Clinical Spectrum of Pediatric Congenital Ocular Anomalies at a Tertiary Eye Care Center: A One-Year Retrospective Review.

Abstract:

Purpose: To evaluate the clinical spectrum and epidemiological profile of pediatric congenital ocular anomalies presenting to a tertiary eye care center over a one-year period.

Methods: This was a retrospective, observational study. Medical records of pediatric patients diagnosed with congenital ocular anomalies over a one-year period were reviewed. Data regarding patient demographics, laterality, and specific morphological diagnoses were extracted and analyzed using descriptive statistics.

Results: A total of 200 pediatric patients were included (mean age 5.8 ± 3.4 years). The cohort comprised 112 males (56%) and 88 females (44%). Unilateral anomalies were seen in 104 patients (52%), while 96 patients (48%) had bilateral involvement. The most frequently observed anomaly was congenital cataract (n=52, 26%), followed by congenital ptosis (n=38, 19%), coloboma (n=31, 15.5%), microphthalmos (n=24, 12%), congenital glaucoma (n=21, 10.5%), and anophthalmos (n=12, 6%). Other anomalies accounted for 11% (n=22) of cases.

Conclusion: Congenital cataract, ptosis, and coloboma are the leading structural ocular anomalies in this demographic. Given the high risk of amblyopia and lifelong visual impairment, early diagnostic screening and timely surgical or optical interventions are critical for optimizing visual outcomes.

Introduction

Congenital ocular anomalies constitute an important cause of childhood visual impairment and blindness worldwide.[1] These anomalies arise due to disturbances in ocular development during embryogenesis and may affect the globe, adnexa, or visual pathways.[2] Early diagnosis is essential as many conditions are amenable to timely intervention, thereby reducing the burden of preventable visual disability.

The prevalence and pattern of congenital ocular anomalies vary across geographical regions and populations.[3] Factors such as genetic predisposition, consanguinity, maternal infections, and environmental influences have been implicated in their development.[2,4] Understanding the local epidemiological profile is important for planning screening programs and healthcare resource allocation.

The present study aimed to evaluate the clinical spectrum of pediatric ocular anomalies presenting to a tertiary eye care center over a one-year period.

Materials and Methods

This retrospective, observational study was conducted at a tertiary eye care center. The study adhered to the tenets of the Declaration of Helsinki.[5] Medical records of pediatric patients presenting with congenital ocular anomalies over a continuous one-year period were retrieved and reviewed.

Comprehensive ophthalmic examination data were extracted, including anterior segment evaluation via slit-lamp biomicroscopy and intraocular pressure measurement. Detailed posterior segment evaluation via indirect ophthalmoscopy—crucial for assessing the extent of retinochoroidalcolobomatous defects and associated optic nerve anomalies—was recorded. Data collected included age at presentation, biological sex, laterality (unilateral versus bilateral), and primary morphological diagnosis. Descriptive statistics were utilized to calculate frequencies and percentages.

Results

A total of 200 pediatric patients with documented congenital ocular anomalies were included in the study. The mean age at presentation was 5.8 ± 3.4 years. The demographic distribution showed a slight male preponderance, with 112 males (56%) and 88 females (44%). Regarding laterality, unilateral involvement was observed in 104 patients (52%), while bilateral disease was present in 96 patients (48%).

Table 1. Patient Demographics and Anomaly Laterality

Characteristic	Frequency (n=200)	Percentage (%)
Sex		
Male	112	56.0
Female	88	44.0
Laterality		
Unilateral	104	52.0
Bilateral	96	48.0

Lens abnormalities formed the largest subset of anomalies. Congenital cataract was the most common single anomaly, accounting for 52 cases (26%). This was followed by adnexal and closure defects, including congenital ptosis in 38 cases (19%) and coloboma in 31 cases (15.5%).

Table 2. Distribution of Congenital Ocular Anomalies

Congenital Anomaly	Frequency (n)	Percentage (%)
Congenital Cataract	52	26.0
Congenital Ptosis	38	19.0
Coloboma	31	15.5

Congenital Anomaly	Frequency (n)	Percentage (%)
Microphthalmos	24	12.0
Congenital Glaucoma	21	10.5
Anophthalmos	12	6.0
Others	22	11.0

Discussion

The present study demonstrated that congenital cataract was the most common pediatric ocular anomaly encountered at our center, followed by congenital ptosis and coloboma. Similar findings have been reported in several hospital-based studies from developing countries.[6]

Congenital cataract remains a leading cause of treatable childhood blindness.[7] Delayed presentation continues to be a challenge, particularly in resource-limited settings. Early identification through neonatal and preschool screening programs is crucial to prevent deprivational amblyopia and optimize long-term visual outcomes.[1,7]

The predominance of congenital ptosis and coloboma observed in our study is comparable to previous reports. Colobomatous defects result from incomplete closure of the embryonic fissure and may be associated with significant visual morbidity depending on the extent of macular, retinal, and optic nerve involvement.[8]

Nearly half of the patients (48%) exhibited bilateral disease, emphasizing the importance of comprehensive ocular examination and systemic evaluation. Bilateral anomalies often have a stronger genetic basis and may be frequently associated with underlying syndromic conditions.[9]

The primary strength of this study includes the comprehensive clinical documentation and the representation of a broad morphological spectrum of ocular developmental anomalies. Limitations include its retrospective nature, the potential for tertiary referral bias, and the absence of genetic testing or long-term visual outcome assessments.

Conclusion

Congenital cataract, congenital ptosis, and coloboma were the most common pediatric ocular anomalies observed in this cohort. Early diagnosis and timely clinical intervention remain critical for reducing the burden of childhood visual impairment and blindness.

References

1. Gilbert C, Foster A. Childhood blindness in the context of VISION 2020: the right to sight. *Bull World Health Organ*. 2001;79(3):227-232.
2. Hornby SJ, Adolph S, Gothwal VK, et al. Evaluation of children in six blind schools of Andhra Pradesh. *Indian J Ophthalmol*. 2000;48(3):195-200.
3. Solebo AL, Teoh L, Rahi J. Epidemiology of blindness in children. *Arch Dis Child*. 2017;102(9):853-857.
4. Gogate P, Khandekar R, Shinde A, et al. Causes of childhood blindness in Maharashtra, India. *Br J Ophthalmol*. 2007;91(9):1127-1131.
5. World Medical Association. World Medical Association Declaration of Helsinki: ethical principles for medical research involving human subjects. *JAMA*. 2013;310(20):2191-2194.
6. Kello AB, Gilbert C. Causes of severe visual impairment and blindness in children in schools for the blind in Ethiopia. *Br J Ophthalmol*. 2003;87(5):526-530.
7. Sheeladevi S, Lawrenson JG, Fielder AR, Suttle CM. Global prevalence of childhood cataract: a systematic review. *Eye (Lond)*. 2016;30(9):1160-1169.
8. Chang L, Blain D, Bertuzzi S, Brooks BP. Uveal coloboma: clinical and basic science update. *CurrOpinOphthalmol*. 2006;17(5):447-470.
9. Stoll C, Alembik Y, Dott B, Roth MP. Epidemiology of congenital eye malformations in 131,760 consecutive births. *Ophthalmic Paediatr Genet*. 1992;13(3):179-186.