Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Ophtalmology

Ectopia Lentis in an Adult Revealing Weill–Marchesani Syndrome

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DOI: https://doi.org/10.36347/sjmcr.2026.v14i08.031 | **Received:** 22.06.2026 | **Accepted:** 14.08.2026 | **Published:** 24.08.2026

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Abstract | **Case Report**

We report the case of a 23-year-old male patient, born of a first-degree consanguineous union, who presented with a history of bilateral visual impairment since childhood and a progressive decline in visual acuity. Ophthalmologic examination revealed bilateral ectopia lentis associated with microspherophakia, zonular dehiscence, and elevated intraocular pressure. Systemic evaluation uncovered multiple clinical features consistent with Weill-Marchesani syndrome, a rare connective tissue disorder. The patient underwent phacophagia with implantation of an iris-fixated intraocular lens, followed by visual rehabilitation and physiotherapy aimed at preventing long-term complications associated with the syndrome.

Keywords: Weill–Marchesani syndrome, microspherophakia, ectopia lentis, glaucoma, iris-fixated intraocular lens.

INTRODUCTION

Weill–Marchesani syndrome (WMS) is a rare congenital disorder characterized by skeletal abnormalities and distinctive ocular manifestations, including microspherophakia, ectopia lentis, and glaucoma. Cardiovascular abnormalities may also be present and are sometimes associated with psychomotor developmental delay.

CASE REPORT

A 23-year-old male patient, with bilateral visual impairment since childhood, born to first-degree consanguineous parents, presented to the emergency department with progressive bilateral visual loss. Ophthalmological examination revealed a visual acuity of hand movements in the right eye and 5/10 in the left eye, corrected with −17.50 D sphere.

Anterior segment examination showed, bilaterally, a clear cornea, an irregular anterior chamber, iridophacodonesis, microspherophakia, and lens ectopia associated with zonular rupture involving more than two quadrants. Corrected intraocular pressure was 35 mmHg.

Gonioscopy revealed bilateral narrow iridocorneal angles. Fundus examination showed complete optic disc cupping in the right eye and a cup-to-disc ratio of 0.6 in the left eye.

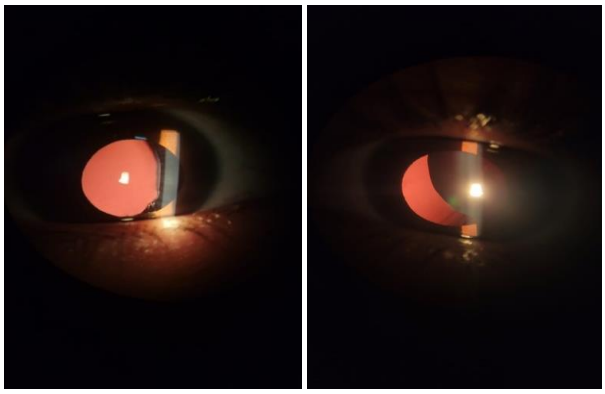

Figure 1: Ectopic lens in both eyes (right and left), with zonular rupture

Furthermore, general examination revealed short stature, muscular hypertrophy of the hands, short feet, and brachydactyly, with no evidence of psychomotor developmental delay. Cardiovascular examination revealed no cardiac abnormalities. A genetic workup was also performed.

Figure 2: Muscular hypertrophy of the hands associated with short stature.

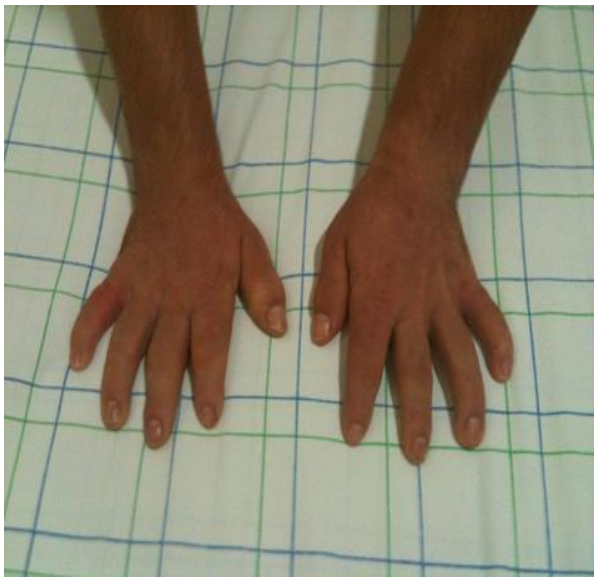

Figure 3: Brachydactyly

The diagnosis of Weill–Marchesani syndrome was established based on clinical criteria after excluding other causes of ectopia lentis.

Management consisted of bilateral lensectomy with implantation of iris-fixated intraocular lenses. The patient was also referred to a rehabilitation and physiotherapy facility to prevent joint stiffness.

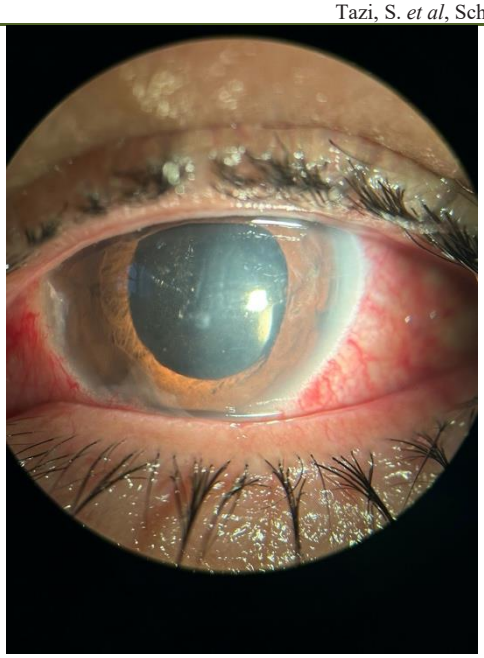

Figure 4: Postoperative Day 1 Following Surgery with Iris-Fixated Intraocular Lens Implantation

DISCUSSION

Weill–Marchesani syndrome is a congenital connective tissue disorder with an autosomal recessive inheritance pattern, most commonly associated with mutations in the *ADAMTS10, ADAMTS17*, or *LTBP2* genes, and occasionally with autosomal dominant inheritance due to heterozygous mutations in the FBN1 gene.[1] It is a rare syndrome, with prevalence varying across published studies.[2]. Diagnosis is primarily clinical and is characterized by short stature, brachydactyly, joint stiffness, and ocular abnormalities, particularly microspherophakia, ectopia lentis, and glaucoma.[3]. Ophthalmological management generally consists of removal of the ectopic lens. Physical therapy may be recommended to manage joint stiffness, along with early detection and appropriate management of associated cardiac abnormalities.[4]

CONCLUSION

Early diagnosis and appropriate management are essential to prevent and treat the ocular and, particularly, cardiac complications of Weill–Marchesani syndrome. Ocular manifestations may often be the presenting feature of the disease. Genetic counseling is recommended for individuals affected by Weill–Marchesani syndrome and their families. Other treatments for this disorder are mainly symptomatic.

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