Print ISSN: 2321-6379
Online ISSN: 2321-595X
DOI: 10.17354/ijss/2017/201

Case Report

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.

Access this article online

IJSS
www.ijss-sn.com

Month of Submission : 02-2017
Month of Peer Review : 03-2017
Month of Acceptance : 03-2017
Month of Publishing : 04-2017

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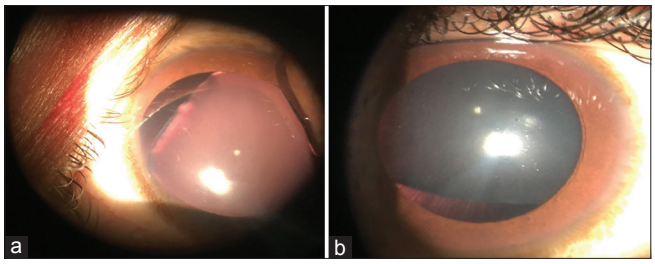

Figure 1: (a) Inferonasal sublaxation of lens in left eye, (b) superotemporal sublaxation of lens in right eye

DISCUSSION

This patient had presented to us with a complaint of decreased vision with a glare. Following refraction, the patient received visual acuity of 6/6. Patient's family history was inconclusive. The patient was referred to a cardiologist with suspicion of MFS for further evaluation. Echocardiography finding of dilatation of aorta supported the diagnosis of MFS in the presence of definitive history. Mutation analysis of FBN1 gene helps in prompt diagnosis.[4]

For ocular system involvement to be used as diagnostic criteria according to Ghent criteria, the major criterion or at least two minor criteria must be present.[3]

The ocular system major criterion is ectopia lentis (lens dislocation)[5] which was present in our case.

Minor ocular system criteria are as follows:

- An abnormally flat cornea
- An increased axial length of the globe, as measured by ultrasound
- A hypoplastic iris or hypoplastic ciliary muscle, causing myopia.

Ectopia lentis is usually bilateral, symmetric, supertemporal in location, and non-progressive entity present in 50-80% of the affected individual.[3] It varies from mild asymptomatic displacement to significant subluxation resulting in monocular diplopia.[6] Anterior dislocation of the lens results into pupillary block glaucoma or chronic angle closure glaucoma. Posterior dislocation results in posterior uveitis or chorioretinal inflammation due to leakage of lens proteins.[6] Non-surgical management includes refractive correction and application of miotic drugs.

Surgical indication for lens extraction includes lens opacity, anisometropia, non-correctable refractive error, impending total luxation of lens, and lens induced glaucoma or uveitis.[6,7]

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 throught 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.

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How to cite this article: Kaur N, Singla E, Saini V, Madan R. Superior and Inferior Lens Subluxation in a Patient of Marfan Syndrome: A Rare Case Presentation. Int J Sci Stud 2017;5(1):255-256.

Source of Support: Nil, **Conflict of Interest:** None declared.