VOLUME - 15, ISSUE - 04, APRIL - 2026 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Ophthalmology

CLINICAL SPECTRUM OF FAMILIAL MARFAN SYNDROME: A CASE REPORT

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ABSTRACT Marfan syndrome is an autosomal dominant connective tissue disorder caused by mutations in the fibrillin-1 (FBN1) gene, resulting in multisystem involvement predominantly affecting the ocular, skeletal, and cardiovascular systems. This case report describes a 13-year-old male who presented with progressive diminution of vision and was subsequently diagnosed with bilateral ectopia lentis and high myopia. Detailed systemic examination revealed classical skeletal features including arachnodactyly, pes planus, high arched palate, and positive Steinberg (thumb) and Walker-Murdoch (wrist) signs. Anthropometric assessment demonstrated an increased arm span to height ratio, further supporting the diagnosis. Family screening revealed similar findings in the father and cousins, confirming autosomal dominant inheritance across two generations. The diagnosis was established using the revised Ghent criteria. Early recognition of ocular signs by ophthalmologists is critical, as cardiovascular complications such as aortic root dilatation and dissection are the leading causes of mortality. This case highlights the importance of early diagnosis, multidisciplinary evaluation, and family screening in preventing life-threatening complications.

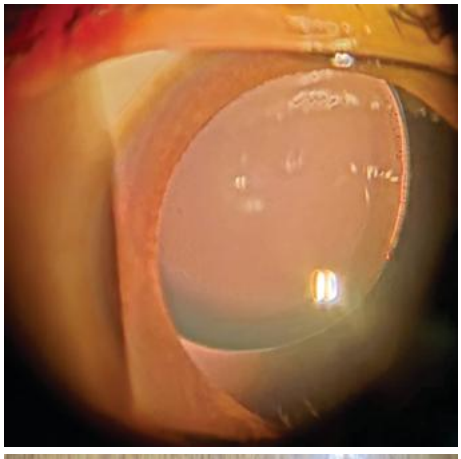
KEYWORDS : Marfan Syndrome, Ectopia Lentis, High Myopia, Arachnodactyly, Ghent Criteria, Familial Disorder

INTRODUCTION

Marfan syndrome is a systemic connective tissue disorder with an estimated prevalence of 1 in 5000 individuals worldwide. It is caused by mutations in the FBN1 gene, which encodes fibrillin-1, an essential component of extracellular microfibrils. Defective fibrillin leads to structural weakness in connective tissue and dysregulation of transforming growth factor-beta (TGF-β) signaling, contributing to the multisystem manifestations of the disease. The condition follows an autosomal dominant inheritance pattern with variable expressivity. Ocular manifestations are among the earliest and most consistent features, with ectopia lentis being present in approximately 60–80% of cases. Other ocular findings include high myopia, retinal detachment, and early cataract formation. Skeletal abnormalities include tall stature, arachnodactyly, joint laxity, scoliosis, pectus deformities, and increased arm span relative to height. Cardiovascular involvement, particularly aortic root dilatation and dissection, represents the most serious complication and is the primary cause of mortality. The revised Ghent criteria provide a standardized diagnostic framework incorporating aortic root measurements, ectopia lentis, systemic features, and family history. Early diagnosis is crucial to allow regular surveillance, timely intervention, and genetic counseling. Ophthalmologists often play a key role in identifying the disease due to early ocular presentation, making awareness of these features essential.

Case Discussion

A 13-year-old male presented to the ophthalmology outpatient department with complaints of progressive diminution of vision in both eyes. There was no history of trauma, ocular surgery, or systemic illness. On examination, best-corrected visual acuity was reduced bilaterally, and refraction revealed high myopia. Slit lamp examination demonstrated bilateral ectopia lentis with superotemporal subluxation of the crystalline lens. Stretched and visible zonular fibers were noted, confirming lens instability. Fundus examination was within normal limits. Systemic examination revealed characteristic skeletal features including arachnodactyly, positive Steinberg (thumb) sign, and Walker-Murdoch (wrist) sign. The patient also had a high arched palate and pes planus. Anthropometric assessment showed an increased arm span to height ratio of 1.052, consistent with Marfanoid habitus. Cardiovascular examination was unremarkable; however, cardiology referral was advised for echocardiographic evaluation to assess aortic root dimensions. Family history revealed similar features in the father and two cousins, suggesting autosomal dominant inheritance. On evaluation, they also demonstrated ectopia lentis and skeletal abnormalities. A pedigree chart confirmed involvement across two generations. Based on the revised Ghent criteria, the presence of ectopia lentis and positive family history was sufficient for diagnosis of Marfan syndrome. The patient was advised regular ophthalmic follow-up, refractive correction, and protective measures, along with cardiology evaluation and genetic counseling.



DISCUSSION

Marfan syndrome is a clinically heterogeneous disorder requiring a high index of suspicion for diagnosis. The revised Ghent criteria emphasize the importance of aortic root dilatation and ectopia lentis as major diagnostic features. In

this case, ocular findings played a pivotal role in diagnosis, highlighting the importance of ophthalmologists in early detection. Ectopia lentis results from weakness of zonular fibers due to abnormal fibrillin, leading to lens subluxation, typically in a superotemporal direction. High myopia is also frequently associated and contributes to visual impairment. Early diagnosis allows timely monitoring of cardiovascular complications, particularly aortic aneurysm and dissection, which significantly increase mortality if untreated. Management requires a multidisciplinary approach involving ophthalmology, cardiology, orthopedics, and genetics. Family screening is essential due to autosomal dominant inheritance, enabling early identification of affected individuals. Genetic counseling should be offered to all families to discuss inheritance patterns, prognosis, and reproductive options.

CONCLUSION

This case underscores the importance of recognizing ocular manifestations such as ectopia lentis as early indicators of systemic connective tissue disorders. Prompt diagnosis using the revised Ghent criteria facilitated early referral for cardiovascular evaluation and identification of additional affected family members. Regular follow-up and multi-disciplinary management are essential to prevent life-threatening complications. Ophthalmologists should maintain a high index of suspicion in patients presenting with high myopia and lens subluxation. Early diagnosis, family screening, and appropriate intervention significantly improve prognosis and reduce morbidity and mortality associated with Marfan syndrome.

REFERENCES (Vancouver)

1. Loeys BL, Dietz HC, Braverman AC, et al. The revised Ghent nosology for Marfan syndrome. J Med Genet. 2010;47(7):476-485.
2. Dietz HC. Marfan Syndrome. GeneReviews. 2017.
3. Judge DP, Dietz HC. Marfan's syndrome. Lancet. 2005;366(9501):1965-1976.
4. Dean JC. Marfan syndrome: clinical diagnosis and management. Eur J Hum Genet. 2007;15(7):724-733.
5. Faivre L, Collod-Beroud G, Loeys BL, et al. Effect of mutation type in FBN1. Eur J Hum Genet. 2007;15(4):415-422.
6. Pyeritz RE. Marfan syndrome: improved clinical history. N Engl J Med. 2014;371:1189-1191.