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Case Series

# Varied ocular manifestations of Marfan's syndrome in a family

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ABSTRACT

Marfan's syndrome may present with varied ocular manifestations, major being ectopia lentis. The purpose of this presentation is to present the case of four members of a family presenting with Marfan's syndrome showing myriad of ocular manifestations. The father, 45 year old male presented with bilateral ectopia lentis (inferiorly subluxated lenses) with raised IOP (33mmHg) in both the eyes. His eldest daughter, 18 year old, also shared similar findings. His second daughter (14 year old) had inferiorly subluxated lens in one eye and anteriorly dislocated lens with corneal opacity in other. Third daughter (13 year old) had intercalary and equatorial staphyloma with anteriorly dislocated cataractous lens with flat AC and raised IOP in right eye and inferiorly subluxated lens in left eye. There was history of death of a male child in the family due to some systemic disease at the age of eight. Systemic examination of the patients revealed no neurological deficits. Cardiovascular system examination was unremarkable. Hands and fingers were long and slender with hyperflexible joints. The ophthalmological management of Marfan's patients is challenging and periodic follow up is needed. Surgical procedure may be required. Each case needs to be evaluated individually to analyse the risks and benefits of the procedures.



## 1. Introduction

Marfan's syndrome (MFS) is a connective tissue disorder involving the cardiovascular, skeletal and ocular systems. Other organs such as skin, adipose tissue, lung, muscle and dura can also be affected.[1,2] It is inherited in autosomal dominant manner and is caused by a mutation in fibrillin–1 (FBN1) gene on chromosome 15.[3] The prevalence of MFS is 1/ 5000 and > 25% of cases are sporadic.[4] Here, we report variable presentations of Marfan's syndrome in four family members of same family.

## 2. Case 1

45 year old father had the complaint of diminution of vision and pain in both eyes. His arm span to height ratio was 1.12. On ophthalmic examination: visual acuity was finger counting at 2 meters in both eyes. Slit lamp examination showed aphakia in right eye and inferior sunset sign due to inferior subluxation of lens in left eye. Refraction showed +14 D in both eyes. Orthoptics showed RDS for near and distance (55BI), ocular motility full in both eyes and BSV negative. IOP was 33mmHg in both eyes. Fundus examination showed small disc in both eyes with a CDR of 0.8 in right eye while 0.4 in left eye. Patient was given the appropriate refraction power and e/d timolol 0.5% BD. (Figure 1)

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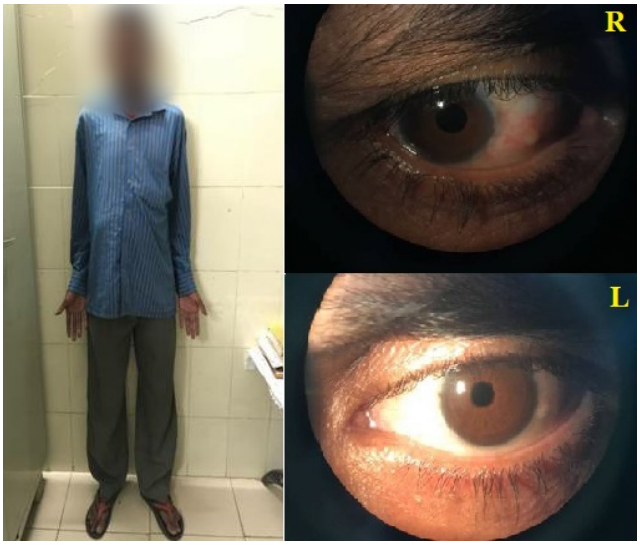


**Fig. 1:** Clinical photograph of case 1, 45 years old male

## 3. Case 2

The eldest 18 year old girl, owing to the problem of poor-sightedness, visited our OPD. On general physical examination, she was tall, thin built, with scoliosis and prominent sternum. She had disproportionately long limbs compared with the trunk (arm span to height ratio =1.09), long spider-like fingers (arachnodactyly) and mild joint hypermobility. Thumb sign and wrist sign were positive. On ocular examination, visual acuity was 2/60 with central nebular corneal opacity and inferiorly subluxated lens in right eye and aphakia in left eye with an IPD of 65mm. The objective refraction of both eyes revealed error of +6 D in both eyes. Orthoptics workup showed ADS for near and distance (25 BI), full ocular motility and BSV negative. Intraocular pressure determined by the Goldmann applanation tonometry method was bilaterally 24 mmHg. Fundus picture showed CDR of 0.8 in both eyes with peripapillary atrophy. After being given a suitable correction of the refractive error and e/d timolol 0.5% BD in both eyes, the girl was then referred to general physician for further consultation. (Figure 2)

## 4. Case 3

The second eldest 14 year old sister, had similar physical features with arm span to height ratio = 1.10. She complained of decrease in vision in both the eyes with pain in right eye. On ophthalmic examination: visual acuity was 5/60 in right eye and hand movement close to face in left eye. Slit lamp examination showed central nebulo-macular corneal opacity and inferiorly subluxated lens in right eye and anteriorly dislocated lens with endothelial pigmentation and leucomatous corneal opacity in left eye. Refraction showed a power of +12 DS/+1DCx180° in both


**Fig. 2:** Clinical photograph of case 2, 18 years old female

eyes. Orthoptics showed LDS (25 BI) for near and distance, full ocular motility and BSV negative. IOP was 28mmHg in right eye and 26mm Hg in left eye. Fundus was normal in right eye and there was no view in left eye. Lens extraction was done in left eye to avoid further damage to cornea and anterior chamber angle. (Figure 3)

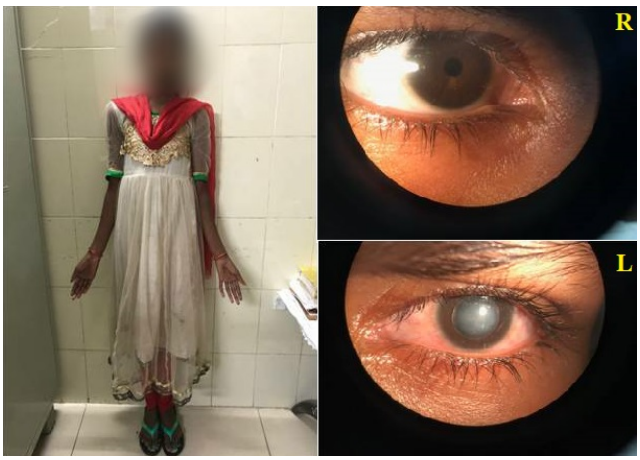


**Fig. 3:** Clinical photograph of case 3, 14 years old female

## 5. Case 4

The youngest one, 13 year old, with similar physical features with arm span to height ratio = 1.10 presented with painful protruding mass in right eye and diminution of vision in both the eyes. On ocular examination: visual acuity was perception of light positive in right eye and 4/60 in left eye. Slit examination showed inferiorly subluxated cataractous lens with posterior synechiae with flat anterior chamber in right eye along with intercalary and equatorial staphyloma while inferiorly subluxated lens in left eye. Orthoptics showed RDS for near and distance (55 BI) and

BSV negative. IOP was 36mm Hg in right eye and 26mmHg in left eye. Fundus could not be seen in right eye while in left eye, cup to disc ratio was 0.7 with normal macula. Enucleation of right eye was done followed by artificial eye shell application 2 weeks later. (Figure 4)


**Fig. 4:** Clinical photograph of case 4, 13 years old female

## 6. Discussion

A typical case of Marfan's syndrome involves manifestations in the cardiovascular, skeletal and ocular systems. Cardiovascular manifestations include progressive aortic root enlargement and abnormal valve leaflets which are thick and elongated. Ascending aortic aneurysm are common in such patients and can lead to life-threatening complications such as aortic regurgitation, dissection or rupture.[2] The most common physical feature which is very evident in such patients is disproportionate increase in linear bone growth that causes overt malformations of the digits, limbs and anterior chest wall as well as craniofacial abnormalities, scoliosis and joint hypermobility.[2] Scoliosis affects around 60% and pectus excavatum affects two-thirds of Marfan's patients. They may cause marked deformity, pain and restricted ventilatory deficit. The prevalence of joint hypermobility in MFS patients is 85% in pediatric population and 56% in adults with many patients suffering arthralgia, myalgia or ligamentous injury. As far as eye is concerned, the prominent features are bilateral ectopia lentis (40 – 56%), myopia (28%) and retinal detachment (due to high myopia). Subluxation usually develops in early childhood, but may first appear in the second decade. Anterior lens dislocation may also occur. Other common features are hypoplasia of dilatator pupillae and angle anomaly whereas microspherophakia, megalocornea, keratoconus and cornea plana are uncommon features.[5] Anisometropia and the possible anterior chamber abnormalities are also need to be addressed. Amblyopia can be prevented by early detection and correction of refractive errors. Laser prophylaxis is essential in high myopes to prevent retinal detachment Surgical management like vitreolensectomy is required for subluxated or dislocated lenses.[6] All family members who are potentially at risk should be motivated for genetic counseling, lifestyle modification and where appropriate, counseling with regard to career options.[1] Prophylactic medical eg beta-blockers and surgical intervention is important in reducing the cardiovascular complications of MFS.[7,8]

## 7. Conclusion

Early ophthalmological assessment including refraction, slit lamp examination and fundoscopy along with thorough general physical examination and referral to cardiologist and orthopaedician is very important especially in children and adolescents. Marfan's syndrome management requires early integrated and multidisciplinary approach to obtain the best quality of life and survival.

## 8. Source of Funding

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## 9. Conflict of Interest

None.

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