IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 14, Issue 5 Ver. IV (May. 2015), PP 136-140
www.iosrjournals.org

Floating Lens In Marfan's Syndrome: A Case Report

*Dr. Laltanpuia Chhangte, *Dr. Kalpana, **Dr. Jaideep Bhatt,
*Dr. Pankaj Kumar
**Department of Ophthalmology, GMC College, Haldwani, Uttarakhand*
***Department of Otorhinolaryngology, GMC College, Haldwani, Uttarakhand*

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)

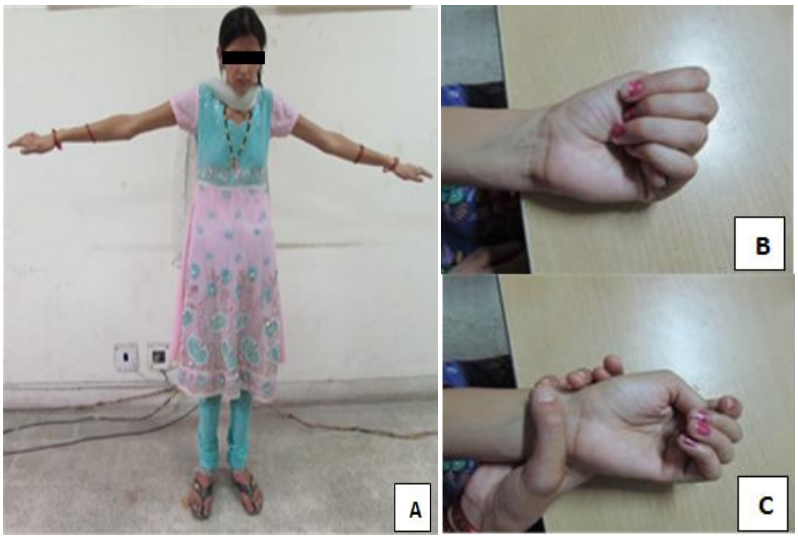
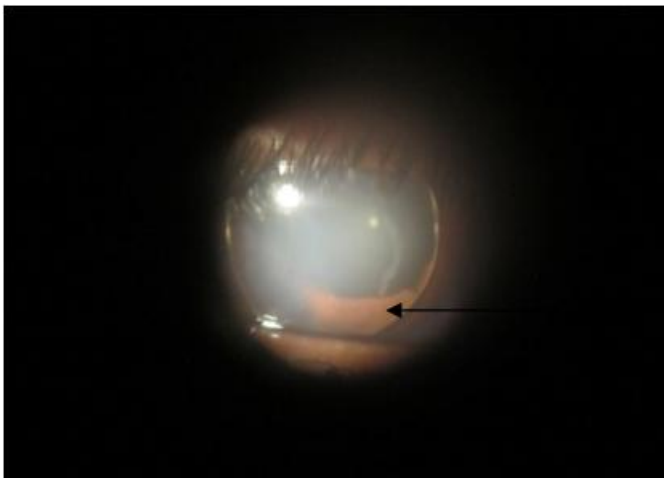


Fig: Arm span -169 cm was greater than her height - 163 cm (A); Steinberg sign-positive (B) Walker murdoch sign- positive (C).

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 accuratein 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.

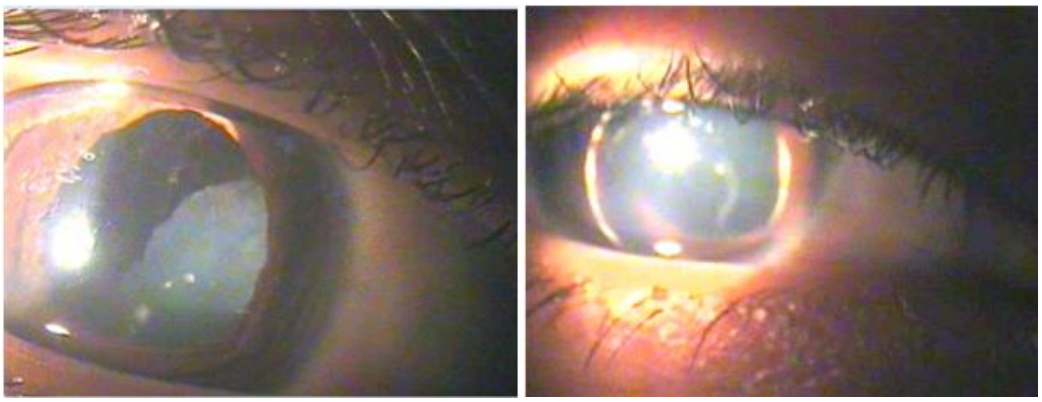


Clear Lens Floating In Anterior Chamber Right Eye

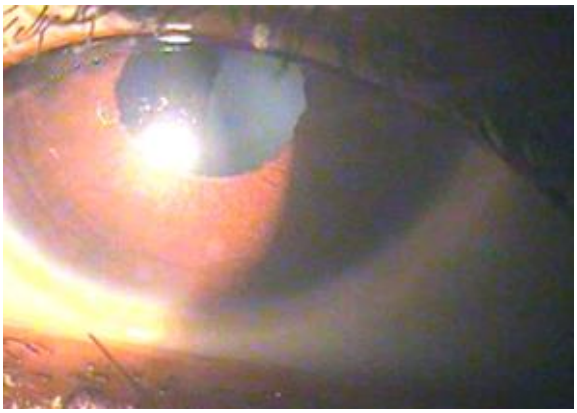
Iris- IridodonesisinRE, normal in left eye

Pupil-Middilated, non reacting in RE, NSNR in LE

LENS- Dislocated clear Lens floating in ACand vitreous cavity in RE. Clearlens was subluxatedsupero-temporally in LE. .



Right eye with lens in vitreous cavity. Right eye with lens in anterior chamber



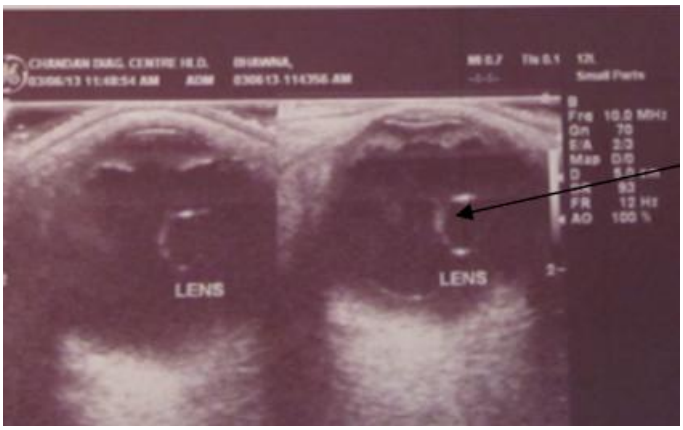
Left eye with superotemporal subluxation of lens

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 LE- K1 -41.5D
K2 -40.50D K2 -40.50D
AL-29.97mm AL-30.63mm
PCIOL + 8.0 D

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- superotemporallysubluxated lens with membranes in vitreous



USG RE showing dislocated lens with vitreous haemorrhage in vitreous cavity

Her neurological and cardiovascular systems were found to be normal. No murmurs were heard. Any cutaneous manifestation or skeletal deformity was absentother 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.[9]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 chromosome15q15-q2.[10]It is found in multiple structuresin the eye and plays an integral role in maintaining the integrityof the healthy eye.[11]In 25 -35% cases,however neither parent has the disorder but instead the syndrome develops because of spontaneous mutation.[12]

The diagnosis is generally complicated because the syndrome presents with varying degree of expression.[13]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.[14]

Clinical diagnosis depends on a combination of major and minor signs, as defined in the revised 1996 Ghent nosology.[15]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.[16]The most common cause of ectopia lentis is trauma, which accounts for nearly onehalf of all cases of lens dislocation.When associated with this syndrome, ectopia lentis is usually bilateral, symmetric and non-progressive.[17]Other systemic conditions associated with ectopia lentis include homocystinuria, Weil-Marchesani syndrome, hyperlysinemia, sulfite oxidase deficiency and isolated familial ectopia lentis.[18]

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.[18]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.[19]

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.[21,22]

Hypoplasia of dilatator pupillae, angle anomaly is common, but microspherophakia, keratoconus, cornea plana, megalocornea are uncommon features.[20]Strabismus is present in 19–45% of individuals with Marfan's syndrome (compared to3–5% in the general population) and may be a presenting sign of the disorder.[23]

Because of zonular weakness and the resultant capsular instability, correction of aphakia with implantation of an intraocular lensin ectopia lentis is challenging. Options include anterior chamberIOL, ciliary sulcus posterior chamber IOL fixed to the sclera and/onto the iris, and scleral fixated capsular tension rings.[24]
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.[25]

References

[1]. Ades L, Members of the CSANZ Cardiovascular Genetics Working Group. Guidelines for the Diagnosis and Management of MarfanSyndrome. Heart Lung Circ 2007; 16: 28-30.
[2]. Ramirez F, Dietz HC. Marfan syndrome: from molecular pathogenesisto clinical treatment. CurrOpin Genet Dev 2007, 17: 1-7.
[3]. Kontić Đ. Sekundarniglaukom. U: Cvetković D, Kontić Đ. Hentova – Senčanić P. Glaukomdijagnoza i lečenje. Zavodzaudžbenike i nastavnasredstva Beograd 1996: 258.
[4]. Harrison's Principles of Internal Medicine. 15th Edition. New York: The Mc-Graw-Hill Companies, Inc.; 2001. pp. 351–355.
[5]. Nelson LB, Maumence IH. Ectopia Lentis. Surv Ophthalmol.1982;27:143 160. [PubMed]
[6]. Pyeritz RE, Whittum-Hudson JA. Immunohistochemical localization of fibrillin in human ocular tissues. Relevance to the MarfanSyndrome. Arch Ophthalmol. 1995;113:103–109.[PubMed]
[7]. Dean JC. Marfan syndrome: Clinical diagnosis and management. Eur J Hum Genet 2007;15:724-33. [PUBMED]
[8]. Grimes SJ, Acheson LS, Matthews AL, Wiesner GL. Clinical consult: Marfan syndrome. Prim Care 2004;31:739-42, xii. [PUBMED]
[9]. Pyretiz RE , Gasnerc.TheMarfan Syndrome, 4th edition ,NewyorkNY:The national marfan syndrome ;1994
[10]. Dietz HC, Pyeritz RE, Hall BD, et al. The Marfan syndrome locus: confirmation of assignment to chromosome 15 and identification of tightly linked markers at 15q15-q21.3. Genomics1991;9:355-61.
[11]. Lee SY, Ang CL. Results of retinal detachment surgery in Marfan syndrome in Asians. Retina 2003;2:24–9.
[12]. Richard Lopez , Julie Berg-McGraw.MarfanSyndrome in a Female Collegiate Basketball Player: A Case Report;J Athl Train. 2000 Jan-Mar; 35(1): 91–95.
[13]. PyretizRE. Toward understanding rate and pathogenesis of Marfansundrome.InHerzetR,Gehle P, Eenker R eds.Cardiovascular aspects of marfansyndrome.Darmstadt,Germany:Steinkopff Verlag;1995:1-8
[14]. Dean JCS, Bradshaw NE, Haites, NE, et al. The Scottish Clinical Guidelines And Integrated Care Pathways For MarfanSyndrome.Genisys, 2003. Available at: http://www.genisys.hw.ac.uk/genisysDR/ NVC/72/Download/msguide.pdf. Accessed 9 June 2007.
[15]. De Paepe A, Devereux RB, Dietz HC, Hennekam RC, PyeritzRE .Revised diagnostic criteria for the Marfan syndrome.Am J MedGenet 1996;62:417–26.
[16]. Dean JC. Marfan syndrome: clinical diagnosis and management.Eur J Hum Genet 2007;15:724–33.
[17]. Rubin SE. Nelson LB. Ocular Manifestations of Autosomal Dominant Systemic Conditions. Duane's Clinical Ophthalmology on CD-Rom. Vol. 3. Ch. 58. Philadelphia: Lippincott Williams & Wilkins, 2006.
[18]. Anteby I, Isaac M, BenEzra D. Hereditary subluxated lenses: visual performances and long-term follow-up after surgery. Ophthalmology 2003;110:1344–8.
[19]. Nemet AY, Assia EI, Apple DJ, Barequet IS. Current concepts of ocular manifestations in Marfan syndrome.Surv Ophthalmol 2006; 51:561–75.
[20]. Kanski JJ. Clinical ophthalmology.A Systematic Approach. Fifth edition Butterworth/Heinemann: Edinburgh – London - New York – Philadelphia - St Louis – Sydney –Toronto, 2003: 698-700.
[21]. Sharma T, Gopal L, Shanmugam MP, et al. Retinal detachment in Marfan syndrome: clinical characteristics and surgical outcome.Retina2002;22:423–8.
[22]. Remulla JF, Tolentino FI. Retinal Detachment in Marfan's syndrome.Int Ophthalmol Clin 2001;41:235–40.
[23]. Ocular Concerns. National Marfan Foundation website. Available at: http://www.marfan.org/nmf/GetSubContentRequestHandler.do?sub_menu_item_content_id=10&menu_item_id=42. Accessed 9June 2007.
[24]. Michaeli A, Assia EI. Scleral and iris fixation of posterior chamber lenses in the absence of capsular support. CurrOpinOphthalmol 2005;16:57–60.
[25]. Dean JCS. MarfanSyndrome: clinical diagnosis and management. Eur J Hum Genet 2007; 15: 724-333.