

Marfan Syndrome: A Case Report

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Abstract: Marfan syndrome is an autosomal dominant systemic disorder of the 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. Marfan syndrome affects most organs and tissues, especially the skeleton, lungs, eyes, heart, and the large blood vessel that distributes blood from the heart to the rest of the body. I will report a case of Marfan syndrome which was incidentally diagnosed in a 9-year-old male presenting with anorexia after routine physical examination revealed positive Steinberg (thumb) and Walker-Murdoch (wrist) signs. Subsequent diagnostic workup confirmed aortic root dilation (Z-score +2.8) and bilateral ectopia lentis. The Revised Ghent Nosology criteria alongside a confirmed FBN1 gene mutation. This case report highlights the clinical utility of physical examination during routine pediatric evaluations.

Keywords: Marfan Syndrome, FBN1 Gene Mutation, Ghent Nosology, Ectopia Lentis, Pediatric Screening.

1. Introduction

Marfan syndrome is an autosomal dominant systemic disorder of connective tissue [1]. 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) [2]. It mostly affects skeleton, lungs, eyes, heart, and the aorta [3]. Affected individuals often are tall and slender, have arachnodactyly, scoliosis, and either a pectus excavatum, pectus carinatum, or ectopia lentis in eyes [4]. The incidence of mitral prolapse in such patients is essentially equal in children and adults of the same sex [5]. Worldwide, the incidence of Marfan syndrome is approximately 7–17/100,000 [6]. The oral cavity shows high-arched palate that result from a narrow maxilla [7].

2. Case Presentation

A 9-year-old male presented to the pediatric outpatient clinic accompanied by his mother with history of poor appetite (anorexia) and failure to gain weight appropriately. The past medical, developmental, and family histories were unremarkable, with no documented inheritance of sudden cardiac death, early-onset aneurysms, or severe visual impairments. Physical examination revealed a tall, slender child with an ectomorphic body habitus, a height in the 97th percentile, and a weight in the 15th percentile for his age and sex. His vital signs were stable, and systemic cardiopulmonary auscultation was completely normal. However, targeted skeletal evaluation demonstrated prominent markers of connective tissue laxity, including a positive Steinberg sign (thumb sign) [Figure 1], a positive Walker-Murdoch sign (wrist sign) [Figure 2], mild pectus excavatum, bilateral arachnodactyly [Figure 3], and an increased arm-span-to-height ratio of 1.06.

Diagnostic Workup & Investigations: Given the distinct positive skeletal signs, a systemic evaluation for Marfan syndrome was initiated:

- Echocardiography: Revealed an aortic root Z-score of +2.8, indicating mild aortic root dilation, without evidence of aortic regurgitation or mitral valve prolapse.

- Ophthalmologic Evaluation: Slit-lamp examination confirmed bilateral ectopia lentis (superior-temporal subluxation of the crystalline lenses).
- Genetic Testing: Subsequent sequence analysis identified a pathogenic missense mutation in the FBN1 gene, confirming the clinical diagnosis.

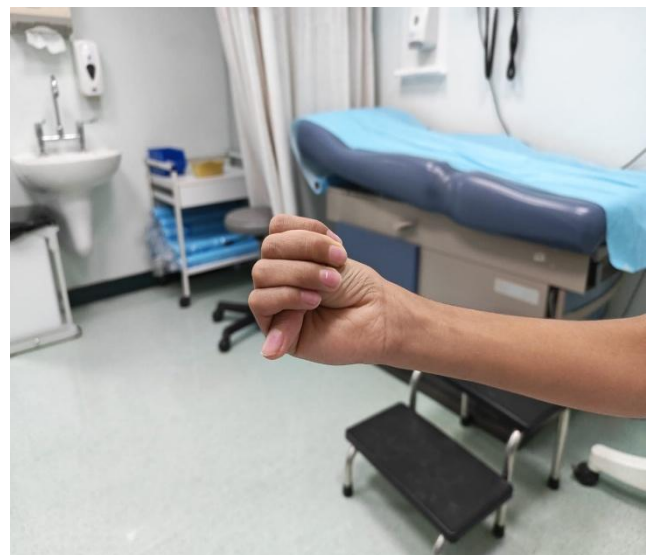


Figure 1: Positive Steinberg sign (thumb sign)



Figure 2: Positive Walker-Murdoch sign (wrist sign)

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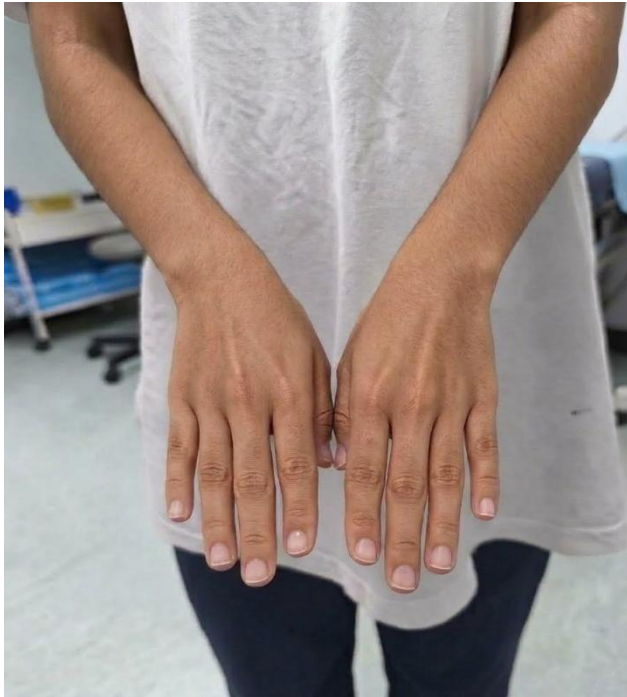


Figure 3: Bilateral arachnodactyly

3. Discussion

The patient's initial complaint of anorexia was entirely unrelated to the underlying genetic pathology, serving instead as an incidental trigger for medical evaluation. According to the Revised Ghent Nosology [8], the coexistence of aortic root dilation ($Z\text{-score} \geq 2$) and ectopia lentis is sufficient for a definitive diagnosis of Marfan syndrome [9], even in the absence of a family history. The presence of positive thumb and wrist signs contributed significantly to a high systemic score, acting as highly sensitive, cost-effective screening features at the bedside.

Diagnosing Marfan syndrome during childhood presents unique challenges due to the age-dependent penetrance of its clinical manifestations, meaning skeletal features and aortic dimensions often progress through adolescence. Tracking the aortic root dimension via body surface area-adjusted $Z\text{-scores}$ is essential in pediatric cohorts, as absolute diameters fail to account for somatic growth. For children under the age of 20 without a family history, an aortic root $Z\text{-score} \geq 2.0$ coupled with ectopia lentis or a pathogenic *FBN1* variant meets the rigorous thresholds of the Ghent criteria to prevent premature or misdiagnosis.

The identification of a pathogenic missense mutation in the *FBN1* gene confirms the diagnosis, with up to 95% of classic Marfan syndrome cases arising from mutations in this specific locus encoding fibrillin-1. Genetic testing plays an increasingly integral part in differentiating Marfan syndrome from overlapping conditions with similar skeletal phenotypes, such as Loeys-Dietz syndrome, Shprintzen-Goldberg syndrome, or Ehlers-Danlos syndrome, which carry distinct vascular risks. Early diagnosis remains a vital modifier of long-term prognosis; standard guidelines advise regular, proactive imaging surveillance alongside prophylactic pharmacological protection using beta-blockers or

angiotensin receptor blockers to mitigate catastrophic aortic dissection risks.

4. Conclusion

This case emphasizes the vital importance of performing comprehensive physical examinations in pediatric patients presenting with non-specific constitutional symptoms. The early, incidental identification of positive wrist and thumb signs permitted timely cardiovascular evaluation and intervention, mitigating the risk of life-threatening complications.

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