Superior and Inferior Lens Subluxation in a Patient of Marfan Syndrome: A Rare Case Presentation

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Abstract

Marfan syndrome (MFS) is a spectrum of disorders caused by a heritable genetic defect of connective tissue involving the musculoskeletal, cardiac, and ocular system predominately. The defect itself has been isolated to the fibrillin1 (FBN1) gene on chromosome 15, which codes for the connective tissue protein FBN. Aortic root dilatation and ectopia lentis are the cardinal clinical features. In the absence of family history, the presence of these two manifestations is sufficient for confirmatory diagnosis of MFS. There’s no cure for MFS, so treatment focuses on managing the symptoms and reducing the risk of complications. Recent advances in diagnosis, improved surgical technique and application of prophylaxis has contributed in the preservation of sight in patients.

Key words: Ectopia lentis, Marfan syndrome, Ocular manifestation

INTRODUCTION

Marfan syndrome (MFS) is the most common cause of heritable ectopia lentis, and ectopia lentis is the most frequent ocular manifestation of MFS, occurring in approximately 75% of patients.¹ MFS is an autosomal dominant disease resulting from various mutations to the fibrillin-1 (FBN-1) gene located on chromosome 15. It is thought that the increased incidence of ectopia lentis with MFS is due to altered FBN microfibrils leading to incompetent zonular fibers and structural abnormalities of the lens capsule.² Lens dislocation in MFS is usually bilateral and occurs most often in the superotemporal direction, though other directions are not uncommon.³

CASE REPORT

A 23-year-old male presented to our Ophthalmology Department on 10th March 2017 with a complaint of diminution of vision and glare. The patient was tall-statured with thin and long extremities. Ocular examination revealed visual acuity in the right eye as 6/6 (With correction of −0.50 D sphere; −3.50 cylinder 160) and left eye as 6/6 (with correction of −4.00 cylinder 180).

Ocular motility was full and free in all direction of gaze. Slit lamp biomicroscope examination of anterior segment revealed clear cornea in both eyes without any corneal ectasia and megalocornea. Both eyes angle were deep without any opposition to cornea on either side. No evidence of anisocoria and relative afferent pupillary defect. Intraocular pressure was 16 mm Hg (with non-contact tonometer) for both eyes. Dilated fundoscopy examination showed normal posterior segment without peripheral retinal degeneration or detachment. Superotemporal subluxation of the lens was noted in the right eye and inferonasal subluxation in the left eye as shown in Figure 1.

Systemic examination revealed skeletal abnormalities such as long, thin extremities, arm span greater than the height (1.06), a positive thumb and wrist sign, pectus excavatum, prominent finger joints, and high-arched palate. The patient was referred to cardiologist and echocardiography revealed mild dilatation of aortic root. The patient gave the history of the long stature of the mother and maternal uncle as well. Diagnosis of MFS was made on clinical and radiological findings.
The presence of zonules weakness and capsular instability makes implantation of an intraocular lens (IOLs) difficult with amplification of usual complication of lens extraction. Surgical options include anterior chamber IOL, ciliary sulcus posterior chamber IOL fixed to the sclera and/or to the iris, and scleral fixated capsular tension rings. Capsular tension rings is a suitable option as it allows preservation of the capsular bag and primary implantation of IOL. It is 2700 open polymethyl methacrylate ring which causes an even distribution of centrifugal forces through the zonules. These rings contain holes that allow centering and fixation of the capsule bag to the scleral wall. Recent reports show good visual outcomes without any serious complications in surgery.6,8

Our patient did not show any other ocular finding on examination. He was advised frequent follow-up with an ophthalmologist for early detection of other ocular features and follow-up with cardiologist and orthopedician was also suggested to improve the quality of life and to help early detection of life-threatening complications like dissection of the aorta.

CONCLUSION

Ophthalmologist has a very key role to play both in diagnosis as well as treatment of MFS. Timely diagnosis and management can help in preserving the vision and hence the quality of life.

REFERENCES


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