

## CASE REPORT

# MARFAN SYNDROME PRESENTING WITH BILATERAL RETINAL DETACHMENT

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## ABSTRACT

Marfan syndrome is an autosomal dominant systemic disorder of the connective tissue. Marfan syndrome affects most organs and tissues, especially the skeleton, lungs, eyes, heart, and the large blood vessels. Eye involvement may be in the form of retinal detachment which is a potentially dangerous manifestation for its sight threatening nature. We report a case where a 17 year old male developed sudden blindness due to spontaneous bilateral retinal detachment. Examination revealed features of Marfan syndrome and was stamped as a case of Marfan syndrome by Ghent criteria. The point to stress upon is that a young male developing spontaneous retinal detachment, a diagnosis of underlying Marfan syndrome should be kept in mind if appropriate clinical stigmata are present.

**Keywords:** Marfan syndrome, bilateral retinal detachment

## INTRODUCTION

Marfan syndrome is an autosomal dominant systemic disorder of connective tissue. 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) [1]. It mostly affects skeleton, lungs, eyes, heart, and the aorta. Affected individuals often are tall and slender, have arachnodactyly, scoliosis, and either a pectus excavatum, pectus carinatum, aortic dilatation, mitral prolapse [2]. Ocular involvement may be in the form of ectopia lentis, myopia, strabismus, squint, retinal detachment. Although retinal detachment in a case of Marfan syndrome has no special treatment issues, but it has greater chance of recurrence [3].

## CASE REPORT

A tall 17 year old man presented with sudden onset of blurred vision in both eyes to the emergency. No history of trauma to the eyes or head was present and he was previously non diabetic and non hypertensive. No previous history of any ocular ailment was present and he was not suffering from myopia. He was 186cms tall and his arm span length was 197cm (arm span:height > 1.05); reduced upper-to-lower body segment ratio, which was 0.73 (normal- 0.86).

Urgent ophthalmological assessment revealed bilateral complete retinal detachment. However lens dislocation was not noted. B-scan ocular ultrasonography confirmed retinal detachment in the both eyes. General examination showed presence of pectus carinatum with

long slender fingers and high arched palate with malocclusion of teeth with generalised joint laxity. Scoliosis was not present. Cardiovascular assessment suggested presence of aortic regurgitation which was later confirmed as mild aortic root dilation on echocardiography.

The thumb and the little finger overlapped well while wrapping the other wrist (positive Walker's sign) and when enclosed within the clenched fist, the thumb protruded beyond the ulnar border (positive Steinberg sign). Keeping a diagnosis of Marfan syndrome in mind, specific interrogation revealed that a strong family history was present. His grandfather and father was also affected by similar physical features. His elder brother died suddenly of dissecting aneurysm of aorta. Routine blood tests including peripheral blood counts, renal and liver function tests, serum electrolytes were within normal limits. Chest X ray showed no bullae or pneumothorax. Aortic root dilation with no mitral valve abnormalities was noted on Echocardiography. Karyotyping was done and it was found to be normal (46 XY) with no structural or numerical abnormalities. In accordance with Ghent criteria, patient was diagnosed as having Marfan syndrome, complicated by bilateral retinal detachment.

Patient was treated urgently along standard surgical lines by vitreolensctomy with cryotherapy and intraocular tamponade with silicon oil. Patient made a slow but uneventful recovery with best vision 6/24 in both eyes after 3 months.



Figure 1: Positive Walker's Sign



Figure 2: High Arched Palate With Malocclusion Of Teeth



Figure 3: Pectus Carinatum

## DISCUSSION

Marfan syndrome is an inherited connective tissue disorder that is transmitted as an

autosomal dominant trait and is named after the French pediatrician Antonin Bernard Marfan, who first summarized the symptoms in 1892. It is a connective tissue disorder with clinical variability and pleiotropic manifestations. The diagnosis is based on Ghent diagnostic criteria.<sup>[4]</sup> In adults, the combination of the major criteria in two different body systems and minor criteria in the third system amongst the cardiovascular, skeletal, ocular, pulmonary, skin, nervous systems provides the clinical diagnosis in majority of the cases. In children, genotyping may contribute to the diagnosis, especially if family history is negative. The clinical manifestations are due to mutation in the fibrillin-1 (FBN-1) gene located on chromosome 15q21.<sup>[1]</sup> It is inherited in approximately 75% of cases and occurs due to spontaneous mutation in the remaining 25%. More than 150 different mutations of the FBN-1 gene have been isolated, and each family often has a unique genetic mutation for the syndrome.<sup>[1]</sup> This could explain the considerable variability in the clinical presentations of Marfan syndrome. The condition may manifest in the cardiovascular, musculoskeletal and ocular systems. Myxomatous degeneration of aortic valve, lens dislocation, pectus excavatum or carinatum, arachnodactyly, dilatation of aorta, high arched palate with malocclusion of teeth with generalised joint laxity are the classical features of Marfan syndrome. Other musculoskeletal manifestations include scoliosis, dural ectasia, protrusion acetabuli, and ligamentous laxity<sup>[2]</sup>. The fingers can be wrapped completely around the opposing wrist with overlapping (positive Walker's sign) and when enclosed within the clenched fist, the thumb protrudes beyond the ulnar border (positive Steinberg sign). Other system involvement includes respiratory and skin manifesting as bullae and spontaneous pneumothorax and multiple striae in skin. Compared with patients with idiopathic scoliosis, patients with Marfan syndrome tend to have scoliosis

that progresses at a faster rate<sup>[5]</sup>. Progression of skeletal abnormalities, especially scoliosis and anterior chest irregularity, can be dramatic during periods of rapid growth, such as puberty. Evaluation and follow up by an orthopedician is indicated in these cases. Aortic root dilation, aortic regurgitation, mitral valve prolapse, dissection of aorta are main cardiovascular manifestations and also the most common cause of death<sup>[6-7]</sup>. It is essential to identify and correct high refractive error or amblyopia in childhood in order to preserve and maximize visual function. Individuals with the Marfan syndrome are at increased risk for glaucoma, cataract formation, and retinal detachment, even in the absence of ectopia lentis. For this reason, the eye evaluations should be performed every year. This case depicts a dangerous as although not a life-threatening but a permanent sight threatening complication of Marfan syndrome which although does not require any underlying disease specific treatment but only timely appropriate management failing which may render a person permanently blind.

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