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CASE REPORT

Timely diagnosis of Weill-Marchesani syndrome can preserve vision and prevent complication: A case report

Nafiz Ahmed Shahriar[1]* and Soma Rani Roy[2]

[1]Chittagong Eye Infirmary and Training Complex, Chattogram, Bangladesh
[2]Department of Ocular Oncology & Oculoplasty, Chittagong Eye Infirmary and Training Complex, Chattogram, Bangladesh

***Correspondence:**
Nafiz Ahmed Shahriar,
nafiz1107@gmail.com

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Purpose: To report a rare case of Weill-Marchesani syndrome (WMS) with its management.
Case report: A 40-year-old female patient presented with bilateral dimness of vision on both eyes, short stature, brachydactyly, and joint stiffness. Ocular examination revealed vision 3/60 in both eyes without improvement with a high myopic correction, bilateral microspherophakia with anterior lens subluxation, and normal fundus. She was diagnosed as a case of WMS and was managed by bilateral clear lens extraction followed by secondary anterior chamber intraocular lens implantation.
Conclusion: Timely diagnosis and meticulous surgical intervention can manage complications of WMS.

Keywords: Weill-Marchesani syndrome, microspherophakia, lens dislocation, brachydactyly, joint stiffness, clear lens extraction

Introduction

Weill-Marchesani (WMS) is a rare connective tissue disorder. It was first mentioned by Weill in 1932 (1) and further described by Marchesani in 1939 (2). It is also called congenital mesodermal dysmorpho-dystrophy or spherophakia-brachymorphia syndrome. The characteristic features of WMS include short stature, brachydactyly, microspherophakia, and ectopia lentis (3). The patients may also have joint stiffness and heart defects. As ocular symptoms and signs are characteristic and symptomatic, most patients present to ophthalmologists first. Characteristic eye abnormalities consist of dislocation of the microspherophakic lens, causing high myopia, acute and/or chronic angle-closure glaucoma, and cataracts. Autosomal recessive and autosomal dominant modes of inheritance have been reported (4, 5). Knowledge of mode of presentation of this syndrome facilitates its timely diagnosis. Here we report a case of WMS to describe its presenting features along with management and outcome.

Case report

A 40-year-old woman presented to us with the complaints of progressive visual disturbance on both eye. Her height was 4 ft 7 inches, which indicates her short stature. She had brachydactyly and joint stiffness. Her uncorrected visual acuity (UCVA) form Snellen eye chart was 3/60 in both eyes. Her retinoscopy showed −9 D myopia in both eyes through phakic portion. Her intraocular pressure (IOP) was 12 mmHg by Goldmann applanation tonometry in both eyes. Slit-lamp examination before pupillary dilation showed normal corneas in both eyes. The anterior chambers were relatively shallow in the middle portion but normal at the periphery. Gonioscopic examination revealed no abnormalities. There was bilateral microspherophakia with anterior subluxation of the crystalline lens, and the equator and zonules of the lens were visible within the pupil. The posterior segment was normal in both eyes. Based on these findings, the patient was diagnosed as WMS. She was advised for bilateral clear lens extraction followed by secondary anterior chamber intraocular lens implantation.

FIGURE 1 | (A) Short stature of the patient comparing height with an average height woman, **(B)** subluxation of the lens, and **(C)** brachydactyly of the patient.

The patient underwent surgery of her left eye within 1 week of diagnosis. After 2 months, she underwent surgery on her right eye. After her surgery on both eyes, her visual acuity was significantly improved to 6/24 with refractive correction. She was followed up on postoperative day (POD) 1, POD 7, and after 1 month of surgery in both times. She was given a final 3-month follow-up after the surgery of the second eye, showing her condition stable and refractive status unchanged (**Figure 1**).

Discussion

WMS is also known as microspherophakia-brachydactyly syndrome. The disease is rare and usually hereditary. It is a connective tissue disorder. Patients present with a lot of different features. Morphologically, short stature and brachydactyly are seen. Ocular findings include lens microspherophakia, lens subluxation, high myopia, and secondary glaucoma. There may be associated cardiac diseases also. Usually, patients carry a gene mutation, which may be autosomal dominant or autosomal recessive. A positive family history is very common. Faivre et al. reported that autosomal recessive and autosomal dominant inheritance found in 45 and 39% of cases, respectively. Besides, the remaining cases were sporadic (2). Patients with WMS are prone to secondary glaucoma. Recurrent glaucoma attacks cause angle adhesion, which may lead to trabecular meshwork damage and long-standing high IOP. This raised IOP eventually causes permanent damage to the optic nerve. Our patient presented with bilateral decreased visual acuity, microspherophakia, lens subluxation, high myopia, short stature, brachydactyly, and without a positive family history.

There is no well-established treatment modality for WMS. Removal of the microspherophakic lens helps control IOP and increase vision (6–8). Decision to undergo glaucoma surgery depends on the course of the disease. If the patient presents with early glaucoma and anterior chamber angle is found open, a simple removal of microspherophakia could be performed. In our case, the IOP and gonioscopic findings were normal, so we did clear lens extraction to prevent glaucoma.

If the patient presents with advanced glaucoma with angle closure, a combined glaucoma surgery with lens extraction should be considered, which may fail or the outcome may not be satisfactory. In our case, we did an early and timely intervention to prevent development of any secondary glaucoma or associated complication. Therefore, a timely diagnosis and treatment in patients with WMS is of vital importance to maintain and rescue their visual function.

Conclusion

Though WMS is a rare disease, clinicians have to be aware of the presenting features. If a patient has high myopia along with narrow anterior chamber but no myopic fundus change, it should be suspected as WMS. Removal of the microspherophakia is a good choice of intervention as it will help control IOP as well as preservation of vision and prevent glaucoma-related complications.

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