diagnosis of "Cataract" in the Republic of Kazakhstan, increased by more than 1.5 times: from 475 children in 2015 to 720 children in 2019 (Figure 2). A high level of absolute growth was set in 2016 and 2018, and a minimum in 2017 (Table 1).

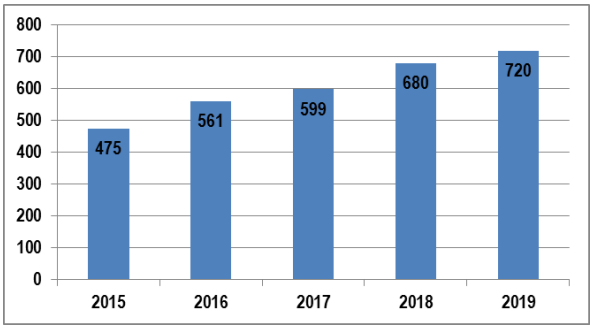

Figure 2. The number of children aged 0-17 years old who are registered with a diagnosis of "Cataract" in the Republic of Kazakhstan

Table 1.

Dynamics of the number of children aged 0-17 years old, registered with a diagnosis of "Cataract" in the Republic of Kazakhstan.

Year	Absolute number	Absolute growth	Indicator of visibility, %	Indicator of growth, %	Growth rate, %	Value of 1% increase
2015	475	-	100,0	-	-	-
2016	561	86,0	118,1	118,1	18,1	4,8
2017	599	38,0	126,1	106,8	6,8	5,6
2018	680	81,0	143,2	113,5	13,5	6,0
2019	720	40,0	151,6	105,9	5,9	6,8

Possible explanation to the increase of the incidence of congenital cataract and the number of children registered with this diagnosis might be the fact that in recent years more ophthalmologists in the Republic of Kazakhstan realized the importance of early detection and surgery for congenital cataract. It is crucial to educate medical practitioners on this disease to decrease the burden of preventable childhood blindness.

Conclusions:

1. Congenital cataract is one of the main causes of preventable childhood blindness and low vision.

2. The prevalence and incidence of congenital cataract varies greatly around the world.

3. Many epidemiological studies indicate more frequent occurrence of this pathology among boys.

4. The bilateral form of congenital cataract is more common, and diffuse, nuclear and posterior subcapsular forms predominate among the clinical and morphological forms.

5. According to the Almaty city branch of the RSE on REM "Republican e-health center" of the Ministry of Health of the Republic of Kazakhstan, there is an increase in the incidence of children with congenital cataracts in the city of Almaty, as well as an increase in the number of children 0-17 years old who are registered with a diagnosis of "Cataract" in the Republic of Kazakhstan.

Conflict of Interest. *The authors declare that they have no conflict of interest.*

Contribution of authors. *All authors were equally involved in the writing of this article.*

Funding: *No funding was provided.*

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