Floating Lens In Marfan’s Syndrome: A Case Report

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Abstract: Marfan’s syndrome (MFS) is an autosomal dominant connective tissue disorder involving the cardiovascular, skeletal and ocular systems. Cardinal manifestations include aortic aneurysm and dissection, ocular lens dislocation and long bone overgrowth. The most common cause of ectopia lentis is trauma, which accounts for nearly onehalf of all cases of lens dislocation. A forward dislocation of the lens into the pupil or anterior chamber may cause pupillary block with acute glaucoma or chronic angle-closure glaucoma. Posterior dislocation can cause harmful vitreous traction on the retina with leakage of lens proteins into the vitreous, which may cause chronic vitritis and chorioretinal inflammation. We presented a case of Marfan’s syndrome who presented with anterior dislocated lens in sitting posture which falls back to posterior segment in supine position which we described it as “floating lens” which has not been described in medical literature so far. Surprisingly, when the dislocated lens was in the anterior chamber, IOP was unrecordably low in contrast to what’s expected in literature (IOP is always raised in anteriorly dislocated lens of Marfan’s syndrome due to pupillary block)

Keywords: Floating lens, Marfan’s syndrome, MFS, Ectopia lentis, dislocated lens.

I. Introduction

Marfan’s syndrome (MFS) is an autosomal dominant connective tissue disorder involving the cardiovascular, skeletal and ocular systems. Cardinal manifestations include aortic aneurysm and dissection, ocular lens dislocation and long bone overgrowth.¹ It was first described by Antoine – Bernard Marfan in an 1896 case report of a young girl with unusual musculoskeletal features,² while Bürger first described ophthalmological features of MFS in 1914.³ Worldwide, the incidence of Marfan’s syndrome is approximately 7–17/100,000.⁴

Ocular features of this syndrome have been repeatedly reported.⁵ Ectopia lentis, the most common ocular feature, occurs in 70 to 80 % of cases.⁶ Progressive aortic dilatation, usually maximal at the sinus of Valsalva, associated with aortic valve incompetence leads to aortic dissection or rupture and is the principal cause of mortality, but mitral valve prolapse with incompetence may be significant. Lens dislocation, myopia, and arthritis associated with chronic joint laxity can cause substantial morbidity.⁷

The diagnosis is commonly considered in a young person with a tall, thin body habitus, long limbs, arachnodactyly, pectus deformities, and sometimes scoliosis. Family history may be helpful, but around 27% of cases arise from new mutation.⁸

II. Case Report

A 22 years old female presented with a chief complaint of decreased vision of right eye for 10 days. She had a history of fall on the floor due to dizziness 10 days back, following which she started complaining of decreased vision. Patient had tall stature and thin built with long face. A family history of marfan’s syndrome was found in her mother.

On examination, her arm span (169 cm) was greater than her height (163 cm)
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Fig: Arm span -169 cm was greater than her height - 163 cm (A); Steinberg sign-positive (B) Walker murdoch sign positive (C).

Steinberg sign-positive ;Walker murdoch sign- positive
B.P-100/60 mm of Hg
Pulse-80/min
Snellen’s Visual Acuity (unaided)
   Right eye - PL +, PR accurate in all quadrants
   Left eye- 6/18

Slitlamp examination
Adenexa- WNL BE
Conjunctiva - Transparent BE
Cornea- Clear BE

Anterior Chamber-
   RE-Phakic, clear floating lens nucleus could be seen in AC in sitting posture while on changing to supineposition, the floating lens went into the vitreous cavity and came back into AC again in sitting posture. AC was full with vitreous. AC was quiet in LE.

Iris- Iridodonesis in RE, normal in left eye

Pupil-Middilated, non reacting in RE, NSNR in LE
**LENS** - Dislocated clear lens floating in AC and vitreous cavity in RE. Clear lens was subluxated superotemporally in LE.

**Fundoscopy**
RE - In sitting posture glow was absent while in supine position glow could be seen. Disc was seen hazily due to vitreous haze. Floating lens could be seen in vitreous in supine position.
LE - Tessellated fundus with large optic disc with inferotemporal crescent
IOP - RE - unrecordably low (at the time of presentation)
LE - 14.6 mm of Hg

**BIOMETERY**
RE - K1 -41.25D
K2 -40.50D
AL -29.97mm
PCIOL + 8.0 D

LE - K1 -41.5D
K2 -40.50D
AL -30.63mm

**Gonioscopy**: Open and WNL in all quadrants.

**Investigations**
Hb - 12.3 gm\%
RBS - 85 mg/dl
ECG - WNL

**X Ray Chest (PA View)** - WNL

**USG B Scan (supine position)**
RE - Dislocated lens present in vitreous with vitreous hemorrhage
LE - Superotemporally subluxated lens with membranes in vitreous
Her neurological and cardiovascular systems were found to be normal. No murmurs were heard. Any cutaneous manifestation or skeletal deformity was absent other than arm span>height & arachnodactyly. Lens extraction RE was done and patient was left aphakic and planned for secondary IOL implantation.

III. Discussion

Marfan’s syndrome is an inherited connective tissue disorder with skeletal, ocular, ligamentous, cutaneous, pulmonary, neurological and cardiovascular manifestation. It is an autosomal dominant disorder where abnormalities of fibrillin protein are encoded by the fibrillin-1 gene (FBN1), a large gene composed of 65 exons on chromosome 15q15-q2. It is found in multiple structures in the eye and plays an integral role in maintaining the integrity of the healthy eye. In 25-35% cases, however neither parent has the disorder but instead the syndrome develops because of spontaneous mutation.

The diagnosis is generally complicated because the syndrome presents with varying degree of expression. The diagnosis is generally based on a health history and a clinical evaluation of the affected system. The main ocular features of Marfan’s syndrome, all of which can result in decreased vision, include bilateral ectopia lentis (lens dislocation), myopia and retinal detachment.

Clinical diagnosis depends on a combination of major and minor signs, as defined in the revised 1996 Ghent nosology. The existence of ectopia lentis is considered a major criterion for the diagnosis of Marfan’s syndrome in this nosology, which unequivocally diagnoses or excludes Marfan’s in 86% of cases. The most common cause of ectopia lentis is trauma, which accounts for nearly one-half of all cases of lens dislocation. When associated with this syndrome, ectopia lentis is usually bilateral, symmetric and non-progressive. Other systemic conditions associated with ectopia lentis include homocystinuria, Weil-Marchesani syndrome, hyperlysinemia, sulfite oxidase deficiency and isolated familial ectopia lentis.

A forward dislocation of the lens into the pupil or anterior chamber may cause pupillary block with acute glaucoma or chronic angle-closure glaucoma. Posterior dislocation can cause harmful vitreous traction on the retina with leakage of lens proteins into the vitreous, which may cause chronic vitritis and chorioretinal inflammation. The second most common ocular manifestation in Marfan’s syndrome is myopia, which is found in 34-44% of Marfan’s patients as compared to 4.8% in the general population in one study.

Marfan’s patients are more prone to develop retinal detachment because globe elongation and axial myopia common in Marfan’s syndrome are associated with early vitreous liquefaction and posterior vitreous detachment, retinal thinning, lattice degeneration, and peripheral breaks – all of which predispose patients to multiple large or even giant retinal breaks.

Hypoplasia of dilatator pupillae, angle anomaly is common, but microspherophakia, keratoconus, cornea plana, megalocornea are uncommon features. Strabismus is present in 19-45% of individuals with Marfan’s syndrome (compared to 3-5% in the general population) and may be a presenting sign of the disorder.

Because of zonular weakness and the resultant capsular instability, correction of aphakia with implantation of an intraocular lens ectopia lentis is challenging. Options include anterior chamber IOL, ciliary sulcus posterior chamber IOL fixed to the sclera and/or the iris, and scleral fixated capsular tension rings. Prophylactic medical (e.g β-blockers) and surgical intervention is important in reducing the cardiovascular complications of Marfan’s syndrome. Ophthalmological assessment is important and regular orthoptic review is recommended, particularly in childhood.
References