CASE REPORT

Recurrent spontaneous scleral rupture in Marfan's syndrome

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Accepted 2 May 2016

SUMMARY

The ocular manifestations of Marfan's syndrome (MS) range from ectopia lentis, microspherophakia, myopia, glaucoma and retinal detachment. Spontaneous scleral rupture is a rare complication and recurrent scleral perforation is extremely rare. We report a rare case of a 26-year-old male with MS who had sequential recurrent spontaneous scleral rupture which required surgical repair. He suffered from a similar problem 4 years later in both eyes in a different location, with overlying thin cystic blebs and hypotony maculopathy. Surgical repair with preserved scleral donor patch graft and conjunctival autograft in one eye, and conjunctival advancement in the other eye was performed. This helped stabilise the eyes, and resulted in complete visual recovery in both eyes.

BACKGROUND

Marfan's syndrome (MS) is an inherited disorder of the connective tissue characterised by musculoskeletal abnormalities, cardiovascular disease and ocular abnormalities.¹ The ocular manifestations of MS include ectopia lentis, microspherophakia, myopia, glaucoma, retinal tears and retinal detachment.¹ The sclera of patients with MS is unusually thin. Scleral thinning and perforation may occur after trauma or surgery or due to chronic increase in intraocular pressure (IOP).^{2–3} Spontaneous perforation due to progressive scleral thinning, although rare, can occur in these patients' eyes.^{4–5} Recurrent bilateral spontaneous scleral rupture in the absence of high IOP is even more rare, and is not reported so far. We report a case of bilateral recurrent spontaneous scleral rupture with hypotony maculopathy in a young man with no glaucoma, which required surgical repair. Our case highlights a rare and sight-threatening ocular complication of MS, and an unusual scleral phenotype.

CASE PRESENTATION

A 14-year-old boy presented to us, in June 2004, with symptoms of decreased vision in the left eye (LE) since 1 month. He had no history of trauma or previous ocular surgery. His visual acuity was 20/60 in both eyes, with myopia of −19D. The IOP was 8 mm Hg in the right eye (RE) and 2 mm Hg in the LE. Anterior segment examination showed clear cornea, shallow anterior chamber, corectopia, microspherophakia, inferiorly subluxated clear lens, superior scleral thinning with underlying uveal show noted at 10–11 o'clock in the RE and a scleral defect with a bleb from 9 to 12 o'clock in the LE. Fundus evaluation was

normal in both eyes. General examination showed features suggestive of MS such as long thin face, high arched palate, tall stature with long extremities and arm span greater than the total height. His cardiovascular examination revealed mitral valve prolapse with mild regurgitation. Surgical repair of the scleral defect in the LE was performed with preserved donor scleral patch graft under general anaesthesia (GA). His postoperative course was uneventful with IOP of 12 and 10 mm Hg in the RE and LE, respectively. He had stable visual acuity and was under our regular follow-up care.

Four years later, in February 2008, he presented to the emergency clinic with symptoms of sudden decrease in vision, pain and redness in the LE of 2 days duration. His best corrected visual acuity (BCVA) was 20/60 in RE and 20/100 in LE. The IOP was 8 mm Hg in the RE and 6 mm Hg in the LE. LE examination revealed dislocation of crystalline lens into the anterior chamber. An emergency pars plana lensectomy (PPL) and anterior vitrectomy was performed under GA, and his postoperative course was uneventful. His visual acuity improved to 20/60 with aphakic correction in the LE. The IOP was 12 mm Hg in both eyes.

Six weeks after this episode, he presented in March 2008 with sudden decrease in vision in the RE. BCVA in the RE was finger counting at 2 m and IOP was unrecordable. On examination, anterior chamber was very shallow with subluxated lens, and a scleral defect was noted at 10–12 o'clock close to the limbus in the area of previous thinning, with overlying conjunctival bleb. Emergency surgical repair with preserved donor scleral patch graft and PPL were performed in the RE under GA. He had an uneventful postoperative period, and was visually rehabilitated with soft contact lens in both the eyes.

Seven years after his first presentation, he presented with sudden decreased vision in his LE. His visual acuity was 20/40 in RE and 20/400 in LE. The conjunctiva showed chemosis with cystic elevation noted in the superior conjunctiva, nasally and temporally in both eyes. Seidel's test was negative, cornea was clear in the RE and showed descemet's folds in the LE, anterior chambers were deep and quiet, pupils were irregular and there was patent surgical iridectomy in the RE. Both eyes were aphakic (figure 1). IOP was 6 mm Hg in the RE and 0 mm Hg in the LE. Fundus examination of RE was normal, and LE showed attached retina, disc oedema, dilated tortuous vessels and radial choroidal folds with hypotony maculopathy (figure 2).

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To cite: Turaga K, Senthil S, Jalali S. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2016-214764

Figure 1 Slit lamp photograph showing thin cystic conjunctival blebs with underlying sclera defect (arrow) in the right eye (OD) and left eye (OS).

INVESTIGATIONS

B-scan ultrasonography of RE was normal, and LE showed few low echo spike opacities in the vitreous cavity; retina was attached, and a thickened choroid and normal optic nerve shadow was noted. This ruled out choroidal/retinal detachment. The central corneal thickness was 540 μm in the RE and 533 μm in the LE.

DIFFERENTIAL DIAGNOSIS

1. Glaucoma filtration surgery is one of the differential diagnosis for cystic blebs. However, our patient did not have any history of previous intraocular surgery, and there were no other clinical signs suggestive of previous trabeculectomy.
2. Retinal or choroidal detachment with hypotony is a possibility; this, however, was ruled out with ultrasound B-scan.

TREATMENT

He was advised to undergo scleral patch graft with conjunctival autograft in LE under GA. The thin cystic conjunctiva was excised and the underlying scleral defect was repaired. Intraoperatively, the sclera was noted to be very thin with underlying uveal show. The sclera was frayed with abnormal scleral architecture, not just at the area of the rupture but also in the adjacent area. The scleral defect was more like a full thickness scleral hole (figure 3). There was no scleral tear noted. The initial patch was in a place adjacent to the new rupture. The abnormal and thin cystic conjunctiva was excised, and the scleral defect was closed with preserved donor scleral patch graft; fibrin glue (Tissel, Baxter, Westlake Village, California, USA) was used and then scleral patch graft edges were sutured with 10-0 nylon sutures to the adjacent sclera which was also thin and abnormal, but we were able to secure the graft. This was then covered with a free conjunctival autograft from the inferior bulbar conjunctiva and was secured with multiple interrupted 10-0 nylon sutures.

OUTCOME AND FOLLOW-UP

At the 1-week follow-up, vision was 20/40 in the RE and 20/160 in the LE. The anterior segment examination of LE showed oedematous lids, conjunctiva showed chemosis, patch

Figure 2 Fundus photograph of the left eye showing oedematous disc and hypotony maculopathy.

graft was in place and the sutures were intact, cornea showed stromal oedema and descemet's folds, anterior chamber was deep with 1+ cells, and the pupil was irregular and fixed. Both eyes were aphakic. IOP was 5 mm Hg in the RE and 6 mm Hg in the LE. He was advised to use topical steroids in tapering doses, topical antibiotics and cycloplegics in the LE. Fundus examination revealed resolving hypotony maculopathy in the LE. One month later, he presented with sudden decrease in vision in his RE and unrecordable IOP. There was a scleral defect noted superonasally, with thin cystic bleb and negative Seidel's test. He underwent surgical repair with scleral patch graft and conjunctival advancement. Vision was 20/25 with +12.00 Dsph/−2.00 Dcyl @ 180° in RE and 20/60 with +12.00 Dsph/−2.00 Dcyl @ 160° in LE. Near vision was N6 in RE and N24 in LE with addition of +3.25 Dsph in both eyes. The IOP recorded was 8 and 6 mm Hg in the RE and LE, respectively. Vision and IOP have remained stable for 3 years, with well-placed patch grafts and healthy conjunctiva (figure 4).

The scleral tissue was biopsied, and the conjunctival and scleral tissues were sent for histopathology. The histopathology of the scleral tissue showed disorganised collagen with focal hyalinisation suggesting either a connective tissue disorder or chronic inflammation. In the absence of inflammation in our case, the histopathology most probably represents a connective tissue disorder.

DISCUSSION

MS is a hereditary systemic connective tissue disorder characterised by musculoskeletal, cardiovascular and ocular abnormalities.¹ MS is caused by mutation of the FBN1 gene on chromosome 15, which encodes fibrillin1; it can also be caused by inactivation of mutation in TGF- β receptor 2 in 20% of the cases.¹ Transmission is autosomal dominant with high penetrance, and ~25–30% of cases are sporadic.¹

The physical features of patients with MS include musculoskeletal defects such as tall stature with long extremities, arm span greater than their total height, disproportionately long and thin fingers (arachnodactyly). Prognathism, high arched palate, kyphoscoliosis, pectus excavatum or pectus carinatum are common.⁶ A generalised muscular hypotony, joint laxity and joint contracture may be seen.⁶ Cardiovascular manifestations of MS include aortic and pulmonary artery dilation, as well as mitral and tricuspid valve prolapse with or without regurgitation.⁶

FBN1 is a main component of the extracellular microfibrils found in a wide range of tissues. It provides stretch and elasticity to connective tissues. Fibrillin, a glycoprotein, is a major structural component of the microfibrils that surround the elastin core in the extracellular matrix of the sclera.¹ These help maintain the rigidity, strength and elasticity of the scleral tissue. The gene defect causes disordered and decreased incorporation of fibrillin into the connective tissue matrix.¹

Ocular structures rich in fibrillin include cornea (at the level of epithelial basement membrane), the suspensory zonules of the lens, the lens capsule and the sclera.¹ Mechanical stretching of the sclera containing abnormal fibrillin molecule may result in axial myopia and progressive scleral thinning. The sclera of patients with MS is very thin, and predisposes to stretching and perforation. The aqueous leak from the scleral defect could lead to bleb formation and hypotony. Scleral perforation following trabeculectomy and scleral buckling procedure has been reported in MS.^{2,3} However, bilateral spontaneous scleral defect is rare and recurrent scleral perforation is even rare.

Figure 3 Intraoperative photograph of the left eye, showing thin cystic conjunctiva (black arrow) superonasally, with an underlying full thickness scleral defect (yellow arrow).

There have been two case reports of spontaneous scleral perforation published.^{4 5} In the first case report, there were two cases, a 11-year-old girl and 23-year-old male who presented with progressive myopia and decreased visual acuity.⁴ The IOP recorded were 11 and 3 mm Hg in RE and LE in case one, and in the second case, 9 mm Hg in both the eyes. There was scleral thinning noted in the superonasal quadrants with blebs in both eyes of the two patients, with upward subluxation of the spherophakic lens. Lensectomy and vitrectomy with iris-fixated intraocular lens implantation was done in both eyes. The scleral defects were not repaired and in one eye with IOP of 3 mm Hg, the IOP increased to 10 mm Hg after lensectomy.

In another similar case, a 18-year-old man with MS presented with bilateral red eye and tearing for 1 week.⁵ IOP in RE and LE was 9 and 8 mm Hg, respectively. In both eyes there was upward subluxation of spherophakic lens with shallow AC. There were areas of scleral thinning in the superior quadrants of both eyes, with exposed uveal tissue covered by a layer of thin conjunctival epithelium with no leak. Surgical repair with scleral patch graft was done in both eyes. Five months after scleral patch graft, vision improved in both the eyes with IOP of 13 and 10 mm Hg in RE and LE, respectively.

In our patient, the lens subluxation was inferior, which is unusual in a MS. The scleral defects were superonasal, as in all the other cases reported so far. The recurrence in the scleral thinning was adjacent to the previously treated area and not in the area of pars plana ports used for lensectomy. Also our patient presented with severe hypotony with maculopathy and had to be treated immediately. High index of suspicion is needed to detect ooze through thin sclera; treatment should be done in time and appropriately to avoid serious hypotony-related complications.

Our case also highlights that these patients need lifelong follow-up; they should be educated about the risk of scleral

thinning and perforation apart from other ocular problems, and the need to report immediately in case of sudden decrease in vision. The patients with suspected sclera thinning should also be advised to avoid contact sports, which could increase the risk of scleral rupture. Prompt surgical repair with preserved donor sclera patch graft and appropriate conjunctival closure technique can preserve the vision and the integrity of the globe.

Learning points

- ▶ Recurrent spontaneous scleral rupture is an extremely rare presentation of Marfan's syndrome, and most often occurs in the superonasal quadrant.
- ▶ Marfan's patients need lifelong follow-up, and should be made aware of the risk of scleral perforation and hypotony.
- ▶ Severe hypotony with maculopathy can pose a threat to vision in these cases, and prompt surgical repair can preserve the vision and the integrity of the eye.
- ▶ Recurrences can occur in the adjacent areas or in a new location, and a high index of suspicion is needed to detect this complication early.

Contributors TK, SS and SJ have contributed to the drafting of the article, and its critical revision for important intellectual content. All the authors have contributed to the conception, design, acquisition and interpretation of the data.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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Figure 4 Final visit slit lamp photograph showing a well-healed conjunctiva with intact scleral patch grafts (arrow) in OD and OS.

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