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Case Report

Ophthalmology Section

Ectopia Lentis with Retinal Detachment in a Patient with Marfan Syndrome: A Case Report

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ABSTRACT

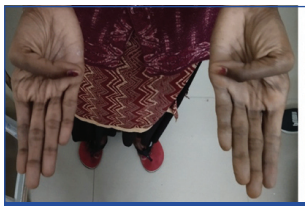
Marfan syndrome is an autosomal dominant hereditary connective tissue disease caused by mutations in the Fibrillin-1 gene located on chromosome 15q15-21. Fibrillin is a specific type of glycoprotein widely distributed throughout the body, contributing to the elasticity and load-bearing capacity of connective tissue. Marfan syndrome is a multisystem disorder that affects the cardiovascular system, musculoskeletal system, and the eyes. Patients with Marfan syndrome can experience life-threatening complications such as aortic aneurysms, aortic dissection, and mitral valve prolapse. However, most patients initially present to an ophthalmologist with ocular symptoms, necessitating the ophthalmologist to diagnose and counsel the patient about the disease and its complications. A 23-year-old female patient presented to the Ophthalmology Outpatient Department (OPD) with complaints of diminished visual acuity in both eyes since birth. The patient exhibited cataractous changes in the lens with dislocation in the superotemporal quadrant in both eyes. Furthermore, the patient experienced rhegmatogenous retinal detachment in her left eye. The patient underwent scleral belt buckling and pars plana vitrectomy with silicone oil insertion in the left eye, as well as cataract extraction with Posterior Chamber Intraocular Lens (PCIOL) implantation in the right eye. Marfan syndrome can impact visual function in various ways. The present case report underscores the ocular manifestations of Marfan syndrome and the management of patients with developmental cataracts, subluxation, and retinal detachment due to high myopia.

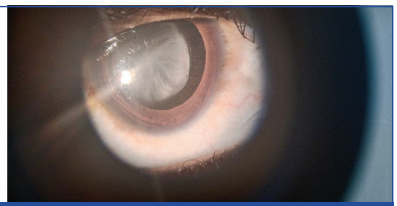
Keywords: Cataract surgery, Connective tissue disorder, Dislocated lens, Myopia, Vitrectomy

CASE REPORT

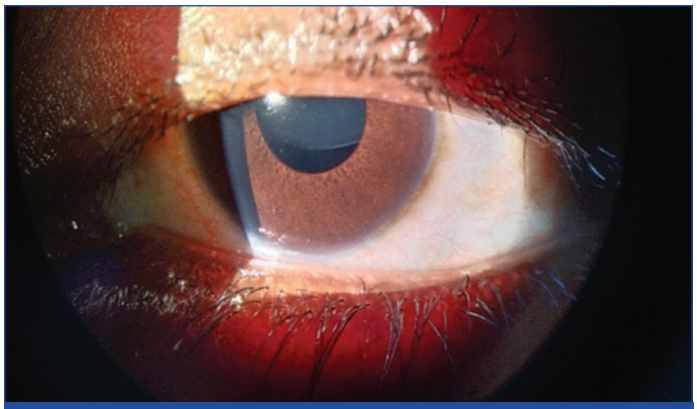
A 23-year-old female presented to the Ophthalmology Outpatient Department (OPD) with the main complaint of reduced visual acuity in both eyes since birth. The patient noted that the decreased visual acuity was more pronounced in the left eye compared to the right eye and had worsened over the last six months. She had no history of ocular pain or trauma. The patient appeared tall with a slender build and an increased arm span [Table/Fig-1]. She exhibited long fingers suggestive of arachnodactyly and a hyperflexible thumb, indicating a positive thumb sign [Table/Fig-2]. Cardiovascular, pulmonary, and dermatological examinations of the patient were all normal. In the right eye, visual acuity was Counting Fingers (CF) 1 metre (m), and in the left eye, there was perception of light with projection of rays. Intraocular pressure measured 18 mm/Hg in the right eye and 5 mm/Hg in the left eye using a non contact tonometer. The axial length was 28.22 mm in the right eye and 28.45 mm in the left eye. Slit lamp evaluation revealed developmental cataracts with superotemporal lens dislocation in both eyes [Table/Fig-3,4]. B-scan ultrasound of the left eye showed retinal detachment [Table/Fig-5], while the right eye was within normal limits, with no signs of retinal detachment [Table/Fig-6].

[Table/Fig-1]: Increased arm span of the patient.

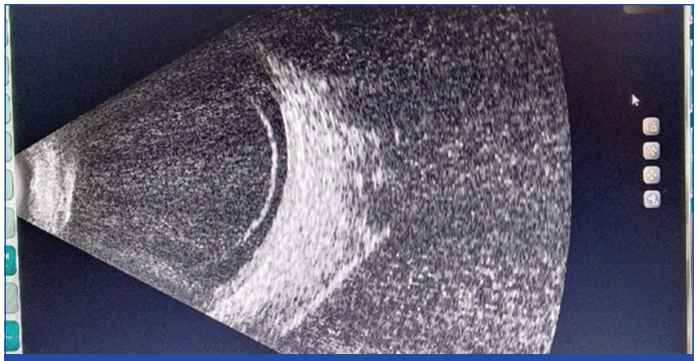




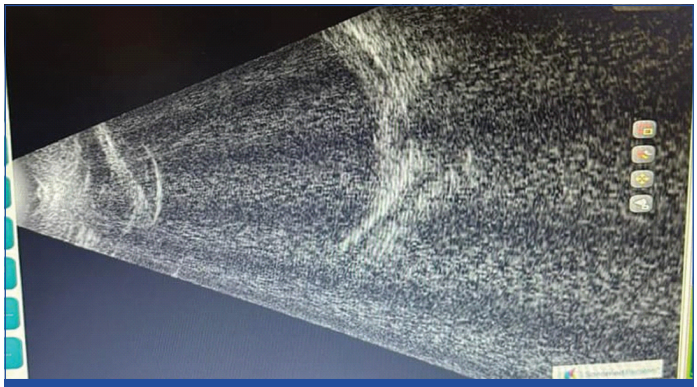
[Table/Fig-2]: Arachnodactyly with hyperflexible thumb joint. **[Table/Fig-3]:** Slit lamp examination of right eye showing dislocated lens along with cataractous changes. (Images from left to right)



[Table/Fig-4]: Slit lamp examination of the left eye showing dislocated lens along with cataractous changes.

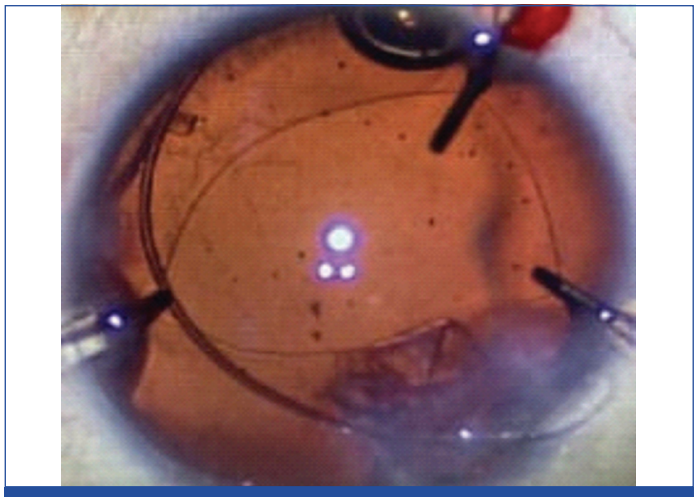


[Table/Fig-5]: B-scan of left eye showing retinal detachment.



[Table/Fig-6]: B-scan of right eye showing no evidence of retinal detachment.

A diagnosis of Marfan syndrome was suspected based on clinical findings. After obtaining informed consent, the patient was scheduled for cataract extraction in the right eye under short sedation. This choice of anaesthesia was made because the patient was unwilling to undergo a peribulbar block due to apprehension about the procedure. A capsular tension ring was utilised to maintain the integrity of the capsular bag in this patient with poor zonules; a zonular dialysis was observed for 6 o'clock hours from 1 o'clock to 7 o'clock [Table/Fig-7]. The right eye was implanted with a hydrophobic acrylic Intraocular Lens (IOL) since the bag's integrity was preserved after Capsular Tension Ring (CTR) implantation, eliminating the need for an Iris claw or a scleral fixated IOL, as initially anticipated. The patient underwent a pars plana vitrectomy under general anaesthesia seven days later, following informed consent. The decision to use general anaesthesia was due to the patient's reluctance to undergo a peribulbar block, and the longer duration of the pars plana vitrectomy procedure. In the left eye, scleral belt buckling and pars plana vitrectomy were performed, followed by the use of silicone oil for retinal tamponade. A month after discharge, cataract extraction for the left eye was recommended. One week post-operation, the visual acuity of the right eye improved to 6/36, the anterior chamber was quiet, and the IOL remained stable.



[Table/Fig-7]: Capsular tension ring implantation.

DISCUSSION

Marfan syndrome is an autosomal dominant condition that causes multiple systemic disorders including cardiovascular, ocular, and musculoskeletal issues. Marfan syndrome is associated with the glycoprotein Fibrillin-1 and Tumour Growth Factor (TGF) beta, a disorder characterised by abnormal connective tissues in various organs [1]. The Ghent criteria is a recognised standard for diagnosing Marfan syndrome, consisting of seven sections: ocular system, musculoskeletal system, cardiovascular system, pulmonary system, dura matter, skin system, and congenital history [2]. Ectopia lentis has been defined as a key clinical feature of Marfan syndrome since 2010, enhancing its sensitivity and specificity [3]. Marfan syndrome affects multiple parts of the eye since fibrillin is present in all ocular tissues [4,5] as described in [Table/Fig-8]. Surgical intervention for ectopia lentis may be necessary in cases involving cataracts, uveitis, anterior or posterior lens displacement, secondary glaucoma, lens-induced glaucoma, posterior lens dislocation into the vitreous, difficulty achieving adequate corrected visual acuity, and the potential for amblyopia in children [6].

Cornea	Flattened cornea
Angle of the anterior chamber	Glaucoma
Iris	Iris coloboma, Iridodonesis
Lens	Dislocation, cataract, microspherophakia
Vitreous	Liquefaction, degenerations
Retina	Detachment, tears, degeneration
Globe	Increased axial length causing high myopia

[Table/Fig-8]: Ophthalmic manifestations in Marfan syndrome [6].

Removing a subluxated lens from these individuals can be challenging. Weakened zonules and an unstable lens can lead to complications such as vitreous humor disruption and loss of the capsular bag [6]. Individuals with Marfan syndrome, due to high myopia and posterior staphyloma, require special consideration when calculating IOL power as inaccurate calculations can result from ultrasound biometry. The prevalence of this disorder is higher in patients with Marfan syndrome (34% to 44%) compared to the general population [7]. Cataract development occurs earlier in life for patients with Marfan syndrome. Significant zonular weakness is a symptom of Marfan syndrome, increasing the risk of complications during ocular surgery. Surgeons may need to use a CTR while performing cataract surgery in adults or children [8]. Glaucoma occurs in approximately one-third of patients with Marfan syndrome [9], with Primary Open Angle Glaucoma (POAG) being the most common type. Two potential causes of POAG include displacement of Schlemm's canal and abnormal placement of the ciliary muscles into the trabecular meshwork [10]. Retinal detachment is more prevalent in individuals with Marfan syndrome due to factors such as ectopia lentis, high myopia, posterior vitreous detachment, and vitreous liquefaction. Additionally, retinal detachment is more commonly seen in younger Marfan syndrome patients, those with ectopia lentis, and those with a history of aphakia [11,12]. In a study by Erdogan G et al., three surgical techniques - intrascleral IOL fixation, scleral IOL fixation with the Z-suture, and IOL implantation after crystalline lens removal - were used to treat ectopia lentis in Marfan syndrome [13]. Visual acuity in each group improved significantly post-surgery. There was no significant difference in ocular residual astigmatism among the groups (p-value=0.51). CTR ring implantation was successfully performed in this case.

Sulcus fixation of an IOL in eyes without capsular support is an option for correcting aphakia in children. In a study by Zetterstrom C et al., haptics of the IOL were sutured to the sclera and the IOL was implanted in the sulcus. Follow-up ranging from 9 to 33 months showed no occurrences of retinal detachment, secondary glaucoma, or posterior capsular opacification [14].

Previously, the most common method for correcting aphakia was flexible open-loop Anterior Chamber Intraocular Lens (ACIOL) implantation. In a short-term follow-up, patients with Marfan syndrome who underwent pars plana lensectomy with ACIOL implantation achieved good postoperative visual acuity without significant complications. However, this approach raises concerns in Marfan syndrome cases where the anterior chamber is often too deep, making proper IOL fitting challenging and leading to excessive IOL movement. Loose fit can result in inflammation, glaucoma, pigment release, and corneal decompensation [15]. In present particular case, a PCIOL implant was chosen due to the maintenance of capsular bag integrity by the CTR, which helped prevent associated complications with other types of intraocular lenses.

CONCLUSION(S)

Marfan syndrome is a multisystem disorder that can cause serious, life-threatening complications such as aortic dissection, aortic aneurysm, and mitral valve prolapse. Ocular involvement is very common, and in many cases, Marfan syndrome is first diagnosed by an Ophthalmologist. Early diagnosis and management can be beneficial for the patient in preventing ocular complications and other systemic, life-threatening issues.

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