

Bilateral subluxation of the lens in a case of sporadic Marfan syndrome

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Marfan syndrome (MFS) is an autosomal dominant disorder caused by a fibrillin-1 gene mutation (FBN1). Atypical MFS is caused by inactivating mutations in transforming growth factor β receptor (TGF β R). About 30% of cases of MFS do not have a family history. These sporadic cases occur due to de novo gene mutations. We report a case of a 16-year-old female, who presented with progressive diminution of vision in both the eyes for the past 5 years. On ocular examination, her right eye visual acuity was 5/60 and left eye visual acuity was 6/60, with a clear cornea.

Keywords: Sporadic, Lens Subluxation, Marfan Syndrome,

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Dear Editor,

Marfan syndrome (MFS) is an autosomal dominant disorder caused by a fibrillin-1 gene mutation (FBN1) [1]. Atypical MFS is caused by inactivating mutations in transforming growth factor β receptor (TGF β R) [2]. About 30% of cases of MFS do not have a family history. These sporadic cases occur due to de novo gene mutations [3].

The defective fibrillin protein in the connective tissue causes ocular, cardiovascular, musculoskeletal, skin, lung, and nervous system manifestations, which evolves gradually as the age advances. Ocular

Manifestations of MFS include subluxation of the lens, flat cornea, glaucoma, cataract, retinal detachment, and amblyopia [4].

We report a case of a 16-year-old female, who presented with progressive diminution of vision in both the eyes for the past 5 years. On ocular examination, her right eye visual acuity was 5/60 and left eye visual acuity was 6/60, with a clear cornea.

Dilated examination revealed a supero-temporal subluxation of the lens with intact zonules in both the eyes and normal fundus (Figure-1).

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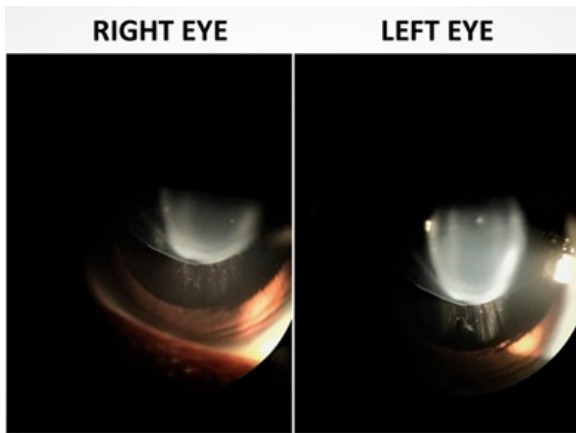


Fig-1: Slit-lamp image of both the eyes showing bilateral supero-temporal subluxation of the lens with intact zonules.

She was tall and her arm span and height ratio was 1.06, and the upper segment lower segment ratio was 0.79. Examination of the cardiovascular system revealed a systolic murmur in the aortic and mitral areas. A 2D-Echocardiogram confirmed aortic and mitral valvular regurgitation with an aortic root diameter of 3.1cm at the sinuses of Valsalva. Her aortic diameter Z-score was 2.23. None of her family members have marfanoid habitus, visual, or cardiac problems. Hence, based on the Revised GHENT nosology, a diagnosis of sporadic Marfan syndrome was made [5]. The patient underwent lensectomy and placement of scleral fixation intraocular lens, stabilized by capsular tension rings in the right eye followed by left eye within 2 weeks. Her vision improved to 6/9 in both the eyes and she is on regular follow-up. Hence, in children and young adults with marfanoid habitus, periodic Ophthalmological screening tests have to be done to diagnose and treat them early.

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