OPEN ACCESS Freely available online

ISSN: 2684-1622
Journal of Eye Diseases and Disorders

Commentary

Advances in Treatment for Congenital Glaucoma in Brittle Cornea Syndrome

Pati Bergmanson*

Department of Ophthalmology, Johns Hopkins University, Baltimore, USA

DESCRIPTION

Brittle Cornea Syndrome (BCS) is a rare genetic disorder primarily affecting the cornea, leading to its fragility and thinning. Among the ocular complications associated with BCS, congenital glaucoma is one of the most severe and sight-threatening. This form of glaucoma, present at birth, can cause increased Intraocular Pressure (IOP), which may eventually result in optic nerve damage and visual impairment if left untreated. Understanding congenital glaucoma within the context of BCS is essential for developing effective treatment strategies, enhancing patient care and improving outcomes.

This article examines the mechanisms underlying congenital glaucoma in individuals with BCS, clinical presentation, diagnostic approaches, current treatments and ongoing research efforts to mitigate the condition. Brittle Cornea Syndrome is linked to mutations in the *ZNF469* and *PRDM5* genes, which play important roles in collagen synthesis and extracellular matrix integrity. These mutations lead to a weakening of the corneal structure, making it more susceptible to ruptures and tears even with minor trauma. The cornea becomes abnormally thin and patients often develop corneal ectasia, a condition where the cornea bulges outward due to the lack of structural support. Congenital glaucoma, a frequent complication in BCS, is thought to arise from structural abnormalities in the anterior segment of the eye, including the trabecular meshwork and Schlemm's canal, which are responsible for regulating aqueous humor outflow. Impaired drainage of aqueous humor leads to increased IOP, putting pressure on the optic nerve and if untreated, causing irreversible damage. The connection between congenital glaucoma and BCS is complex. The trabecular meshwork and the outflow channels, essential in maintaining normal intraocular pressure, may be underdeveloped or functionally compromised in individuals with BCS. The structural weakness of the cornea also extends to other parts of the anterior segment, which might explain the frequent occurrence of congenital glaucoma in these patients.

Several studies suggest that mutations in the ZNF469 and PRDM5 genes disrupt the normal development of ocular tissues, resulting in compromised aqueous humor dynamics. In addition, the sclera, which shares structural components with the cornea, may also be thinner and less rigid in individuals with BCS. This contributes to the difficulties in maintaining the pressure balance within the eye, ultimately leading to increased IOP and optic nerve damage. Children with congenital glaucoma often present with classic signs, including enlarged eyes (buphthalmos), corneal edema, excessive tearing (epiphora) and photophobia (sensitivity to light). However, in the context of BCS, these symptoms may be exacerbated due to the underlying fragility of the cornea. Early recognition of these symptoms is vital, as prompt intervention can prevent permanent vision loss.

Corneal thinning and ruptures, which are hallmarks of BCS, complicate the clinical course of congenital glaucoma. Increased IOP may lead to additional stress on an already weakened cornea, raising the risk of spontaneous corneal perforation. Therefore, managing congenital glaucoma in BCS patients requires a delicate balance between controlling intraocular pressure and preserving the structural integrity of the eye. Diagnosing congenital glaucoma in BCS requires a comprehensive ocular examination that includes measurement of intraocular pressure, assessment of the optic nerve head and detailed evaluation of the anterior segment. Gonioscopy, a technique used to visualize the drainage angle, may reveal developmental abnormalities in the trabecular meshwork that impede aqueous humor outflow.

Imaging techniques such as Optical Coherence Tomography (OCT) and Ultrasound Biomicroscopy (UBM) are often employed to assess the thickness and structure of the cornea and other ocular tissues. These imaging modalities can provide valuable information about the extent of corneal thinning and the integrity of the anterior segment, guiding treatment decisions.

In addition to ocular evaluations, genetic testing is an important component of the diagnostic process for individuals with BCS. Identifying mutations in the *ZNF469* or *PRDM5* genes can

Correspondence to: Pati Bergmanson, Department of Opthalmology, Johns Hopkins University, Baltimore, USA, E-mail: pati@bergmanson.it

Received: 26-Aug-2024, Manuscript No. JEDD-24-27066; **Editor assigned:** 28-Aug-2024, PreQC No. JEDD-24-27066 (PQ); **Reviewed:** 11-Sep-2024, QC No. JEDD-24-27066; **Revised:** 18-Sep-2024, Manuscript No. JEDD-24-27066 (R); **Published:** 25-Sep-2024, DOI: 10.35248/2684-1622.24.9.247

Citation: Bergmanson P (2024). Advances in Treatment for Congenital Glaucoma in Brittle Cornea Syndrome. J Eye Dis Disord. 9:247.

confirm the diagnosis of BCS and provide insight into the risk of developing congenital glaucoma. Genetic counseling may also be beneficial for affected families, allowing them to understand the inheritance pattern and potential risks for future offspring. The management of congenital glaucoma in BCS presents significant challenges due to the fragility of the cornea and the potential for complications arising from increased intraocular pressure. Treatment typically involves a combination of medical and surgical interventions aimed at reducing IOP and preserving vision while minimizing the risk of corneal damage.

Initial treatment for congenital glaucoma often involves the use of topical medications, such as beta-blockers, prostaglandin analogs and carbonic anhydrase inhibitors, to lower intraocular pressure. However, the effectiveness of these medications may be limited in BCS patients and their use requires close monitoring for potential side effects. Systemic medications, such as oral carbonic anhydrase inhibitors, may also be employed in cases where topical therapy is insufficient. However, medical management alone is rarely sufficient to control IOP in the long term, especially in patients with structural abnormalities of the anterior segment. Surgery is often necessary for congenital glaucoma, particularly when medical therapy fails to adequately control IOP. However, surgical interventions in BCS patients must be approached with caution due to the increased risk of corneal perforation and other complications.