Case Report on Marfans Syndrome

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Abstract

Marfan syndrome (MFS) is one of the most common inherited disorders of connective tissue. It is an autosomal dominant condition with a reported incidence of 1 in 3000 to 5000 individuals. There is a wide range of clinical severity associated with MFS with classic ocular, cardiovascular and musculoskeletal abnormalities, while some patients demonstrate significant involvement of the lung, skin and central nervous system. A 11 years old female child came to the ophthalmology OPD with chief complaints of diminution of vision in both eyes, more in right eye since last 2 months. Patient also complains of pain in right eye since last 3 days. On examination patient had a tall stature, arachnodactyly with positive wrist and thumb signs, right eye had superotemporal subluxation of lens,the left eye had temporal subluxation with iridodonesis on autorefractometer patient was having high myopic reading

Keywords: *Marfan syndrome, Connective tissue disorder, radiological and vascular abnormality, case report.*

Introduction

Marfan syndrome is an autosomal dominant systemic disorder of connective tissue[1,2]. Children affected by the Marfan syndrome carry a mutation in one of their two copies of the gene that encodes the connective tissue protein fibrillin-1 (FBN 1).[3] It mostly affects skeleton, lungs, eyes, heart, and the aorta[4]. Affected individuals often are tall and slender, have arachnodactyly, scoliosis, and either a pectus excavatum, pectus carinatum, or ectopia lentis in eyes[5]. The incidence of mitral prolapse in such patients is essentially equal in children and adults of the same sex [6]

Worldwide, the incidence of Marfan syndrome is approximately 7–17/100,000. The incidence of aortic dilatation and mitral prolapse in patients with Marfan's syndrome was essentially equal in children and adults of the same sex.

Material And Method

A 11 years old female child came to ophthalmology OPD with chief complaints of diminution of vision in both eyes since 2 months. Patient also complains of pain in right eye since 3 days.

History of glare present in both eyes.

No history of

- Trauma
- Floaters
- Flashes of light
- Diplopia

Family history: No known family members with Marfan Syndrome.

On general examination

- Blood pressure: 110/80 mmHg
- Stature: tall

On neurological examination patient was fully alert and orientedand had fluent speech. There were no signs of meningeal irritation.

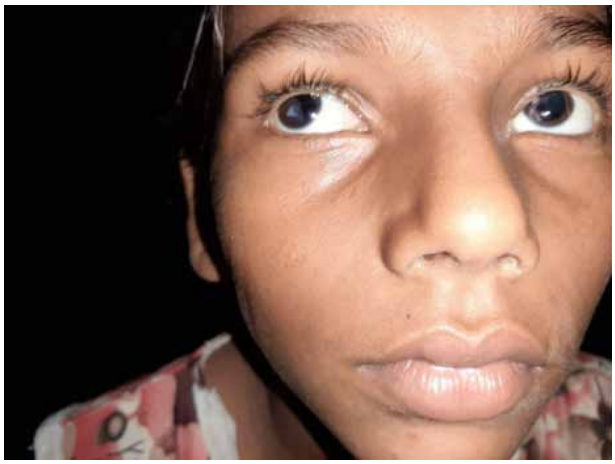
- All cranial nerve examination was normal.
- High arched palate +

Ophthalmic Examination:

	Right Eye	**Left Eye**
Vision	CF 1M-No improvement	6/60p-no improvement
conjunctiva	Normal	Normal
Cornea	Bright	Bright
Anterior chamber	Deep	Deep
Iris	Normal colour pattern	Normal colour pattern
Pupil	Normal size reacting to light No anisocoria and no relative afferent pupillary defect	Normal size reacting to light No anisocoria and no relative afferent pupillary defect
lens	Supero temporal subluxation of lens	Temporal sublaxation +
Extraocular movements	Free and full in all gazes of direction	Free and full in all gazes of direction
Fundus examination	Normal	Normal
Intra-ocular pressure (GAT)	18	20

Pic showing superotemporal sublaxation of lens in right eye

Pic 2. showing high arched palate .



Pic 3 . showing dysmorphic fascial features and also temporal sublaxation of lens in left eye

Plan of Management:

- Lens extraction for ectopia lentis with Contact lens or IOL (anterior chamber, sutured to sclera/iris)
- Regular screen for myopia, amblyopia, strabismus, keratoconus, and glaucoma
- Chest x-ray and 2D- ECHO advised

Discussion

Marfan syndrome is a autosomal dominant genetic condition that causes weakening of connective tissue in the cardiovascular, musculoskeletal and ocular systems. The major ocular abnormality in Marfan syndrome is ectopia lentis (lens subluxation or dislocation). While relatively little is known about the exact mechanism of this ocular pathology in Marfan syndrome, a number of theories have been suggested.

Al (1995) noticed that while fibrillin is located on the surface superior to the capsule and on the ciliary epithelial surface when zonules are attached to normal eyes, Marfan patients lack such position and have irregular ciliary processes with missing or extremely disorganized zonules. This pathology was found to be positively correlated with lens subluxation.

Clinically, ectopia lentis is bilateral in 60-87% of Marfan patients and is stable from childhood. Symptoms consist of blurring of vision, diplopia, and pain. On examination, patients show refractive instability with myopia and astigmatism, iridodonesis, phacodinesis, and a recessed angle. The most common direction of dislocation on examination is superotemporal. In addition, there may be secondary complications from lens movement such as phacolytic uveitis from posterior subluxation of the lens to the vitreous. Retinal tears and detachments are also quite common in patients with Marfan syndrome.

Other ocular manifestations of Marfan syndrome include flattened cornea (causing astigmatism), keratoconus, increased globe length (causing myopia), iris coloboma, cataracts, glaucoma, strabismus, amblyopia, and vascular malformations. Central to each of these findings is the ubiquitous abnormality of fibrillin in the ocular connective tissue of the Marfan patient.

The systemic manifestations of Marfan syndrome are well-studied, the most obvious of which are the musculoskeletal abnormalities. Again, the common thread in the variety of Marfan phenotypes is the weakness or incompetence of the connective tissues due to defects in fibrillin. The Marfan patient is often very tall with long, flexible extremities and marked scoliosis. They exhibit arachnodactyly (spider fingers) with the ability to dramatically encircle the wrist (Walker-Murdoch sign). In addition, they often have pectus excavatum, a high-arched palate, and facial abnormalities. The cardiovascular findings range from mild mitral valve prolapse to severe aortic aneurysm or dissection; the severe cardiovascular complications are the primary causes of mortality among Marfan patients. Pulmonary diseases include apical blebs or spontaneous pneumothorax. Articles on few other rare syndromes[7-9] are available.

Ethical Clearance: Taken from institutional ethics committee.

Source of Funding: Self.

Conflict of Interest: nil.

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