Early Recognition and Surgical Intervention in Bilateral Congenital Glaucoma: A Case Report Highlighting Genetic Predisposition and Multidisciplinary Management

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Abstract

Background: Childhood glaucoma is a significant yet often underrecognized cause of pediatric blindness. Unlike adult glaucoma, which is typically asymptomatic, childhood glaucoma presents with observable signs that can be detected by pediatricians and even parents. However, there is a lack of epidemiologic and clinical data on this condition, leading to delayed diagnoses and suboptimal outcomes. Methods: A detailed ophthalmologic evaluation under general or local anesthesia is essential for accurate diagnosis and management planning. While medical therapy plays a limited role, surgical intervention is the primary treatment modality. Goniotomy and trabeculotomy are effective in congenital glaucoma, with primary combined trabeculotomy–trabeculectomy recommended for advanced cases. Results: The Childhood Glaucoma Research Network (CGRN) has introduced a new classification system for pediatric glaucomas, enhancing the diagnosis and management of these conditions. Early diagnosis, prompt surgical intervention, and proper refractive correction are critical for successful outcomes. This comprehensive approach can lead to a long, morbidity-free life for affected children. Conclusion: The management of childhood glaucoma requires a holistic approach, including early detection, surgical intervention, and lifelong follow-up. Visual rehabilitation and the management of residual vision are integral components of care. Ensuring early and effective treatment can significantly improve the prognosis, allowing children with glaucoma to maintain functional vision and quality of life.

Keywords: Congenital glaucoma, trabeculotomy, trabeculectomy, combined trabeculotomy–trabeculectomy.

Significance | Early diagnosis and surgical intervention in congenital glaucoma are crucial to preventing irreversible vision loss in pediatric patients.

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Introduction

Glaucoma in childhood represents a significant, albeit often underrecognized, cause of pediatric blindness (Dandona et al., 1998; Fung et al., 2013). This ocular condition, characterized by increased intraocular pressure (IOP) leading to optic nerve damage, is particularly devastating in children due to its potential to impair vision during critical periods of visual development. Unlike adult glaucoma, which is typically asymptomatic in its early stages, childhood glaucoma often presents with observable signs and symptoms that can be identified by pediatricians and even parents. However, despite its clinical presentation, there remains a scarcity of epidemiologic and clinical data regarding childhood glaucoma, contributing to delayed diagnoses and suboptimal outcomes (Gupta et al., 2016; Taylor et al., 1999).

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The management of childhood glaucoma necessitates a specialized approach due to the unique challenges it presents. Diagnosis typically requires a comprehensive ophthalmologic evaluation, which may need to be conducted under general or local anesthesia to ensure accuracy and patient comfort. Early intervention is crucial, as delayed treatment can lead to irreversible vision loss. While medical therapy can offer temporary control of IOP, it is generally insufficient as a standalone treatment. Surgical intervention remains the cornerstone of management, with procedures such as goniotomy or trabeculotomy being particularly valuable in cases of congenital glaucoma (Møller, 1977). For advanced cases, a combined approach using trabeculotomy and trabeculectomy has been shown to offer the best outcomes, preserving vision and minimizing complications (Mukkamala et al., 2015; Sood et al., 2018).

Recent advancements in the classification and understanding of childhood glaucoma, particularly through the work of the Childhood Glaucoma Research Network (CGRN), have provided a more structured framework for diagnosing and managing this complex condition. These developments underscore the importance of early and accurate diagnosis, followed by prompt surgical intervention. Additionally, ongoing management, including refractive correction and visual rehabilitation, is essential for optimizing long-term outcomes and ensuring that affected children can lead a life with minimal visual morbidity.

Childhood glaucoma is a critical yet manageable condition that demands early recognition and intervention. With the proper approach, including timely surgery and lifelong follow-up, the prognosis for children with glaucoma can be significantly improved, allowing them to maintain functional vision and quality of life. This case report and subsequent discussion aim to highlight the importance of comprehensive management in childhood glaucoma, drawing attention to the need for heightened awareness and early action in pediatric populations.

Case Report Presentation

A one-day-old male infant, born at full term via emergency lower segment cesarean section (LSCS) at Sree Balaji Medical College and Hospital on September 24, 2019, was presented with bilateral bluish discoloration of the eyes (Figure 1). The baby, weighing 2.330 kg, had an otherwise uneventful delivery, and both mother and baby were in stable condition post-delivery. The bluish discoloration of the corneas was noted upon examination by an ophthalmologist who was called to the neonatal intensive care unit (NICU) for further evaluation. The examination revealed an increased corneal diameter and diffused corneal edema, which obscured the view of the posterior structures (Figure 3).

Given the presentation, congenital glaucoma was highly suspected. Intraocular pressure (IOP) measurements and corneal diameter assessments were conducted under local anesthesia. The initial management included the administration of topical beta-blocker eye drops along with antibiotics. The infant was then referred to a specialist with expertise in congenital glaucoma for further evaluation and treatment. After a comprehensive examination under anesthesia, trabeculotomy was performed on the right eye, followed by a similar procedure on the left eye at a later date. Post-surgical management included regular follow-ups with both the ophthalmologist and the pediatrician to monitor the infant's progress and ensure the effectiveness of the treatment.

Family History

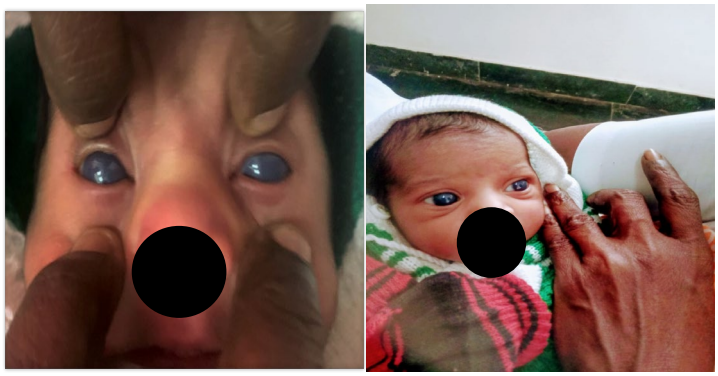
A detailed family history revealed that the infant's maternal grandmother had experienced a twin delivery, with both daughters (one of whom is the infant's mother) presenting similar ocular complaints at birth. The grandmother reported that both daughters underwent a surgical procedure approximately 40 days after birth, which, based on the current examination of the mother, appears to have been a peripheral iridotomy, possibly performed using the Scheie's procedure. Examination of the mother revealed an alternating divergent squint (exotropia), an updrawn pupil in the right eye, and a peripheral iridotomy scar in the left eye at the 7 o'clock position (Figure 5). Bedside visual acuity testing indicated that both eyes had a visual acuity of greater than 5/60.

Results and Discussion

Primary Congenital Glaucoma

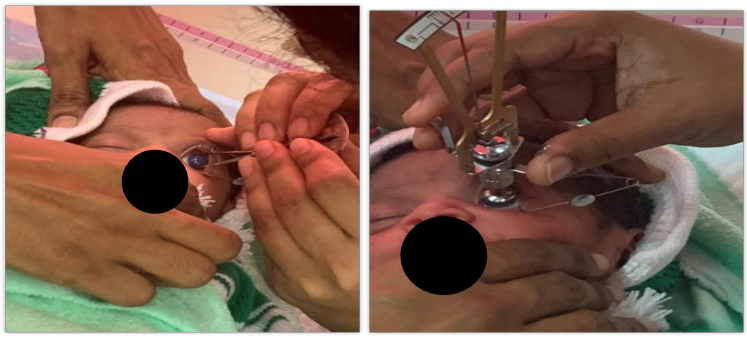
Primary congenital glaucoma (PCG) is the most common form of pediatric glaucoma, accounting for approximately 38% of all childhood glaucoma cases. The incidence of PCG in India, as calculated in a large-scale study in Andhra Pradesh, is around 1 in 3300 live births, although the global incidence is estimated to be 1 in 10,000. PCG can be categorized into three types: true congenital glaucoma, which manifests in utero; infantile glaucoma, which appears before the age of three; and juvenile glaucoma, which is less common and develops between the ages of 3 and 16 years.

The pathogenesis of PCG is primarily linked to impaired aqueous outflow due to maldevelopment of the anterior chamber angle. The prognosis depends significantly on the severity of the condition and the age at diagnosis. The classical triad of congenital glaucoma includes epiphora (excessive tearing), photophobia (sensitivity to light), and blepharospasm (involuntary tight closure of the eyelids). Affected patients are predominantly male (65%) and typically present with bilateral involvement (70%) within the first year of life (80%). If left untreated, the disease can lead to visual loss due to Descemet's breaks, corneal edema, optic neuropathy, buphthalmos



A B

Figure 1. (A, B) bilateral bluish discoloration of the cornea



A B

Figure 2. measurement of IOP and corneal diameter under LA

Table 1. IOP, corneal diameter, and overall ocular health

		Right eye	Left eye
Corneal diameter	Horizontal	12mm	12mm
	Vertical	11mm	11mm
IOP	5.5gm	40.0 mmhg	41.5 mmhg
	7.5gm	36.0 mmhg	35.8 mmhg

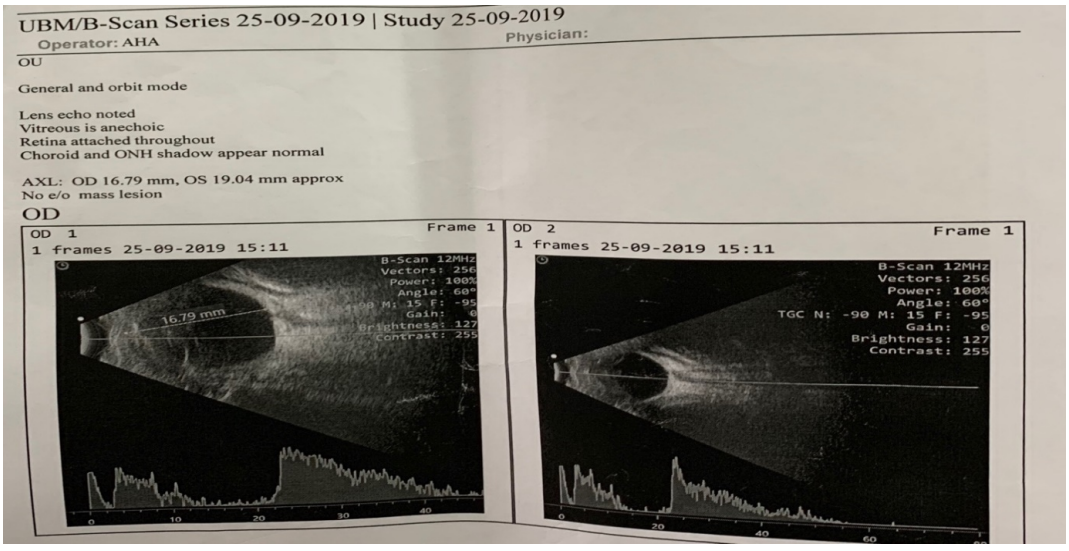


Figure 3. (USG) Bscan was done to rule out posterior segment pathology

A B

Figure 4. A) post trabeculotomy of the right eye. B) post trabeculotomy of both eyes

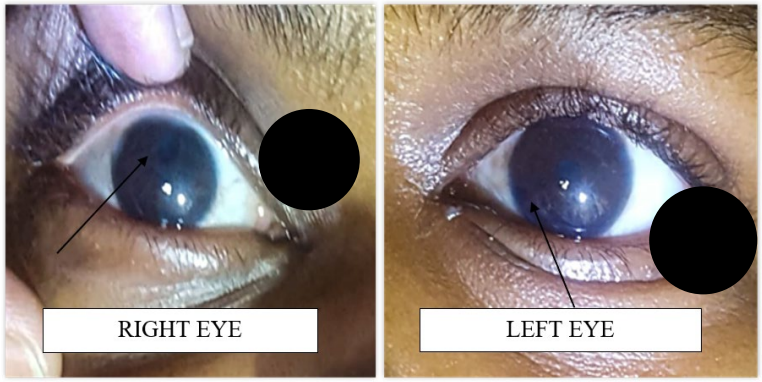


Figure 5. right eye of the mother showing updrawn pupil. Left eye of the mother showing a peripheral iridotomy scar

(enlarged eyeball due to increased IOP), and amblyopia (lazy eye) (Figure 2) (Fung et al., 2013). Given the rapid progression and potential for severe visual impairment, early diagnosis and intervention are crucial. In this case, the timely recognition of symptoms by healthcare providers led to prompt surgical intervention, significantly improving the prognosis (Mukkamala et al., 2015).

Medical and Surgical Management

The initial management of PCG often involves medical therapy to temporarily reduce IOP and clear the cornea to facilitate further examination and surgical intervention (Dandona et al., 1998). In this case, the infant was started on a combination of topical beta-blocker and antibiotic eye drops. However, medical therapy is typically a temporary measure, as surgical intervention is considered the definitive treatment for PCG.

Goniotomy and trabeculotomy are the two primary surgical procedures used to treat PCG. Goniotomy involves making an incision in the trabecular meshwork to create a drainage route for the aqueous humor. The procedure has an approximate success rate of 80% with a single intervention. However, in cases where corneal clouding is present, as seen in this infant, goniotomy is technically challenging, and trabeculotomy becomes the preferred option. Trabeculotomy, which disrupts the tissue between Schlemm's canal and the anterior chamber, allows for direct communication and drainage, with reported success rates of 87% to 92% in cases treated before one year of age.

In this case, trabeculotomy was performed on both eyes, starting with the right eye (Figure 4). The procedure was successful in reducing IOP and preventing further damage to the optic nerve and other ocular structures. Regular follow-up appointments were scheduled to monitor IOP, corneal diameter, and overall ocular health (Table 1).

Genetics of Childhood Glaucoma

Genetic factors play a significant role in the development of PCG, with 10% to 40% of cases being familial, often associated with consanguinity. The condition is typically inherited in an autosomal recessive manner, with variable expression and penetrance ranging from 40% to 100%. Three genetic loci associated with PCG have been identified: GLC3A (2p21), GLC3B (1p36), and GLC3C (14q24.3). The most commonly implicated gene is CYP1B1, with several mutations identified in various ethnic populations, including those in India.

In this case, the family history of congenital glaucoma in the infant's mother and maternal aunt strongly suggests a genetic predisposition, likely linked to mutations in the CYP1B1 gene or other related loci. Genetic counseling was recommended for the family to provide information about the hereditary nature of PCG and the potential risks for future offspring.

Conclusion

This case highlights the critical importance of early diagnosis and intervention in managing primary congenital glaucoma. The infant, who presented with classical signs of PCG, including bilateral bluish corneal discoloration and increased corneal diameter, was promptly diagnosed and treated with trabeculotomy. The positive outcome in this case underscores the effectiveness of timely surgical intervention in preventing long-term visual impairment in children with PCG.

The involvement of a multidisciplinary team, including ophthalmologists, pediatricians, and genetic counselors, is essential in managing PCG, particularly in cases with a strong family history. Regular follow-up is crucial to monitor the progression of the disease, manage any residual vision, and address complications such as amblyopia and refractive errors.

In conclusion, the management of PCG requires a comprehensive approach that includes early detection, appropriate surgical intervention, and ongoing monitoring. With proper care, children with PCG can achieve a long and morbidity-free life, minimizing the risk of blindness and other severe complications associated with this condition.

Ethics

Ethics approval was obtained from the Institutional Review Board of Sree Balaji Medical College and Hospital prior to the presentation of this case report. Written informed consent was obtained from the infant's parents for the publication of the case details and accompanying images, ensuring confidentiality and compliance with ethical standards.

Author contributions

P.M. conceptualized the study and led the overall project. A.J.R. conducted the data collection and analysis. M.R.S. and A.M. provided critical input on the methodology and interpretation of the results. All authors (P.M., A.J.R., M.R.S., and A.M.) discussed the results, contributed to the writing of the manuscript, and approved the final version.

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Competing financial interests

The authors have no conflict of interest.

References

Dandona, L., Williams, J. D., Williams, B. C., & Rao, G. N. (1998). Population-based assessment of childhood blindness in southern India. Archives of Ophthalmology, 116(4), 545-546.

Fung, D. S., Roensch, M. A., Kooner, K. S., Cavanagh, H. D., & Whitson, J. T. (2013). Epidemiology and characteristics of childhood glaucoma: Results from the Dallas Glaucoma Registry. Clinical Ophthalmology, 7, 1739. https://doi.org/10.2147/OPTH.S45480

Gupta, P. C., Singh, R. R., Pandav, S. S., & Ram, J. (2016). Rhexis fixation of intraocular lens in a case of buphthalmos with cataract. Nepalese Journal of Ophthalmology, 8(1), 71-73. https://doi.org/10.3126/nepjoph.v8i1.16140

Møller, P. M. (1977). Goniotomy and congenital glaucoma. Acta Ophthalmologica, 55(3), 436-442. https://doi.org/10.1111/j.1755-3768.1977.tb06120.x

Mukkamala, L., Fechtner, R., Holland, B., & Khouri, A. S. (2015). Characteristics of children with primary congenital glaucoma receiving trabeculotomy and goniotomy. Journal of Pediatric Ophthalmology and Strabismus, 52(6), 377-382. https://doi.org/10.3928/01913913-20151014-51

Sood, D., Rathore, A., Sood, I., Singh, G., & Sood, N. N. (2018). Long-term outcome of combined trabeculotomy-trabeculectomy by a single surgeon in patients with primary congenital glaucoma. Eye, 32(2), 426. https://doi.org/10.1038/eye.2017.207

Taylor, R. H., Ainsworth, J. R., Evans, A. R., & Levin, A. V. (1999). The epidemiology of pediatric glaucoma: The Toronto experience. Journal of the American Association for Pediatric Ophthalmology and Strabismus, 3(5), 308-315. https://doi.org/10.1016/S1091-8531(99)70028-5