Handle with Caution: Scleral Lenses for Brittle Cornea Syndrome

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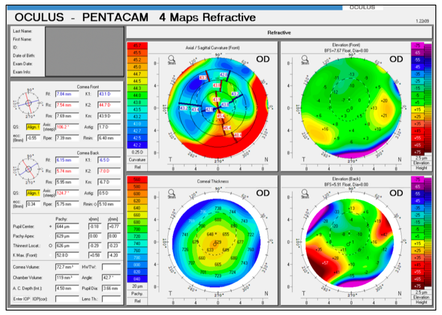
BACKGROUND

Brittle Cornea Syndrome is a rare autosomal recessive form of Ehlers-Danlos syndrome. The prevalence of brittle cornea syndrome is less than 1 in 1,000,000. Ehlers-Danlos syndrome is a group of 13 genetic disorders that affect the body's connective tissue primarily the skin, joints and blood vessels. Individuals with Ehlers-Danlos may present with skin hyper-extensibility, tissue fragility, and generalized joint hypermobility. It is caused by genetic mutation that affects the function and structure of collagen. Since collagen is the basis of many connective tissues, Ehlers-Danlos can affect multiple organ systems. Manifestations of Ehlers-Danlos include: thin cornea (less than 400 um), corneal scarring, blue sclera, progressive keratoglobus, high myopia, retinal detachment, deafness, developmental dysplasia of the hip, scoliosis, and small joint hypermobility.

There are two types of Brittle Cornea Syndrome. Type 1 is caused by mutation in the ZNF469 gene, which is primarily responsible for the regulation of genes involved in production and organization of collagen fibers. Type 2 is caused by mutation in the PRDM5 gene, which is a gene regulator required for synthesis and maintenance of collagen fibers. As a result of this genetic mutation the most common clinical presentation of Brittle Cornea Syndrome is corneal thinning. Individuals with Brittle Cornea Syndrome are at high risk for corneal rupture. Most patients present with greatly reduced visual acuity and excessive corneal thinning. There is no current treatment for this condition. Ocular management includes: monitoring for progressive vision loss, corrective spectacle lenses, and pachymetry to prevent corneal rupture. Another viable management option is scleral lenses.

CASE HISTORY

A 10-year-old Asian female presents for an initial evaluation for contact lens in the setting of Brittle Cornea Syndrome. As a review, she was referred by a cornea specialist for a contact lens fitting. Her ocular history is remarkable for Brittle Cornea Syndrome. Her thinnest pachymetry was 185/179 OD/OS. She underwent a corneal transplant in March 2021. Her transplant was a difficult case due to her thin peripheral cornea and posterior pressure. Following her corneal transplant, there was signs of graft rejection. She demonstrated diffuse edema and inferior keratic precipitates. This caused a reduction in her visual acuities. She was counting fingers in both eyes at this point. She was treated with Brimonidine BID, Timolol BID and Pred Forte Q3H. After additional follow up visits, her vision stabilized to 20/200 OD and CF OS. She was referred to the Eye Institute for a refraction and contact lens fitting. It is notable that the patient's medical history is remarkable for premature birth. On physical examination, the patient also has long slender fingers. Additionally, there is also a positive family history of Brittle Cornea Syndrome through the patient's younger brother as well. She presented with progressive decline in vision OS>OD. She has been wearing spectacle glasses for many years. She presented as a first time contact lens wearer and was fit in scleral lenses using hyper Dk Optimum Infinite material.



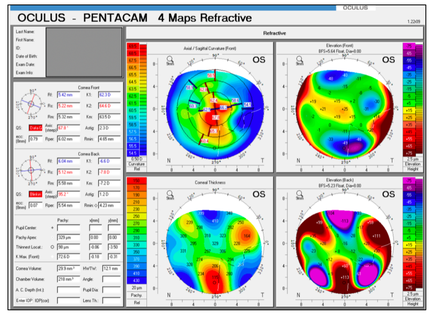


Figure 1 & 2 demonstrates Pentacam topography scans show oblate shape s/p PKP OD (left image) and central steepening OS (right image).

CASE FINDINGS

INITIAL VISIT		
SLIT LAMP FINDINGS	OD: blue hue sclera, superior pannus, ectasia, resolved KPs, PAS with small iris cyst at 5 o'clock	OS: blue hue sclera, ectasia, vertical anterior and mid-stromal haze
LENS PARAMETERS	OD: CS Elite 16.8 Sag: 6.00 mm BC 49.00/6.89 mm Pwr: -8.0D Dia: 16.8 mm C.T 400 um S.L 6.00/-4.0 OR: plano	OS: CS Elite 16.8 Sag: 6.00 mm BC 49.00/6.89 mm Pwr: -8.0D Dia: 16.8 mm C.T 400 um S.L 6.00/-4.0 OR: plano
PLAN	Increase diameter OS to avoid lens touch at superior nasal limbus. Steepen peripheral curves by 60 microns to reduce edge lift and lens movement OU. No lens were dispensed a this visit. Trial lenses were order	
Patient present with spectacle glasses correction. Entering visual acuity were 20/200 OD and CF OS		
SLIT LAMP FINDINGS	OD: blue hue sclera, superior pannus, ectasia, resolved KPs, PAS with small iris cyst at 5 o'clock	OS: blue hue sclera, ectasia
LENS PARAMETERS	OD: Optimum Infinite CS Elite Sag: 6.00 mm BC 49.00/6.89 mm Pwr: -8.0D Dia: 16.8 mm C.T 400 um S.L 4.00/-6.0 OR: plano	OS: Optimum Infinite CS Elite Sag: 6.00 mm BC 49.00/6.89 mm Pwr: -8.0D Dia: 17.8 mm C.T 400 um S.L 4.00/-6.0 OR: plano
PLAN	**Plan:** Steepen Flat Meridian 60 microns to improve comfort, increase limbal clearance by 100 microns OU. Initial lens dispensed at this visit.	
FOLLOW UP #2: Patient present with trial contact lenses. Entering visual acuity: 20/125 OD, 20/400 OS.		
LENS PARAMETERS	OD: Optimum Infinite CS Elite Sag: 6.00 mm BC 49.00/6.89 mm Pwr: -8.0D Dia: 16.8 mm C.T 400 um S.L 2.0/-4.0 OR: plano	OS: Optimum Infinite CS Elite Sag: 6.00 mm BC 49.00/6.89 mm Pwr: -8.0D Dia: 17.8 mm C.T 400 um S.L 2.0/-4.0 OR: plano
PLAN	**Plan:** Adequate fit. Scleral lens dispensed. Follow up in 2-3 weeks after wearing lenses for 3-4 hours.	
FOLLOW UP #3: Patient present with trial contact lenses OD only. She could not get the lenses in OS and lost the lenses before the appointment. Her father states that she occasionally has difficulty getting the lenses in. Entering visual acuity were 20/125 OD and CF OS		
LENS PARAMETERS	OD: Optimum Infinite CS Elite Sag: 6.00 mm BC 49.00/6.89 mm Pwr: -8.0D Dia: 16.8 mm C.T 400 um S.L 2.0/-4.0 OR: plano	OS: Optimum Infinite CS Elite Sag: 6.00 mm BC 49.00/6.89 mm Pwr: -8.0D Dia: 17.8 mm C.T 400 um S.L 2.0/-4.0 OR: plano
LENS EVALUATION	200 microns clearance centrally (-) limbal touch, no touch at GHJ and no edge lift or blanching.	300 microns central clearance (-) limbal touch, no edge lift or blanching.
PLAN	**Plan:** Adequate fit. Patient to return for re-evaluation in 6 months. In the meantime, continue ocular care at Will's Eye for cornea and glaucoma monitoring.	

TREATMENT & MANAGEMENT

Brittle Cornea Syndrome is a complex and progressive disorder and given its rarity. There is no current treatment for this condition; rather, it is recommended that early detection or diagnosis is pertinent to ensure proper measures are taken to prevent corneal perforation. Additionally, the condition can be managed through various interventions. These include the following: protective eyewear, routine comprehensive eye exam and specialty contact lenses. Surgical management options include corneal transplant and corneal crosslinking. Patients with brittle cornea syndrome should use protective polycarbonate glasses and avoid contact sports. These measures will reduce risk of corneal rupture in the event of accidental injury. Routine comprehensive eye exams are important to monitor changes in corneal thickness or visual acuity. Specialty contact lenses such as scleral lenses can assist in the visual and ocular challenges of brittle cornea syndrome. A corneal transplant is indicated in severe cases. However, given the fragile nature of the cornea in these patient's, corneal transplants are challenging and there is also a high risk of graft rejection in brittle cornea syndrome patients. In addition, corneal transplants may only help reinforce peripheral cornea with little to no effect on improvement of visual acuity. Collagen cross-linking has had promising results with keratoconus, a corneal ectasia. A modified corneal crosslinking method has been recently trialed as a treatment option for brittle cornea syndrome; though, there have been varied outcomes. Therefore, collagen crosslinking remains a controversial management option for brittle cornea syndrome.

CONCLUSION

The patient presented in case presentation exhibited many characteristics of Brittle Cornea Syndrome such as corneal ectasia, blue sclera and corneal scaring. Given this case presentation, it is important to protect her cornea and reduce the risk of rupture. Scleral lenses are an ideal management option for patients with Brittle Cornea Syndrome. With the use of scleral lenses, she was able appreciate mildly improved visual acuity and comfort. Due to the highly customizable nature of scleral lenses, it is a great tool in cases with complex and irregular corneas. Scleral lenses provide stability, comfort, and improved vision. Due to the large diameter of a scleral lenses, it is able to vault over the cornea and provide protection. It does this by shielding the cornea from external environmental factors and reduce the likelihood of corneal perforation. Additionally, scleral are also often used in patients with corneal ectasias to improve visual acuity by providing a smooth optical surface between the lens and corneal reservoir. As Brittle Cornea Syndrome is a rare progressive disease with potential devastating outcomes, it is important look for signs and symptoms of this disorder to ensure early detection and treatment. It is also equally pertinent to understand and utilize viable management options such as scleral lenses to prevent corneal rupture and improve the prognosis of this condition.