CASE REPORT

Unusual case of globe perforation: the brittle cornea without systemic manifestations

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SUMMARY

Brittle cornea syndrome is a rare generalised connective tissue disorder with ocular features like keratoglobus or keratoconus, severe corneal thinning and a high risk of perforation. Various authors in different case reports and case series have brought out the fact that brittle cornea is a disorder with characteristic systemic manifestations such as deafness, joint hypermobility, hyperelasticity of skin, kyphoscoliosis and dental abnormalities along with ophthalmic features. We report a case of globe perforation following trivial trauma, in an individual with brittle cornea without any extraocular manifestations, posing a challenge in the diagnosis and dilemma in surgical repair of cornea, restoration of globe integrity and visual rehabilitation. The absence of systemic manifestations decreased the index of suspicion and led to a surprise in the theatre—a point this case emphasised.

BACKGROUND

Corneal tears and lacerations represent a significant percentage of all cases of traumatic ocular injuries. Usually, the primary management of these cases is by suturing of the tear with additional procedures like cortical wash, AC reformation, etc if needed. However, in the case that we report below, suturing for restoration of globe integrity was not possible due to extreme fragility of the cornea. In the absence of any systemic manifestations, especially musculo-skeletal abnormalities, the diagnosis and management of this case posed a challenge and several surgical procedures were required to salvage the eye.

CASE PRESENTATION

A girl aged 15 years was presented in the emergency room of our hospital. The girl had a history of sudden diminution of vision, pain and redness in the right eye following a blunt trauma with a baby's foot while playing. She gave a similar history of loss of vision in the left eye following trivial trauma 5 years. Her previous medical records showed corneal laceration in the left eye with no evidence of infectious keratitis. She had undergone corneal tear suturing in the left eye at a local ophthalmic hospital. Following surgery, eye was quiet, although there was no improvement of vision in the left eye and the patient gave history of gradual reduction in the size of that eye.

On examination, her uncorrected visual acuity was hand movements in the right eye and was found to have perception of light with inaccurate projection of rays in the left eye. On slit-lamp examination, the right eye showed a full thickness

Figure 1 (A) OD—preoperation, diagrammatic presentation of corneal tear. (B) OD—preoperation, diagrammatic presentation of corneal tear—optical section. OD, right eye.

paracentral corneal tear extending vertically from 6:30 to 12:00 clock position with iris incarceration in the wound, exudative membrane over iris, flat anterior chamber and grossly hypotonous globe. There was no evidence of infection in the wound. Sclera did not appear blue. The left eye was phthisical with total corneal opacity and presence of few corneal sutures suggestive of previous surgical intervention (figures 1A, B, 2 and 3). The patient was posted for primary repair of corneal tear after

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Figure 2 OD—postkeratoprosthesis, OS—phthisical eye. OD, right eye; OS, left eye.

Figure 3 OS—corneal scarring postcorneal suturing. OS, left eye.

doing haemogram and checking fitness for general anaesthesia. Since the patient presented in an emergency setup and was immediately taken up for corneal tear repair, preoperative clinical photo could not be taken. However, schematic presentation of corneal tear has been shown in [figure 1A, B](#).

INVESTIGATIONS

Detailed history, ocular and systemic examination was undertaken, after the primary surgery. The patient was of south Asian origin. Old case records showed she was diagnosed with keratoconus in both eyes, few years earlier, but topography or pachymetry was not performed. The parents gave history of consanguineous marriage, but no history of similar corneal ectatic condition in sibling. The parents and two younger siblings aged 14 and 11 were examined. Both parents were emmetropic. The siblings had moderate myopia and best corrected visual acuity of 20/20 with glasses in both eyes. On examination, the parents and siblings did not show any signs of keratoconus, their corneal topography and pachymetry were within normal limits.

Directed medical history of patient revealed no history of hearing loss, frail skin, excessive skin scarring or laxity of joints. Examination revealed a well-nourished female with normal vital signs. She had black hair and her skin had unremarkable texture, thickness, elasticity with no evidence of excessive scarring. Orthopaedic evaluation showed no skeletal deformity and no dysmorphism of the face, hands, feet or back. There was no scoliosis or any hypermobility of joint (Beighton score 2/9).¹ Oral examination revealed poor dental hygiene. The patient was investigated for RF, ANA, liver function test, kidney function

test and urine analysis to rule out any connective tissue disorder, which were reported normal.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis of non-accidental injury (NAI) in the paediatric population needs to be borne in mind when such a clinical picture is encountered. Conversely, patients with brittle cornea syndrome (BCS) and their families are at great risk of a mistaken diagnosis and it is very important to stress that this condition, although rare, does occur and may present in the absence of systemic features. Since NAI and BCS are in the differential diagnosis: all investigations, including genetic testing for the diagnosis of BCS, must be carried out without delay to prevent wrongful conviction!

TREATMENT

The patient was undertaken for corneal tear repair in the right eye under general anaesthesia. Intraoperatively, total iridocorneal touch with choroidal bulge alongwith cortical matter and exudates in the wound was noted. There was failure to approximate extremely thin and friable edges of corneal wound. Restoring anatomical integrity by suturing corneal laceration was not possible in this patient; therefore, it was decided to perform a primary tectonic penetrating keratoplasty. A 9 mm host tissue and 9.5 mm donor graft was trephined; however, the remaining thinned out fragile host cornea torn off completely when suturing of donor button was attempted. Finally, a 12 mm sclerocorneal graft was trephined, iris was cleared of membranes, cortex in the wound was removed and graft was sutured to sclera.

OUTCOME AND FOLLOW-UP

On first day postoperatively, the patient had vision of CF1 m in the right eye; slit-lamp examination showed intact sutures, well opposed Graft-Host junction and graft clarity of grade 3 with few Descemet membrane folds, formed anterior chamber and aphakia ([figure 4](#)). On follow-up, the patient maintained an ambulatory vision until 15 months later, when she was diagnosed with failed sclerocorneal graft in the right eye and phthisis bulbi in the left eye. A decision was made to perform optical penetrating keratoplasty in the right eye after a thorough discussion with the patient and her parents. Seven millimetre central grafting was performed; however, intraoperatively, the patient was left aphakic as there was loss of posterior capsular integrity and broken anterior vitreous phase.

Figure 4 OD—postprimary tectonic keratoplasty. OD, right eye.

Figure 5 OD—postfailed optical penetrating keratoplasty. OD, right eye.

Figure 6 OD—postkeratoprosthesis. OD, right eye.

On subsequent follow-up, the patient was diagnosed with dry eye for which she was medically managed and later on underwent punctal occlusion with Herrick's canalicular plug. She developed dry ocular surface and a failed regrant in the right eye another 10 months later (figure 5). After a thorough discussion with the patient and her parents, she underwent keratoprosthesis implantation (Auro K-pro, Boston Type 1-based, Aurolabs, Arvind Eye Care) and lateral tarsorrhaphy in the right eye. The patient is being regularly followed up for last 3 years for vision, intraocular pressure (IOP), status of K pro, retroprosthetic membrane and any evidence of endophthalmitis or vitritis. At her last follow-up visit, visual acuity in her right eye was 20/80. Further, slit-lamp examination showed in situ keratoprosthesis with no evidence of infective or sterile stromal melt, no retroprosthetic membrane, normal IOP and normal disc and central fundus (figure 6).

DISCUSSION

Corneal trauma is an important cause of significant ocular morbidity and visual difficulty among ocular trauma patients. Corneal lacerations and perforations account for 6.8–14.7% of all ocular traumatic injuries.^{2–4} Primary treatment is by suturing, which is required for restoration of globe integrity, if not, for visual rehabilitation. Very rarely, cornea surgeons may come across a situation like a brittle cornea where wound suturing is not possible.

BCS is an autosomal recessive generalised connective tissue disorder characterised by corneal perforation, either after a

minor trauma or spontaneously.⁵ There are two types of BCS; BCS type 1 is caused by mutations in the ZNF469 gene and BCS type 2 is caused by mutations in the PRDM5 gene. Both the genes have a regulatory and a structural role in the assembly of collagen fibrils. Mutation results in corneas which are unable to bear normal biomechanical stress and hence cannot maintain their structural integrity, thus leading to corneal rupture. Other classical ophthalmological features include extreme corneal thinning (220–450 µm), irregular corneal astigmatism, high myopia, blue sclera, keratoglobus or keratoconus and retinal detachment. Additionally, there is generalised connective tissue damage with extraocular dysfunctions, including auditory, skin and musculo-skeletal features.⁶

BCS, in the literature, has been described in association with systemic mesodermal disorders like osteogenesis imperfecta, Marfans syndrome and Ehler Danlos syndrome. However, recently BCS has been identified as an entity different from the kyphoscoliotic type of EDS, which has a much poorer prognosis.⁷ In BCS, multiple tissue involvement can usually be demonstrated; but, recently, a patient of BCS with an ocular-only phenotype has been identified.⁸ This presentation is in line with our case suggestive of ocular only phenotype of BCS. Genetic studies would have been interesting to perform in this patient but was not feasible because of lack of funds. Simple surgical repair of corneal rupture in BCS is challenging owing to extremely thin cornea. Various alternative methods for repair of corneal perforations have been reported like using tissue adhesives and viscoelastic agents,⁹ corneal sealing by a combination of sutures with application of bandage contact lens, pressure patches and systemic carbonic anhydrase inhibitors,¹⁰ onlay epikeratoplasty with a donor corneoscleral button,¹¹ and intraocular gas C3F8 tamponade.¹² Cases where primary repair has been attempted, complication due to scarring have been noticed. When primary repair of corneal perforation is not possible, or fails, affected individuals usually require an evisceration.¹³

Primary penetrating keratoplasty is a good option to restore ocular integrity in patients with corneal perforations due to BCS, especially when tissue loss precludes simple repair as reported by Natarajan *et al.*¹⁴ In our patient too, suturing the corneal tear as a primary procedure was not possible due to pre-existing corneal pathology (brittle cornea). Hence, management of our case was drastically different from routine corneal tear repair. We performed primary tectonic penetrating keratoplasty as globe-saving procedure. Since it was a large sclerocorneal graft, eventually there was graft rejection and graft failure. Invariably, fate of most large diameter grafts(>=8 mm) is rejection and failure as reported by Collaborative Corneal Transplantation Studies Research Group and various other studies, requiring a second penetrating keratoplasty for visual rehabilitation.^{15–18} In our case too, an optical penetrating keratoplasty and finally type I keratoprosthesis was required as this was only eye with visual potential.

With this case report, we have attempted to highlight the need of keeping BCS, though very rare, as a cause of corneal perforation in mind so as to aid us in emergent management to salvage the eye. The diagnosis and management of these cases is difficult irrespective of the presence or absence of systemic features. Examination of family members and siblings can help in diagnosis, especially if facilities for genetic testing are lacking. Lifestyle counselling of family, use of polycarbonate reinforced spectacles in affected children, avoidance of contact sports may prevent globe perforation. Referral to clinical geneticist and genetic counselling may be required. In patients of corneal perforation with BCS, visual rehabilitation is a long drawn process requiring multiple surgeries as was evident in the present case.

Learning points

- ▶ Corneal rupture after a relatively trivial trauma can be the sole presenting feature in some cases of keratoconus/keratoglobus with brittle cornea.
- ▶ A high index of suspicion is required to diagnose brittle cornea in patients without systemic features. It needs to be differentiated from non-accidental injury.
- ▶ Lifestyle counselling of family and genetic counselling may be required to prevent globe perforation.
- ▶ Surgical closure of corneal perforation in brittle cornea is challenging as conventional corneal wound suturing is not possible because of extreme fragility leading to 'a surprise in operation theatre' and management dilemma.
- ▶ Primary penetrating keratoplasty is a good option to restore globe integrity when tissue loss precludes simple repair.

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