

Spontaneous Scleral Perforation and Optic Nerve Dural Ectasia in Marfan Syndrome

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Abstract

Purpose: To report two patients with spontaneous scleral perforation and bleb formation associated with Marfan syndrome (MFS); which one of them also showed bilateral optic nerve dural ectasia.

Case reports: An 11-year-old girl was referred with progressive myopia. Past medical history was negative. Slit-lamp examination revealed bilateral scleral thinning and perforation with bleb formation, shallow anterior chamber (AC), and microspherophakia. Systemic evaluation confirmed the diagnosis of MFS. B-scan ultrasonography and orbital MRI showed bilateral optic nerve dural ectasia. A 23-year-old man, known case of MFS, also presented with spontaneous scleral perforation with bleb formation and microspherophakia. In this case, ancillary tests didn't show optic nerve dural ectasia. Both patients underwent lensectomy with vitrectomy and iris-fixated intraocular lens implantation in both eyes.

Conclusion: MFS may be associated with new and rare findings as part of generalized connective tissue disorder.

Keywords: Scleral Perforation, Optic Nerve, Dural Ectasia, Marfan Syndrome

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Introduction

Marfan syndrome (MFS) is a hereditary systemic connective tissue disorder characterized by musculoskeletal, cardiovascular, and ocular abnormalities. It is most commonly caused by mutations in the fibrillin-1 (FBN1) gene on chromosome 15.¹

The main ocular manifestations of MFS include ectopia lentis, microspherophakia, cataract, myopia and astigmatism, glaucoma, retinal tear and detachment, and strabismus.¹

The abnormal fibrillin weakens the mechanical properties of the sclera and may be the cause of scleral thinning in MFS.

Scleral perforation following trabeculectomy,² and scleral buckling procedure,³ have been reported in MFS but spontaneous scleral perforation is rare.⁴ Dural ectasia is a major diagnostic criterion specially in young patients with Marfan and has been reported to present almost always in lumbar area.^{1,5}

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Herein, we report two patients with MFS and bilateral spontaneous scleral thinning and perforations, one of them also showed bilateral optic nerve dural ectasia.

Case reports

Case 1

An 11-year-old girl was referred to our clinic with progressive myopia and decreased visual acuity (VA). According to her optometrist, over a period of six months her refraction had increased from -10-1x180 to -12-1x180 in the right eye and from -10-2x180 to -14-2x180 in the left eye. Best corrected visual acuity (BCVA) was $20/30$ in the right eye and $20/50$ in the left eye. Past medical history was negative for trauma and surgery. Slit-lamp examination revealed an area of scleral thinning measured 1.5x4 mm in supranasal quadrants of both eye adjacent to limbus leading to perforation and bleb formation (Figure 1). Fluorescein staining revealed no leakage. Both corneas were clear and measured 11.5 mm in diameter. Anterior chamber (AC) was extremely shallow specially on the left side and upward subluxation of microspherophakic lenses were observed in both eyes after dilation of the pupils. Intraocular pressure (IOP) was 11 and 3 mmHg in the right and left eyes, respectively. Funduscopy revealed normal retina and vessels; average sized optic discs with sharp margin, pink rim, and C/D ratio of 0.3. Axial lengths were 21.87 mm (right eye) and 21.59 mm (left eye).

The diagnosis of MFS was confirmed after systemic evaluation and rule out rheumatic diseases, with tall stature and long extremities as well as dilation of aorta demonstrated by echocardiography. Family history was negative for MFS. To evaluate the patient completely, B-scan ultrasonography was performed and showed bilateral cystic dilation of the optic nerves (Figure 2). Orbital MRI confirmed optic nerve dural ectasia (Figures 3 and 4). Dural ectasia was not found on lumbar MRI scan.

The patient underwent lensectomy and vitrectomy with iris-fixated intraocular lens (IOL) implantation in both eyes. After surgery the bleb decreased in size in the left eye and IOP increased to 10 mmHg. At three months BCVA was 20/25 in both eyes.

Case 2

A 23-year-old male, known case of MFS was referred for ophthalmic examination. BCVA was $20/40$ in both eyes with a refraction of -11 -4x40 and -14 -4x115 in the right and left eyes, respectively. Slit-lamp examination revealed bilateral scleral perforation and bleb formation in supranasal quadrants, shallow AC and microspherophakia. IOP was 9 mmHg in both eyes. Axial lengths were 21.33 mm (right eye) and 21.67 mm (left eye). Other examinations including funduscopy and B scan ultrasonography were unremarkable. The patient underwent lensectomy and vitrectomy with iris-fixated IOL implantation in both eyes. After surgery AC depth increased, although the blebs did not change in appearance.

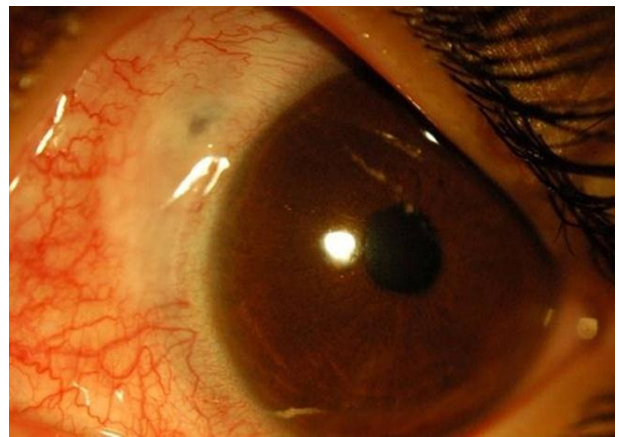


Figure 1. Slit-lamp photograph of the left eye of an 11-year-old girl with Marfan syndrome showing spontaneous scleral perforation and bleb formation

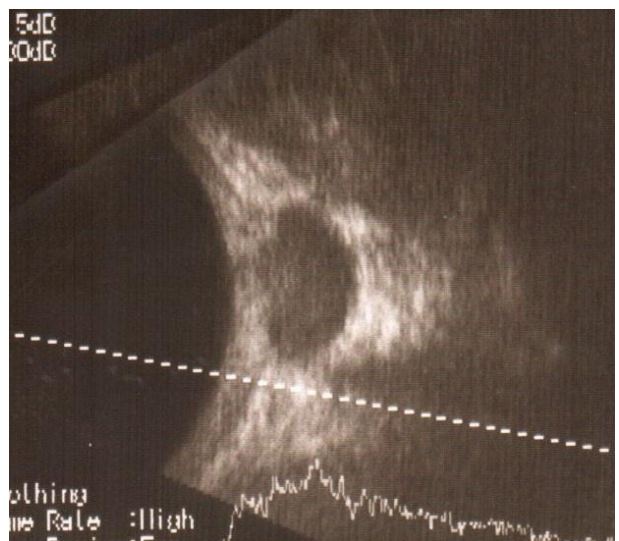


Figure 2. Case 1: B-scan ultrasonography shows cystic dilation of the optic nerve.

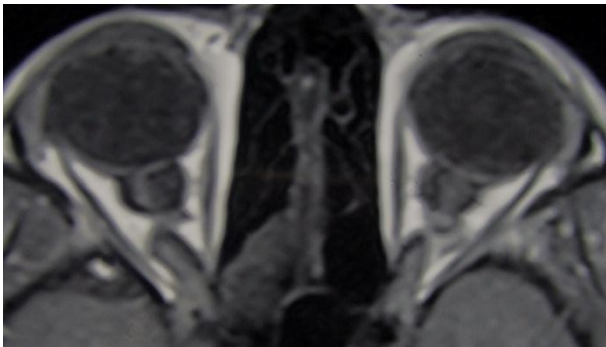


Figure 3. Case 1: T1 axial orbital MRI showing bilateral optic nerve dural ectasia with expansion of cerebrospinal fluid spaces around normal optic nerves. Normally, there is a very thin layer of cerebrospinal fluid around the optic nerve.

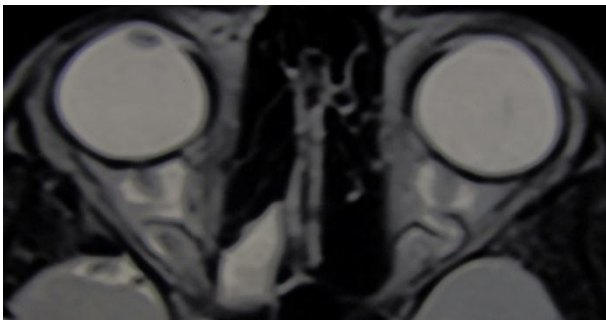


Figure 4. Case 1: T2 axial orbital MRI clearly shows dilated perioptic nerve sheaths with accumulation of cerebrospinal fluid (white on T2-weighted images) around normal size optic nerves. Also note the absence of lens signal on the left side after lensectomy.

Discussion

MFS is a hereditary systemic connective tissue disorder with ocular, skeletal and cardiac manifestations. It is the second most common inherited connective tissue disorder, after osteogenesis imperfecta.¹ While usually inherited as an autosomal dominant trait, 25-30% of cases present sporadically.¹ MFS is most commonly caused by mutations in the FBN1 gene on chromosome 15. However, it can also be caused by inactivation mutations in TGF- β receptor 2 (TGFB2) on chromosome 3.⁶

The diagnosis of MFS remains clinical. The Ghent diagnostic criteria are used for diagnosis in seven areas including cardiovascular system, pulmonary system, ocular system, skeletal system, skin and integument, dura mater, and family or genetic history.^{1,5} Patients may be systemically normal

with only ocular signs, such as dislocated lenses, to suggest the diagnosis.

The systemic manifestations of MFS include tall stature, long extremities, arachnodactyly, chest wall deformity, aortic and pulmonary artery dilatation, and mitral valve prolapsed.^{1,5}

FBN1 is the major structural component of the microfibrils present in elastic fibers and the main defective gene product in MFS. 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

Ocular manifestation of Marfan could be explained by structural deficiency of FBN1. Mechanical stretching of the sclera containing abnormal fibrillin molecule may result in axial myopia and progressive scleral thinning.

Scleral thinning and perforation may occur after trauma or surgery.^{2,3} Rodriguez-Ares et al described scleral perforation after trabeculectomy with mitomycin-C in MFS.² Deramo et al reported full-thickness scleral erosion following sclera buckling procedure secondary to a silicone buckle causing hypotony in a patient with MFS.³

Spontaneous scleral perforation in MFS is rare. Law et al reported an 18-year-old male with bilateral spontaneous scleral perforation requiring surgical repair with a scleral patch graft.⁴ The perforations were in supranasal quadrants of both eyes.

Our patients had no history of trauma or surgery and scleral perforations occurred spontaneously in supranasal quadrants. It caused bleb formation with resultant decrease in AC depth and progressive myopia. Whether the occurrence of thinning in supranasal quadrants in our patients and also in Law report is accidental or not is not known. Another interesting point in our patients is that considering axial lengths, none of them was axial myope.

Dural ectasia is a major diagnostic criterion specially in children.^{1,5} Studies have shown that dural ectasia is present in 63-92% of patients with MFS.^{1,7} In these patients, the dilation of the dural sac is almost always at the level of the lumbar region and presents with low back pain. This has been attributed to the effects of gravity and higher subarachnoid pressure in upright position.⁷ Dural ectasia is considered a sensitive and specific marker for

confirmation of a diagnosis of MFS in young patients.¹

Optic nerve dural ectasia in our patient was asymptomatic and was found accidentally on a routine ultrasound exam. To the best of our knowledge, this has not been reported previously. We are not aware of any visual impact of this finding. Additionally, optic nerve dural ectasia cannot be explained by the effects of gravity. However, it shows the possibility of the presence of dural ectasia in

other areas of the central nervous system and the need for performing routine B-scan and/or MRI in these patients even in the presence of normal looking optic discs.

Conclusion

In summary, MFS may be associated with rare or new presentations. This report warrants performing ancillary tests in Marfan patients with apparently normal optic discs.

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