



## Case Report

### Marfan Syndrome: A Case Report

Mohammad Golam Rob Mahmud<sup>1</sup>

<sup>1</sup>Associate Professor, Department of Medicine, Jalalabad Ragib-Rabeya Medical College, Sylhet.

#### ABSTRACT

*Marfan syndrome is a rare autosomal dominant, multisystem connective tissue disease, caused by mutation in the extra cellular matrix protein fibrillin-1 gene located on chromosome 15. It has variable phenotypic expression with a reported incidence of 1 in 3000 to 5000 individuals. There is a wide range of clinical severity associated with Marfan syndrome with classic ocular, cardiovascular and musculo-skeletal abnormalities. The diagnosis of Marfan syndrome relies essentially on the fulfilment of clinical diagnostic criteria. We report a case of an 18 years old male with positive family history including lean and thin body stature, joint hypermobility, high arched palate, pectus excavatum, total arm span more than total body height (1.11:1), lower segment greater than upper segment (1.34:1) and mitral valve prolapse.*

**Keywords:** Marfan syndrome, Mitral value prolapse, Connective tissue disease.

[Jalalabad Med J 2023; 20 (1): 31-34]; DOI: <https://doi.org/10.3329/jmj.v20i1.79416>

#### INTRODUCTION

Marfan syndrome is a rare (0.02% of the population) inherited autosomal dominant disorder of connective tissue with a high risk of aortic aneurysm and dissection. It results from mutations in the FBN1 genes, which lead to a lack of fibrillin-1 and a decrease in microfibril production. This disrupts the mechanical integrity of connective tissue, giving rise to a wide range of clinical features<sup>1</sup>. The syndrome is inherited as an autosomal dominant trait with complete penetrance but with phenotypic expression that varies considerably both between and within families. About 75% of people with classic Marfan syndrome have a positive family history, and 25-26% of the cases have no family history<sup>2</sup>.

Affected individuals develop varying patterns of organ involvement, including the cardiovascular, ocular, skeletal, lung, skin, and dura. In classical Marfan syndrome, many manifestations present during puberty, but severe complications rarely develop before

adulthood. Such complications include severe scoliosis or pectus excavatum, spontaneous pneumothorax, retinal detachment or sight-threatening glaucoma resulting from dislocated lenses<sup>3</sup>. Further, less specific features include a high arched palate, crowding of the teeth, and skin striae<sup>2</sup>. The typical life expectancy of people with Marfan syndrome is close to 70 years, despite the increased risk of cardiovascular issues connected to the condition. The life expectancy for this syndrome has increased to greater than 25% since 1972<sup>4</sup>. Treatment with b-blockers or angiotensin receptor blockers may reduce the risk of aortic dilatation and should be given to all patients with Marfan syndrome. Activities that are associated with increases in cardiac output are best avoided. Surgery to replace the aortic root can be performed in patients with progressive aortic dilatation<sup>1</sup>. Here, I am going to discuss a case that had some classic symptoms of Marfan syndrome.

#### CASE REPORT

An 18-year-old Bangladeshi young male patient had been brought to the hospital with complaints of palpitation and shortness of breath. His parents reported that he had been healthy up to the age of 10, but after that, he started to get a little bit taller and

#### Address of Correspondence:

Dr. Mohammad Golam Rob Mahmud, Associate Professor,  
Department of Medicine, Jalalabad Ragib-Rabeya Medical  
College, Sylhet. Email: [shuyebgolamrob@gmail.com](mailto:shuyebgolamrob@gmail.com).

thinner than other children of his age. His parents and one sister were healthy, but his younger brother had a similar presentation. A clinical examination revealed that he looked lean and thin (Figure-1). His pulse was 96 beats per minute with a normal rhythm, and his blood pressure was 110/70 mmHg. He had a high arched palate (Figure-2) and pectus excavatum (Figure-3). His joints were hypermobile (Figure-4), thumb and little fingers gripped more than his wrist, the Steinberg sign was positive (Figure-5) and arachnodactyly was present (Figure-6).

The total height of the patient was 167 cm, and the arm span was 186 cm. The ratio of arm span to height was 1.11:1. His lower segment length from the public

symphysis to the heel was 96 cm, and his upper segment length from the vault to the public symphysis was 70 cm. The ratio of the lower segment to the upper segment was 1.34:1. His cardiovascular system examination revealed that the 1<sup>st</sup> and 2<sup>nd</sup> heart sounds appeared normal and there was a late systolic murmur in the left 3<sup>rd</sup> and 4<sup>th</sup> intercostal spaces. There was no abnormality on the ophthalmological or spine examinations. Echocardiography revealed a relatively dilated aorta and mitral valve prolapse (MVP) (Figure-7). The patient was treated with oral propranolol and referred to National Institute of Cardiovascular Diseases (NICVD), Dhaka for further management.



**Figure-1:** Lean and thin body structure



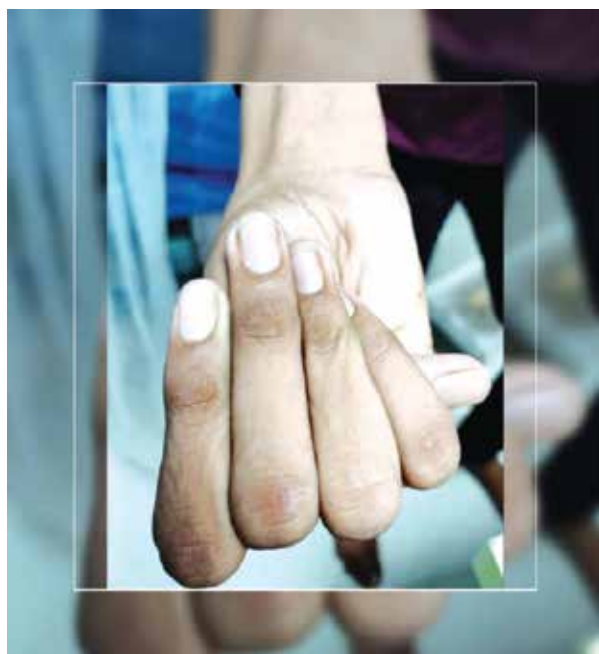
**Figure-3:** Pectus excavatum



**Figure-2:** High arched palate



**Figure-4:** Hypermobile joint



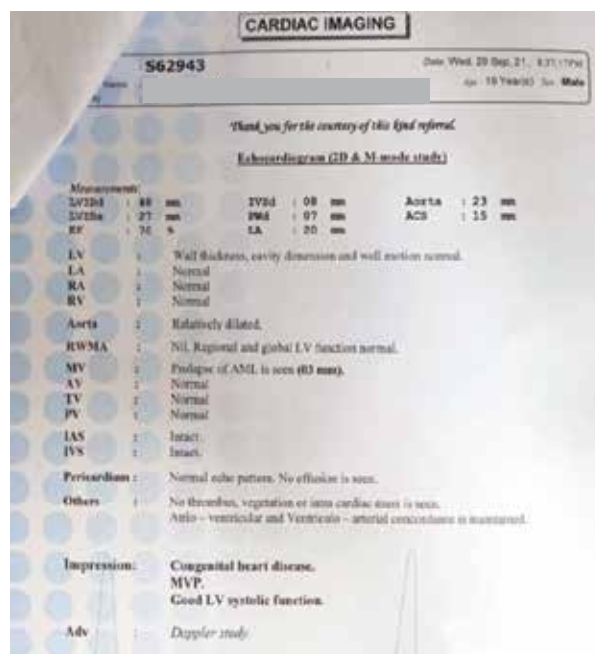
**Figure-5: Steinberg sign**



**Figure-6: Arachnodactyly**

## DISCUSSION

Fibrillin is an essential component of the microfibrillar system that serves as a scaffold for elastogenesis. A mutation in the gene FBN1 that codes for the protein fibrillin-1 is linked to the classic form of Marfan syndrome. The pathophysiological outcomes of the degeneration of elastic fibres in Marfan syndrome seem to explain the majority of manifestations of this condition. The primary and most significant effects of elastin degradation include stiffness and decreased



**Figure-7: Echocardiogram report**

aortic distensibility in response to elevated pulse pressure<sup>4</sup>. Another theory has recently emerged in an effort to clarify the pathophysiology of Marfan syndrome. The cytokine transforming growth factor (TGFB), which controls cell morphogenesis, may play a role in the phenotype of Marfan syndrome. Abnormal fibrillin prevents TGFB's latent precursor from being sequestered, which leads to an excessive amount of TGFB activation and thus produces the phenotypical manifestations of Marfan syndrome<sup>6</sup>.

A scoring system that incorporates multiple diagnostic items is necessary to diagnose Marfan syndrome because there is no single molecular test that can do so. The so-called Ghent nosology subdivides diagnostic features into "Major Criteria" "Minor Criteria" "Organ involvement" and manifestations that only in combination with other manifestations constitute a "Major" or "Minor" criterion<sup>5</sup>. Individuals without a family history of Marfan syndrome require major criteria in at least two different organ systems and involvement of a third organ system. Marfan syndrome or cases with a positive family history require one major criterion and involvement of an additional organ to establish Marfan syndrome<sup>7,8</sup>.

Major criteria that were present in this case were an arm-span to height ratio >1.05, positive wrist and thumb sign, and a first-degree relative who independently meets Marfan criteria. Minor criteria were pectus excavatum not requiring surgery, joint hypermobility, a high-arched palate, and mitral valve prolapse.

## CONCLUSION

Marfan syndrome is a common inherited connective tissue disease with diverse mode of presentation. Mortality of the disease is related to ocular and cardiac complications. Cardiovascular and ocular evaluations should be performed in all patient of Marfan syndrome along with skeletal survey.

## REFERENCE

1. Penman ID, Ralston SH, Strachan MWJ, Hobson RP. Cardiology. In: Newby DE, Grubb NR, editors. Davidson principles and practice of medicine. 24<sup>th</sup> ed. London: Elsevier; 2022. p 445-6.
2. Dean JCS. Management of Marfan syndrome. *Heart* 2002; 88 (1): 97-103.
3. Silverman DI, Burton KJ, Gnay J, Bosner MS, Kouchoukos NT, Roman MJ, et al. Life expectancy in the Marfan syndrome. *Am J Cardiol* 1995; 75 (2): 157-60.
4. Kumar K. Life expectancy of someone with Marfan syndrome. *Medicine Net*; 2022 [Cited 4 January 2023]. Available from: [https://www.medicinenet.com/25\\_heart-healthy\\_foods\\_pictures\\_slideshow/article.htm](https://www.medicinenet.com/25_heart-healthy_foods_pictures_slideshow/article.htm)
5. Loeys BL, Dietz HC, Braverman AC, Callewaert BL, De Backer J, Devereux RB, et al. The revised Ghent nosology for the Marfan syndrome. *J Med Genet* 2010; 47 (7): 476-85.
6. Lacro RV, Dietz HC, Wruck LM, Bradley TJ, Colan SD, Devereux RB, et al. Rationale and design of a randomized clinical trial of beta blocker therapy (atenolol) versus angiotensin II receptor blocker therapy (losartan) in individuals with Marfan syndrome. *Am Heart J* 2007; 154 (4): 624-31.
7. De Paepe A, Devereux RB, Dietz HC, Hennekam RC, Pyeritz RE. Revised diagnostic criteria for the Marfan syndrome. *Am J Med Genet* 1996; 62 (4): 417-26.
8. von Kodolitsch Y, Robinson PN. Marfan syndrome: an update of genetics, medical and surgical management. *Heart* 2007; 93 (6): 755-60.