MARFAN SYNDROME: A CASE REPORT

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ABSTRACT

We report on a patient from Peshawar, Pakistan diagnosed with Marfan syndrome. Our reported case of Marfan syndrome is exceptional due to the myriad of musculoskeletal, cardiovascular, and clinical signs present in a single patient.

Keywords: Marfan syndrome; Ocular, Musculoskeletal, Cardiovascular.

INTRODUCTION

Marfan syndrome is one of the most commonly occurring genetic disorders of the connective tissue. It presents in an autosomal dominant pattern with a mutation in the FBN1 gene causing the formation of defective fibrillin. 75% of cases are familial, while 25% are due to de novo mutations. The reported incidence is about 1 in 3000 to 5000 individuals[1]. Marfan syndrome is associated with a constellation of clinical signs including ocular, cardiovascular, and musculoskeletal abnormalities. Diagnosis of the condition is based on Ghent nosology which is divided into two parts based on the presence or absence of family history. Greater diagnostic value is given to the clinical signs of ectopia lentis and aortic root dilatation[2].

Case Presentation

A 19-year-old male presented to the Ophthalmology outpatient department, Hayatabad Medical Complex, Peshawar with the complaint of a significant decrease in vision. He was unable to comprehend writing on the class board. Visual impairment was gradually progressive with the recent development of glare and headache. He was a previously diagnosed case of Marfan syndrome and had undergone cardiac surgery for an atrial septal defect. (figure 1) The patient did not have a family history of Marfan syndrome. On general examination, he was a tall (6 feet 6 inches) thin male sitting comfortably. He had characteristic musculoskeletal features of Marfan syndrome; exophthalmos, malar hypoplasia, pectus carinatum, scoliosis, increased arm span, arachnodactyly, and positive wrist and thumb signs (figure 2). Ophthalmic examination revealed a left

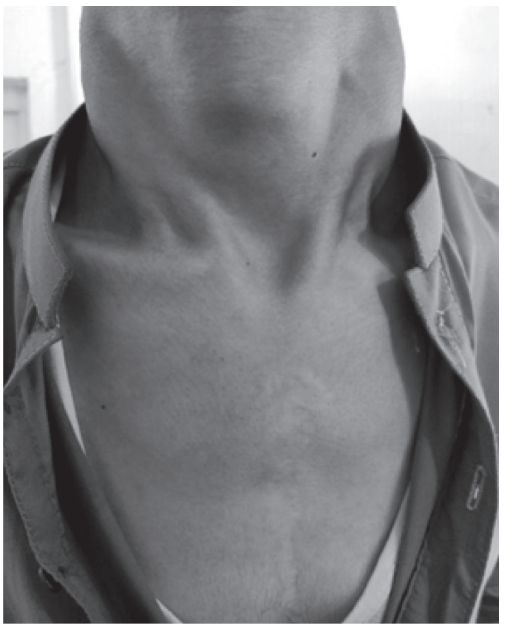

Figure 1

Figure 2

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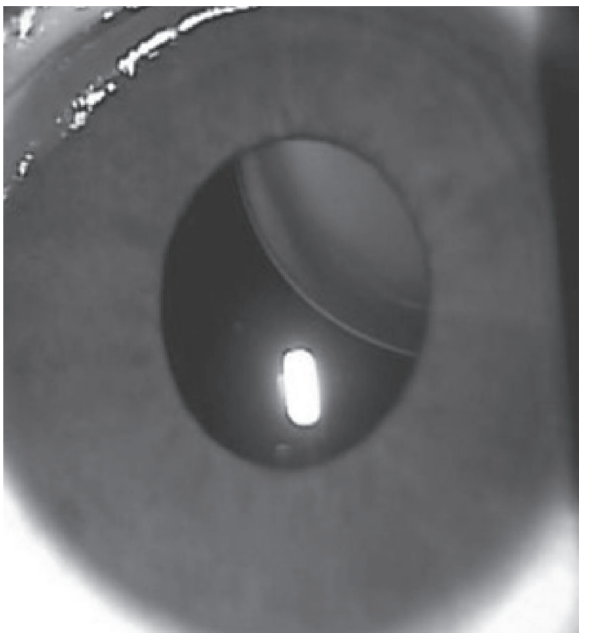

Figure 3

lens superior subluxation, iridodonesis, and slight improvement of vision with refraction (-10.00 glasses). The right eye was pthysical due to old retinal detachment (figure 3). All laboratory tests were within normal limits. The patient's previous diagnosis of Marfan syndrome was confirmed with Ghent nosology diagnostic criteria of aortic root dilatation Z score 6 and ectopia lentis in the absence of a family history of Marfan syndrome.

DISCUSSION

Marfan syndrome is an autosomal dominant genetic disorder due to a structural defect in the fibrillin-1 protein which functions as a scaffold for elastin[3]. Mutations in the FBN1 gene result in this connective tissue disease. Our patient did not undergo genetic testing and family history was negative for Marfan syndrome. He presented with characteristic features of the syndrome such as cardiac septal defects, ectopia lentils, and various musculoskeletal abnormalities. Disease mortality is linked to cardiac complications; most frequently aortic dissection[4].

Marfan syndrome is associated with multiple ocular abnormalities. The pathognomonic abnormality is ectopia lentis (lens subluxation) which occurs in about 10% of patients. This occurs because of the weakness of ciliary zonules; the connective tissue ligaments which suspend the lens within the eye. The most frequently affected are inferior zonules, resulting in upward and outward lens subluxation[5]. Other ocular features may include increased axial length, myopia, corneal flatness, and strabismus. Affected individuals are also at a higher risk of developing cataracts and glaucoma.

Learning Points

Although Marfan syndrome is one of the most common connective tissue disorders our patient deserves special consideration because of the distinctive array of pleiotropic effects; ocular, vascular, cardiac, and musculoskeletal. These patients are in greater need of routine clinical evaluation due to the higher risk of multidisciplinary complications.

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