Timely Diagnosis of Weill-Marchesani Syndrome Can Preserve Vision and Prevent Complication: A Case Report

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Abstract.
Purpose: To report a rare case of Weill–Marchesani syndrome with its management.

Case Report: A 40-year-old female patient presented with bi-lateral 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 Weill-Marchesani syndrome and was managed by bi-lateral clear lens extraction followed by secondary anterior chamber intra-ocular lens implantation.

Conclusion: Timely diagnosis and by meticulous surgical intervention, complications of Weill-Marchesani syndrome can be managed accordingly.

Keywords: Weill–Marchesani syndrome, microspherophakia, lens dislocation, bradydactyly, joint stiffness, clear lens extraction.

INTRODUCTION

WMS is a connective tissue disorder. It is a rare condition. In 1932, It was first mentioned by Weill [1], and further described by Marchesani (1939) [2]. It is also called congenital mesodermal dysmorpho dystrophy or spherophakia-brachymorphia syndrome. Weill-Marchesani syndrome is distinguished by ectopia lentis, brachydactyly, microspherophakia, and short stature. Patients may also experience joint stiffness and cardiac problems. As ocular symptoms and signs are symptomatic and characteristic, most patients present to ophthalmologists first. Dislocation of the microspherophakic lens, which causes extreme myopia, acute and/or chronic angle-closure glaucoma & cataracts are all common eye disorders. There have been reports of autosomal recessive and autosomal dominant inheritance mechanisms [4, 5] Knowing how this condition manifests itself helps with timely diagnosis. We discuss the presenting characteristics of Weill-Marchesani syndrome, as well as management and outcome, in this case report.

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 Snellen visual acuity (UCVA) was 3/60 in both eyes. Her retinoscopy showed $-9$ D myopia in both eyes through phakic portion. By Goldmann applanation tonometry, her intraocular pressure (IOP) in both eyes was 12 mmHg. Before pupillary dilatation, a slit-lamp examination of both eyes revealed normal corneas in both eyes. In the centre, the anterior chambers were rather shallow, while the perimeter was typical. Gonioscopic examination revealed no abnormalities. The lens equator and zonules were evident within the pupil due to bilateral microspherophakia and anterior subluxation of the crystalline lens. In both eyes, the posterior portion was normal. Based on these findings, the patient was diagnosed with Weill-Marchesani syndrome. She was advised for bi-lateral clear
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The patient underwent surgery of her left eye within one week of diagnosis. After 2 month, she underwent surgery on her right eye. After her surgery on both eye, her visual acuity was significantly improved to 6/24 with refractive correction. She was followed up on 1st post-operative day, on 7th POD and after 1 month of surgery in both times. She was given a final follow up 3 month after the surgery of 2nd eye, where we found her condition stable and refractive status unchanged.

DISCUSSION

Microspherophakia-brachydactyly syndrome is another name for Weill-Marchesani 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 is seen. Ocular findings inculde 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. Autosomal dominant inheritance and autosomal recessive were detected in forty five percent and Thirty nine percent of cases, respectively, according to Faivre et al. Besides that the remaining cases were sporadic [2]. Patients with Weill-Marchesani syndrome are prone to secondary glaucoma. Recurrent glaucoma attacks causes angle adhesion which may lead to trabecular meshwork damage, it may cause 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 Weill-Marchesani Syndrome. The microspherophakic lens is removed to assist manage intraocular pressure and improve vision [6–8]. Decision to undergo glaucoma surgery depends on the course of the disease. If the patient has early glaucoma and the anterior chamber angle is discovered to be open, the microspherophakia can be removed easily. In our case, the IOP and gonioscopic findings were normal, so we did clear lens extraction to prevent glaucoma.

A combination glaucoma surgery with lens extraction might be considered if the patient has advanced glaucoma with angle closure, 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. As a result, it is critical to diagnose and treat people with WMS as soon as possible in order to preserve and restore their vision.

CONCLUSION

Though Weill-Marchesani syndrome is a rare disease, clinicians have to be aware of presenting features of it. 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 to control intraocular pressure, as well as preservation of vision and to prevent glaucoma related complications.

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REFERENCES


