Sequential traumatic corneal open globe rupture in a patient with osteogenesis imperfecta type I

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\section*{ARTICLE INFO}

\textbf{Keywords:}
Osteogenesis imperfecta
Trauma
Open globe

\section*{ABSTRACT}

\textbf{Purpose:} To report a case of sequential open globe rupture in a young patient with osteogenesis imperfecta type I following minor accidental blunt injury. This represented the patient's sole clinical manifestation of connective tissue disease, leading to a diagnosis of osteogenesis imperfecta type I at the age of 12 years old.

\textbf{Observations:} A 12-year-old male presented with right eye pain following accidental blunt trauma at school while wearing protective lenses. One year ago, he required surgical repair of a left open globe following blunt trauma during a middle school basketball game. His exam was significant for a full-thickness corneal laceration, necessitating open globe repair of his right eye, which was remarkably difficult given the poor tissue constitution of the cornea and sclera. He was referred to a genetics specialist, where he was found to have a pathogenic heterozygous splice site variant in the \textit{COL1A1} gene, consistent with osteogenesis imperfecta type I.

\textbf{Conclusions and importance:} Connective tissue disease should be considered in any case of open globe rupture following minor trauma, even in the absence of other clinical manifestations of the disease. The surgical management of these patients is particularly challenging due to the fragility of the connective tissue. Early diagnosis of connective tissue disease is important to preserve visual acuity and prevent further damage to the eyes.

\section*{1. Introduction}

Osteogenesis imperfecta (OI) is a rare, inherited, connective tissue disorder (CTD) caused by mutations in genes that encode type I collagen, a critical structural protein found in the cornea and sclera.\textsuperscript{1} Some CTDs such as Ehlers-Danlos syndrome (EDS) confer an increased risk of globe rupture,\textsuperscript{2} but there are very few reported instances of open globe injury in patients with OI.\textsuperscript{3-5} We report a case of sequential traumatic open globes in a patient ultimately diagnosed with OI type I.

\section*{2. Case report}

A 12-year-old boy presented with right eye (OD) pain and reduced acuity following minor trauma. His ocular history was significant for high myopia in both eyes with an eyeglasses prescription of $-18.25 + 2.00 \times 180$ in the right eye and $-14.75 + 1.50 \times 070$ in the left eye. In 2016, he experienced minor blunt trauma to the left eye (OS) while wearing glasses during a basketball game and required open globe repair (Fig. 1), resulting in functional monocularity OD. One year later, on this particular occasion, a classmate accidentally elbowed his face, pressing his spectacles against his right eye. Medical history revealed joint hypermobility and prior tibia fracture following a motor vehicle accident (MVA). Family history was negative for ophthalmic and musculoskeletal conditions, and the patient’s parents and three sisters were healthy.

Slit-lamp examination of the right eye showed a semicircular full-thickness corneal laceration from 10 to 4 o’clock inside the limbus with iris prolapse. The anterior chamber was flat, but the lens appeared intact. He did not have blue or tinted sclera. He underwent emergent repair of the ruptured cornea under general anesthesia. Obtaining water-tight closure of the corneal wound rupture was remarkably difficult given the poor tissue constitution of the cornea and sclera, requiring twelve 10-0 interrupted nylon sutures and glue. Over-tightening the suture caused the nylon to cut through the intact corneal tissue. Similar difficulty with primary closure occurred previously in the left eye.

He was known to the genetics service and had been seen in genetics clinic a few years prior for joint hypermobility, but his clinical examination at the time did not prompt further genetic testing. Following the second open globe repair, he was again referred for genetics.
consultation, and a pathogenic heterozygous splice site variant in the COL1A1 gene was identified, consistent with OI type I. Gene testing for kyphoscoliotic EDS was negative. His postoperative course has been complicated by significant corneal thinning in the right eye (Fig. 2).

3. Discussion

In patients with traumatic corneal rupture after minor trauma, especially in both eyes, EDS, OI, and brittle cornea syndrome should be considered. This patient experienced a previous tibia fracture in the setting of MVA but otherwise did not depict a typical clinical portrayal of OI. The extent of this patient’s ocular phenotype with OI is unique, and to our knowledge, cases of open globe injury in patients with OI have been reported merely three times in the literature.3–5

Patients with OI show reduced thickness of the corneal and scleral collagen fibers,6 resulting in low ocular rigidity. Thin corneas, small corneal diameters, and keratoconus are also associated with OI,3 but corneal rupture is an unusual occurrence. Blue sclera is a common finding in OI, as the thin scleral collagen reveals the darker underlying choroid. However, this patient did not have blue sclera. The only ophthalmic finding suggestive of connective tissue disease was high myopia in both eyes, which is associated with thin sclera.7 There is evidence that immature collagen fibers are particularly slender and less densely packed in the episclera and superficial cornea near the limbus,8 which may explain the curvilinear nature of the open globe rupture in this patient and in previous reports.3–5 While the sample of similar cases is quite small, curvilinear open globe rupture may warrant genetic testing, even in the absence of other signs of connective tissue disease, which is evidenced by this case report. The surgical management in this case is consistent with previous reports.3 However, one may need to be cautious grafting normal tissue against abnormal corneal host tissue interface since sutures may cut through the host tissue interface making it difficult to seal that surgical interface. The exquisite fragility of this patient’s sclera was particularly astonishing, as even gentle manipulation with forceps teased the tissue apart.

In contrast to other cases,3–5 this patient was not known or highly suspected to have OI prior to corneal rupture in either eye. This report contributes to the very limited but growing body of literature of ocular trauma in patients with OI and reinforces the importance of maintaining a high degree of clinical suspicion for CTDs in patients with globe rupture following minor trauma or whenever the integrity of the cornea seems unusual during repair, particularly in a child. Use of protective eyewear to guard against potential permanent vision loss in any patient with a clinical suspicion of underlying CTD cannot be overstated.

Patient consent

Consent to publish the case report was not obtained. This report does not contain any personal information that could lead to the identification of the patient.

Acknowledgements and disclosures

Funding: No funding or grant support.

Conflicts of Interest: The following authors have no financial disclosures: GC, ZA, SK, EC.

Authorship: All authors attest that they meet the current ICMJE criteria for Authorship.

Acknowledgements: None.

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