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Ectopia Lentis in Marfans Syndrome - A case report

Dr Abhishek Karra, Dr Elika Gupta, Dr Daksh Sharma

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ABSTRACT
Marfan syndrome is a connective tissue disorder with ocular, musculoskeletal and cardiovascular manifestations that are caused by mutations in fibrillin-1. A young male presenting with characteristic ocular and systemic features of Marfan's syndrome. Diagnosis can be made with detailed ocular and general physical examination along with a detailed family history. Genetic counselling and timely examination of patients and their families can help reduce the morbidity and mortality associated with complications of Marfan's syndrome.

I. INTRODUCTION

Marfan's syndrome is a genetic disorder of connective tissue. It is an autosomal dominant disorder involving the cardiovascular, skeletal and ocular system. [1]
Ectopia lentis is the dislocation or displacement of the natural crystalline lens.
Marfan's syndrome is the most common cause of heritable ectopia lentis, and ectopia lentis is the most frequent ocular manifestation of Marfan's syndrome occurring in approximately 75% of patients. [2] It has a worldwide prevalence rate of 1 per 5000 live births. [3]

II. CASE REPORT

A 20-year-old male born out of a non-consanguineous marriage presented to the eye OPD with B/L, painless, progressive, decrease in vision for both far and near since a few months. Detailed medical history and family history was taken. In the family, siblings and relatives were normal.
On ophthalmological examination, the best corrected visual acuity was 6/60 OU and N8 OU with no further improvement. Anterior segment examination in both eyes revealed.

- superotemporal subluxation of crystalline lens (Figure 1)
- Pupils were round, regular and reacting to light.
- Fundus – Clear media with normal disc with normal foveal reflex (Figure 2)
- IOP – within normal limits
- Gonioscopy – Open angles

On general examination,

- Tall stature with increased armspanto body height ratio(Figure 3)
- Kyphoscoliosis (Figure 4A)
- Pectus excavatum(Figure 4B)
- Arachnodactyly(Figure 5)
- Hypermobile joints (Figure 5)
- Pes planus(Figure 6)

Patient was referred to cardiology and general medicine for further systemic evaluation and clearance for surgery under local anesthesia.

III. DISCUSSION

Marfan syndrome is a connective tissue disorder with ocular, musculoskeletal and cardiovascular manifestations that are caused by mutations in fibrillin-1, the major constituent of extracellular microfibrils.[4]

Ectopia lentis is potentially visually debilitating but visual acuity varies with the degree of abnormal position of the lens. The presence of ectopia lentis is a major criterion for the diagnosis of Marfan's syndrome which unequivocally establishes the diagnosis of Marfan's syndrome in 86% of cases [5]. It may be subtle and detectable only by observing phacodonesis or iridodonesis, sometimes visible by gonioscopy. Lens dislocation is characteristically, but not always, supertemporal.[6].
Diagnosis of Marfan's syndrome can be made with Revised Ghent Criteria.
In our patient, systemic score was calculated as

- Wrist and Thumb Sign – 3
- Pectus Excavatum/Chest asymmetry – 1
- Pes planus – 1
- Scoliosis – 1
- Increased armspan to body height ratio – 1

Systemic score of ≥ 7 indicates systemic involvement.

Common non-surgical interventions in ectopia lentis include refractive aid and pharmacological manipulation of the pupil. Indications for lens extraction include

- Lens opacity with poor visual function,
- Anisometropia,

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- Refractive error not amenable to optical correction,
- Impending luxation
- Lens-induced glaucoma or uveitis [7]

In this patient, lensectomy with vitrectomy was done followed by secondary SFIOL fixation as this patient was not amenable for optical correction.

Genetic counselling should also be done as it is an autosomal dominant condition.In order to prevent complications secondary to cardiac manifestations, cardiac consultation should not be missed. Management of patients with Marfan's syndrome requires multidisciplinary approach and early diagnosis is important.

Figure 1 – B/L Supertemporal Subluxation of crystalline lens

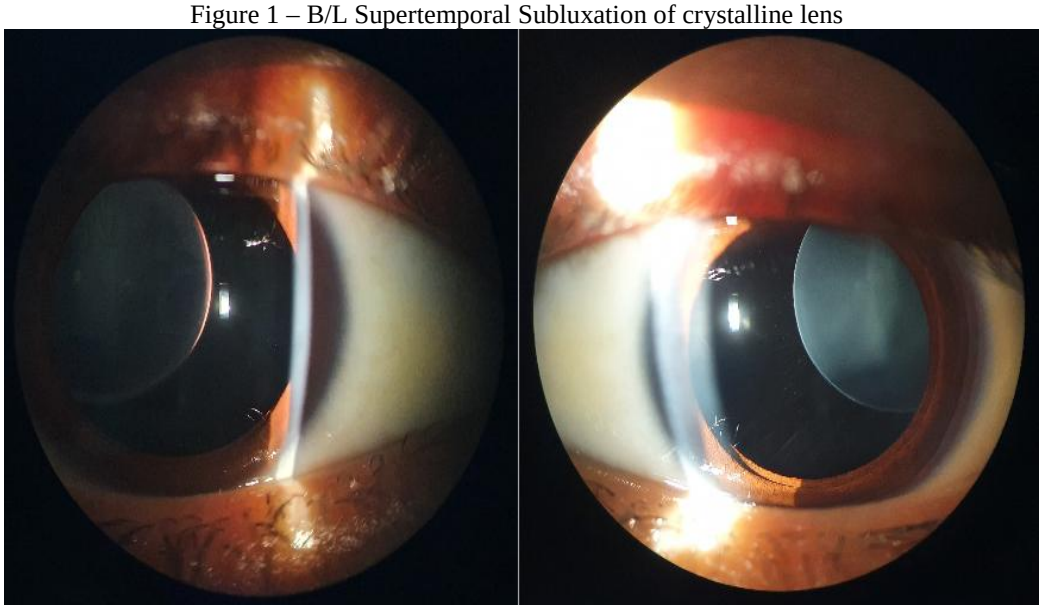

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FIGURE 2 – Fundus (Normal)

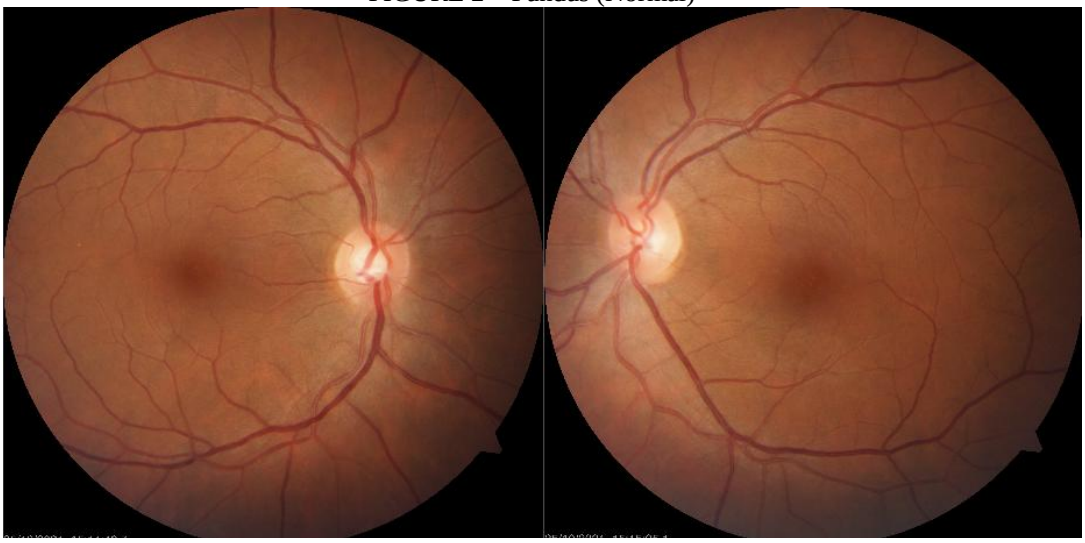

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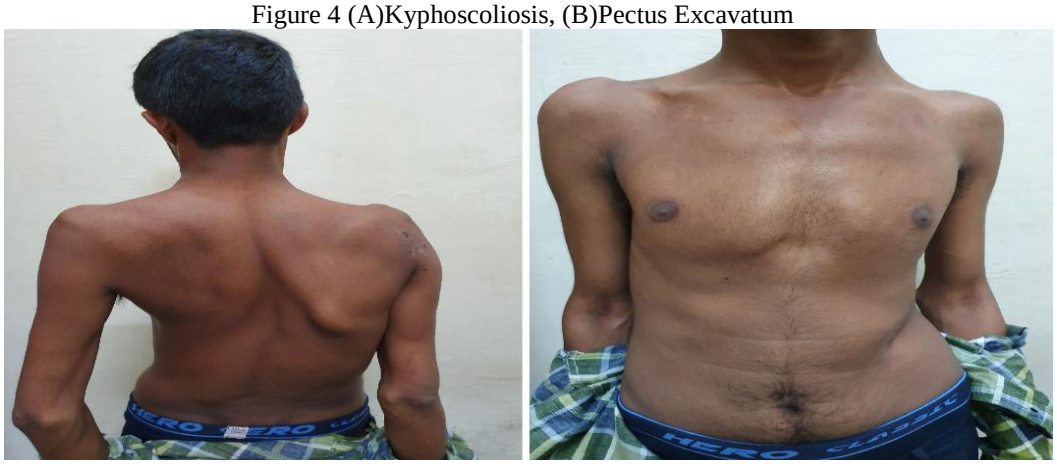
Figure 3 – Increased armspan to body height ratio (>1.05)

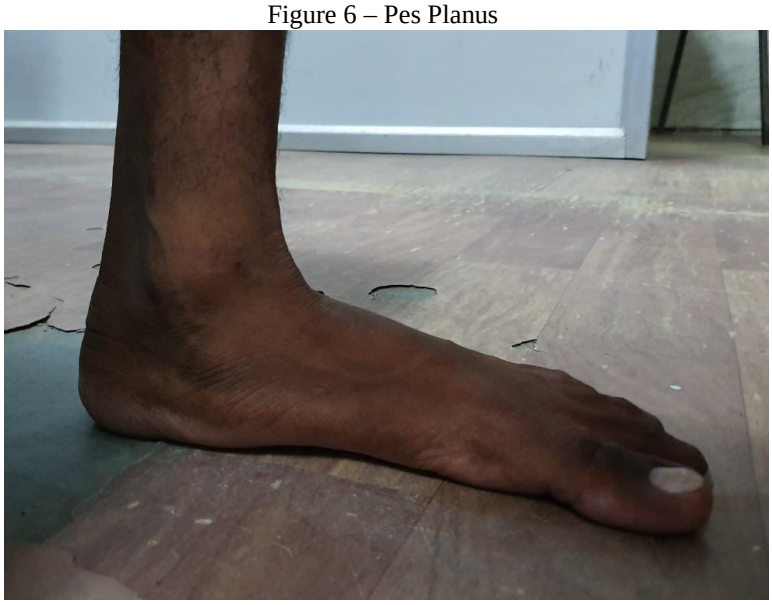
Figure 4 (A)Kyphoscoliosis, (B)Pectus Excavatum



A B

Figure 5 – Thumb Sign(Upper left) , Wrist Sign(Upper Right) , Arachnodactyly(Below)

Figure 6 – Pes Planus



REFERENCES

[1]. Stankovic Babic G, Vujanovic M, Dordevic-Jocic J, Cekic S. Ocular features of Marfan's syndrome. Medicine and Biology. 2008;15(1):37-40

[2]. Wakita, M., et al. "[Statistical analysis of Marfan's Syndrome]." Nippon Ganka Gakkai Zasshi 93.6 (1989): 682-690

[3]. Robinson P, Godfrey M . The molecular genetics of Marfans syndrome and related micro□brillopathies. J Med Genet 2000; 37:9 -25

[4]. Ramirez F, Dietz HC. Marfan syndrome: from molecular pathogenesis to clinical treatment. CurrOpin Genet Dev. 2007 Jun;17(3):252-8. doi: 10.1016/j.gde.2007.04.006. Epub 2007 Apr 27. Erratum in: CurrOpin Genet Dev. 2007 Aug;17(4):367. PMID: 17467262.

[5]. Dean JC. Marfan's syndrome: Clinical Diagnosis and Management. Eur J Hum Genet. 2007;15(7):724-33.

[6]. Rubin SE, Nelson LB. Ocular manifestations of autosomal dominant systemic conditions. Duane's Clinical Ophthalmology on CD –Rom. Philadelphia: Lippincott Williams and Wilkins. 2006;3(58)

[7]. Anteby I, Isaac M, BenEzra D. Hereditary sub-luxated lenses: visual performances and long term follow-up after surgery. Ophthalmology. 2003;110(7):1344-8