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Epidemiological Characteristics of Hereditary Diseases of the Visual Organ in Surkhandarya Region

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Abstract

Hereditary diseases of the visual organ represent a significant medical and social problem, especially in regions such as the Surkhandarya region of Uzbekistan, where access to specialized ophthalmological care is limited. Failure to detect these pathologies promptly leads to progressive vision loss and patient disability. This study aimed to determine the prevalence and the actual detection rate of hereditary eye diseases in the Surkhandarya region. A retrospective epidemiological study was conducted based on data from the Termez branch of the Republican Specialized Scientific and Practical Medical Center of Eye Microsurgery. Outpatient medical records, hospital discharge summaries, and ophthalmological examination reports of patients for the period 2022–2024 were analyzed. The assessment included nosological forms, sex, age, place of residence, and family history. A significant discrepancy was identified between the expected and the actual number of hereditary diseases of the visual organ. The greatest mismatch was observed in retinitis pigmentosa and hereditary retinal dystrophies. For all nosological forms, the detection rate was significantly lower than expected values, indicating substantial underdiagnosis of these conditions in the region. The obtained results emphasize the need to improve early detection systems for hereditary eye diseases in the Surkhandarya region, enhance physician training, and expand access to ophthalmological care.

Keywords: hereditary diseases, visual organ, epidemiology, diagnostics.

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Introduction

Hereditary diseases of the visual organ represent a serious medical and social problem, especially in regions such as the Surkhandarya region of Uzbekistan, where access to specialized ophthalmological care is limited. These pathologies often lead to progressive vision loss and disability, significantly reducing patients' quality of life [1,2,5].

According to the World Health Organization, congenital and genetic pathologies constitute a substantial

proportion of eye diseases among children and young people worldwide [3,4]. In Uzbekistan, the incidence of diseases of the eye and its accessory apparatus has increased; between 2012 and 2024, the overall morbidity rate rose by 32.8%. This trend highlights the urgent need to develop effective strategies for the diagnosis, treatment, and rehabilitation of patients with ophthalmic diseases, including hereditary conditions.

In the Surkhandarya region, insufficient public awareness and limited access to modern diagnostic technologies create significant challenges in the timely detection of hereditary eye diseases. In addition, there is a shortage of qualified ophthalmologists and specialized medical institutions in remote areas, which complicates the provision of adequate care to patients with hereditary disorders of the visual organ.

Aim of the Study

To determine the prevalence and the actual detection rate of hereditary diseases of the visual organ in the Surkhandarya region.

Materials and Methods

The study was retrospective and epidemiological in nature and was conducted in 2024. The research was based on data obtained from the Termez branch of the Republican Specialized Scientific and Practical Medical Center of Eye Microsurgery (RSSPMCMEM).

The analysis was carried out in two stages:

- **Desk (analytical) stage.** A review of contemporary scientific literature and major epidemiological studies on the prevalence of hereditary eye diseases (retinitis pigmentosa, Leber congenital amaurosis, congenital glaucoma, hereditary retinal and optic nerve dystrophies, and developmental anomalies of the anterior segment of the eye) was conducted.
- **Practical stage.** Medical records of patients who applied to the Termez branch of the RSSPMCMEM between 2022 and 2024 were analyzed. The following sources were examined: outpatient medical records, hospital discharge summaries, and ophthalmological examination reports.

The dataset included the following variables: nosological form of the disease, patients' sex and age, place of residence, and family history (presence of hereditary pathology).

To calculate prevalence, the official population of the Surkhandarya region in 2024 (2,877,000 people) was used. The expected number of cases was determined based on data from international literature regarding the prevalence of the corresponding nosological forms. The actual number of cases reflected the number of patients registered in the medical documentation database of the Termez branch of the RSSPMCMEM. The detection rate was assessed by comparing expected and actual cases, taking into account underdiagnosis-related bias.

Results

The study compared the expected and actual prevalence of hereditary diseases of the visual organ in the Surkhandarya region. According to the data presented in Table 1, a significant underdetection of cases was identified for all nosological forms compared with expected values. Thus, for retinitis pigmentosa, the actual number of patients was 207, compared to an expected 575–792 cases, while for Leber congenital amaurosis, 13 cases were identified versus an expected 29–58 cases. A similar trend was observed for other conditions, including congenital glaucoma, hereditary retinal and optic nerve dystrophies, as well as developmental anomalies of the anterior segment of the eye.

The graphical representation of the results (Figure 1) clearly demonstrates a pronounced discrepancy between the expected and actual number of registered cases across all studied nosologies. The most significant mismatch was observed in retinitis pigmentosa and hereditary retinal dystrophies, where the proportion of detected cases was less than half of the expected level. A less pronounced, yet still considerable, discrepancy was noted for congenital glaucoma and hereditary optic nerve dystrophies. The

lowest detection rate was observed for developmental anomalies of the anterior segment of the eye.

Table 1. Estimated and actual number of cases of hereditary diseases of the visual organ in the Surkhandarya region

(Population in 2024: 2,877,000)

Disease	Prevalence (per 100,000 population)	Expected number of cases	Actual number of detected cases
Retinitis pigmentosa	20–27.5	575–792	207
Leber congenital amaurosis	1–2	29–58	13
Congenital glaucoma	~1 case per 2,500 live births	29	9
Hereditary retinal dystrophies	10.4	299	90
Hereditary optic nerve dystrophies	3	86	26
Developmental anomalies of the anterior segment of the eye	0.3–0.6	9–17	4

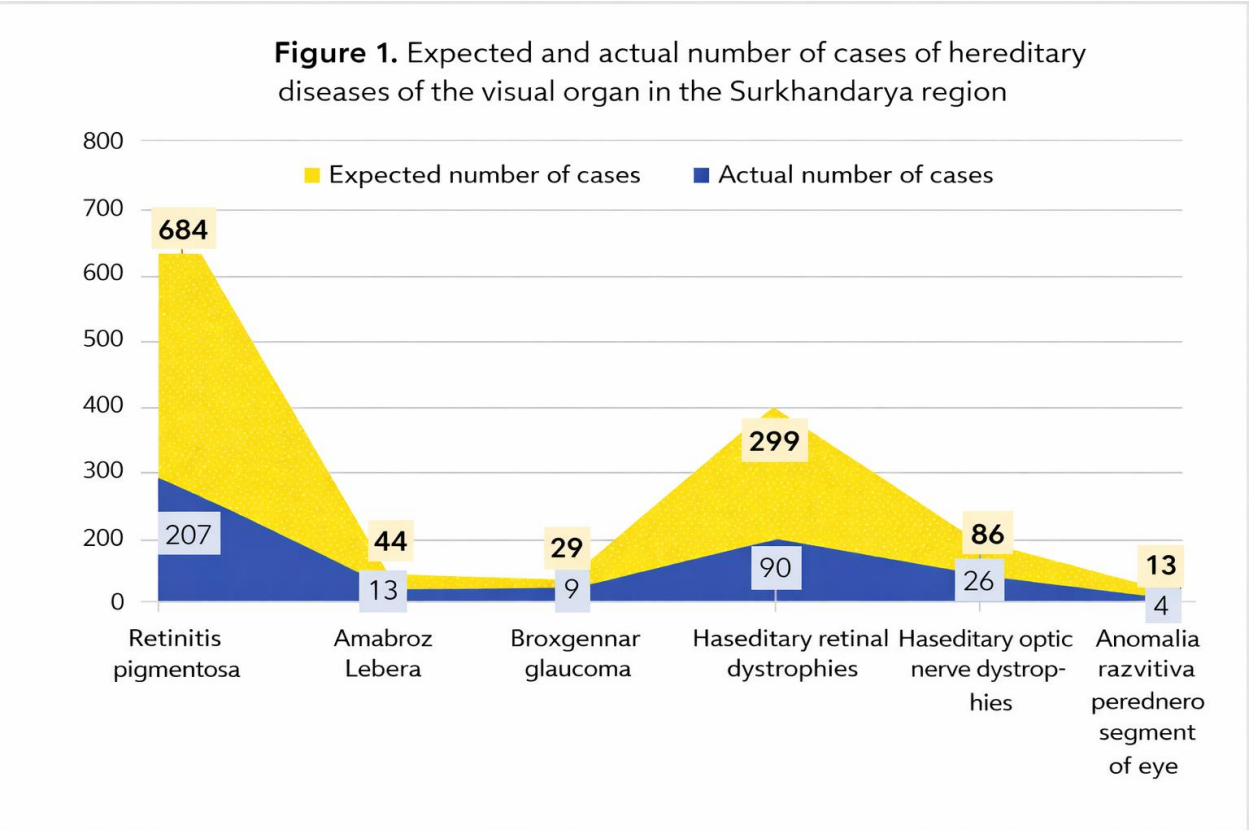


Figure 1. Ratio of expected and actual number of cases

Discussion

The obtained data confirm the existence of a significant problem of underdiagnosis of hereditary diseases of the visual organ in the Surkhandarya region. For all studied nosological forms, the actual indicators were markedly lower than the expected values based on data from international literature. This finding indicates the low effectiveness of the existing system for early diagnosis of ophthalmic diseases, particularly those of hereditary origin.

Analysis of the identified discrepancies shows that the most difficult conditions to diagnose are diseases characterized by slow and progressive symptom development, such as retinitis pigmentosa and hereditary retinal dystrophies. The low detection rates of congenital glaucoma and developmental anomalies of the

anterior segment of the eye also indicate insufficient alertness among general practitioners and pediatricians, who often serve as the primary link in patient referral pathways. In addition, the absence of specialized ophthalmological centers in remote areas of the region, limited availability of modern diagnostic methods (including genetic testing), and low levels of medical awareness among the population contribute to delayed diagnosis.

The results of this study are consistent with findings from other studies conducted in developing countries that address the prevalence of ophthalmic diseases [2,6,7], where the problem of late diagnosis of hereditary pathologies has also been reported.

Conclusion

Thus, the findings demonstrate insufficient diagnostic coverage of hereditary visual disorders in the region, which is likely associated with limited access to specialized ophthalmological care, low public awareness, and a shortage of qualified specialists. To improve the situation, it is necessary to strengthen preventive programs aimed at early disease detection, expand screening networks, enhance training programs for primary healthcare physicians in the diagnosis of hereditary eye diseases, and integrate modern molecular genetic methods into standard ophthalmological practice in the region.

References

1. Hanany M, Rivolta C, Sharon D. The worldwide landscape and genetic architecture of inherited retinal disorders. *Proceedings of the National Academy of Sciences of the United States of America*. 2020;117(5):2710–2716. https://doi.org/10.1073/pnas.1913179117
2. Hanany M, Shalom S, Ben-Yosef T, Sharon D. Global prevalence and complex genetic architecture of inherited retinal disorders and their variable interactions. *Cold Spring Harbor Perspectives in Medicine*. 2024;14(2):a041277. https://doi.org/10.1101/cshperspect.a041277
3. Jain M, Anjani P, Krishnamurti G, Sachdeva V, Kekunnaya R. One-year profile of pediatric eye diseases at secondary (rural) eye care centers in South India. *Indian Journal of Ophthalmology*. 2021;69(4):906–909. https://doi.org/10.4103/ijo.IJO_1084_20
4. Jammeih S, Malik S, Okashah S, Zayed H. Genetic epidemiology of primary glaucoma in 22 Arab countries: A systematic review. *Ophthalmic Epidemiology*. 2022;29(1):1–12. https://doi.org/10.1080/09286586.2021.1883676
5. Mai CT, Isenburg JL, Canfield MA, Meyer RE, Correa A, Alverson CJ, et al.; National Birth Defects Prevention Network. National population-based estimates for major birth defects, 2010–2014. *Birth Defects Research*. 2019;111(18):1420–1435. https://doi.org/10.1002/bdr2.1589
6. Sheck LHN, de Esposti SD, Mahroo OA, Arno G, Pontikos N, Wright G, Webster AR, Khan KN, Michaelides M. Panel-based genetic testing for inherited retinal disease screening across 176 genes. *Molecular Genetics & Genomic Medicine*. 2021;9(12):e1663. https://doi.org/10.1002/mgg3.1663
7. Solebo AL, Teoh L, Rahi J. Epidemiology of blindness in children. *Archives of Disease in Childhood*. 2017;102(9):853–857. https://doi.org/10.1136/archdischild-2016-310532