

Research Journal of Pharmaceutical, Biological and Chemical Sciences

Inherited Marfan Syndrome.

T Vidhya, Gokul D Yatheendranathan*, M Divya, S Raashida, and A Saranya.

Sree Balaji Medical College & Hospital, Bharath University, No.7, CLC Works Road, Chrompet, Chennai – 600044, Tamil Nadu, India.

ABSTRACT

Marfan syndrome named after Antoine Marfan, a French paediatrician who first described the condition in 1896 is an autosomal dominant genetic disorder caused by the misfolding of the protein fibrillin-1 which is encoded by the gene FBN1.^[1,2,3] Here we present a case report of 6 year old developmentally normal girl child who presented to our OPD for fever and was found to have features characteristic of Marfan syndrome. What was unique was the fact that her mother and her maternal grandfather also showed features of marfan syndrome.

Keywords: Marfan syndrome, genetic, tall stature, thin limbs, pectusexcavatum, pes planus, arachnodactyly, mitral valve regurgitation, high arched palate.

**Corresponding author*

Case Presentation

A 6 year old developmentally normal girl child came to our OPD with complaints of fever for two days which was intermittent and high grade. She was accompanied by her mother who was tall (172cm) with thin long limbs. On examination of the child she was found to weigh 21 kg and her height was 120 cm. Her arm span was 126 cm; arm span to height ratio was 1.06 and upper segment to lower segment ratio was 1.03. On head to foot examination, her head was normal, eyes had a bluish sclera which was normal for her age, her neck, skin and spine was normal. Her chest showed pectusexcavatum (fig.I). Her limbs were slender, could be easily put into hyper extension (fig. III) and there was medial displacement of the medial malleolus (fig. II). She had long fingers and toes. She was found to have joint hypermobility which was 4/9 on the Beighton Scale. Right and left elbow extension maxed at 190 degree and right and left knee extension at 200 degrees. On examination of the CVS the apex beat was felt in the 4th intercostal space $\frac{1}{2}$ inch lateral to the mid clavicular line. First & second heart sounds were heard along with a short systolic murmur in the pulmonary and mitral areas, grade 3/6 (Levine grading scale). Her respiratory and central nervous system was normal and abdomen soft with no organomegaly, free fluid or any palpable mass. An Echo scan was done which revealed a mild Mitral valve regurgitation. Her mother aged 36 was also evaluated and found to have features of marfan syndrome. She was tall (172 cm) with thin long limbs, an arm span to height ratio of 1.1, a high arched palate, myopia, arachnodactyly, pes planus and mitral valve regurgitation. The mother's father was also examined and found to have similar features including pectusexcavatum. The fever was evaluated and was determined to be of viral etiology. She was admitted and treated symptomatically for the fever. She recovered in 5 days and was later discharged.



Figure I: Pectusexcavatum of the child



Figure II: Medial dislocation of the medial malleolus



Figure III:Hyper extension of the elbow joint

DISCUSSION

The estimated incidence of MFS ranges from 1 in 5,000 to 2-3 in 10,000 persons. The mutation in the fibrillin gene causes pleiotropic effects; thus, a wide range of phenotypic features is derived from a single gene mutation. Several other diseases have presentations similar to MFS, making it exceedingly difficult to determine the exact incidence [1,2].

In most cases, Marfan's is recognized medically from a person's physical characteristics. The connective tissue abnormality usually affects the tissues of the skeletal system, the eyes and the cardiovascular system [3-5]. People with Marfan's tend to be considerably taller than others.

The lower half of the body is especially long and the arm span is usually greater than the person's height. Fingers tend to be long and spidery; the spine is often curved (a condition called scoliosis); and the chest bone can bend inward or outward [3,4, 9-11]. Other characteristics include leanness, small muscle mass, loose-jointedness, flat feet, backward curve of the legs at the knees and a narrow mouth and small jaw. People with Marfan's tend to be nearsighted.

The most serious complications are loss of vision because of dislocation of the eyes' lenses or detached retinas [5], and weakening of the aorta rendering it more prone to rupture, which can be fatal. Both problems underscore the importance of avoiding vigorous physical activities [6,7,13].

The diagnosis is tricky and must be made by an expert since many people without Marfan's may share some or even all these characteristics.

To reduce the risk of such complications, patients are commonly treated with a drug like propranolol (Inderal) to reduce stress on the aorta by decreasing the force of heart-muscle contractions. They should also undergo annual cardiovascular examinations called echocardiograms to monitor the condition of the aorta and detect other heart problems [7,8].

Many Marfan patients also have weakened heart valves and must take antibiotics before undergoing dental work or any other procedure that could spew infectious organisms into the bloodstream and cause a cardiac infection [14-16]. Frequent eye examinations are also advisable, and patients must seek eye treatment without delay when they notice any visual abnormality [17].

REFERENCES

- [1] Dietz HC, Cutting GR, Pyeritz RE, et al. *Nature* 1998;352(6333):337-9.
- [2] Ammash NM, Sundt TM, Connolly HM. *Curr Probl Cardiol* 2008;33(1):7-39.
- [3] Tachdjian MO. Marfan's syndrome. In: Herring JA, ed. *Tachdjian's Pediatric Orthopaedics*. 3rd ed. Philadelphia, Pa: WB Saunders; 829-37, (1990)
- [4] McKusick VA. *Circulation* 1955;11(3):321-42.
- [5] Maumenee IH. *Trans Am Ophthalmol Soc* 1981;79:684-733.
- [6] Judge DP, Dietz HC. *Annu Rev Med* 2008;59:43-59.
- [7] Gould RA, Sinha R, Aziz H, Rouf R, Dietz HC 3rd, Judge DP, et al. *PLoS One* 2012;7(9):e44639.
- [8] Matt P, Habashi J, Carrel T, et al. *J Thorac Cardiovasc Surg* 2008;35(2):389-94.
- [9] Demetracopoulos CA, Sponseller PD. *OrthopClin North Am* 2007;38(4):563-72.
- [10] Sponseller PD, Hobbs W, Riley LH 3rd, Pyeritz RE. *J Bone Joint Surg Am* 1977;77(6):867-76.
- [11] Robins PR, Moe JH, Winter RB. *J Bone Joint Surg Am* 1975;57(3):358-68.
- [12] Mommertz G, Sigala F, Langer S, et al. *Eur J VascEndovasc Surg* 2008;35(2):181-6.
- [13] Murdoch JL, Walker BA, Halpern BL, Kuzma JW, McKusick VA. *N Engl J Med* 1972;86(15):804-8.
- [14] Cañadas V, Vilacosta I, Bruna I, Fuster V. *Nat Rev Cardiol* 2010;30.
- [15] Iams HD. *Curr Sports Med Rep* 2010;9(2):93-8.
- [16] Sakai LY, Keene DR, Engvall E. *J Cell Biol* 1986;03(6 pt 1):2499-509.
- [17] http://www.nhlbi.nih.gov/health/dci/Diseases/mar/mar_diagnosis.html.