

Photo essay

Weill Marchesani Syndrome: Microspherophakia

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Abstract

A rare case of Weill Marchesani syndrome in a 6 years old girl who presented with short stature, short broadhead, thickened skin with short stubby fingers and toes. On ocular examination, she had bilateral small spherical crystalline lens (microspherophakia) with stretched zonules causing anterior subluxation of the crystalline lens resulting in pupillary angle-closure glaucoma. Pentacam scheimpflug image measurement confirmed the presence of microspherophakia crystalline lens.

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A 6-year-old girl presented with complain of maintaining a close reading and writing distance, noticed by parents for 1 year. On general physical examination, she had a short stature, short broadhead (figure 1A), thickened skin with short stubby fingers and toes (figure 1B). Best-corrected visual acuity (BCVA) was 20/200 with myopic correction of -25.00 diopters spherical in both eyes. A-scan biometry using LENSTAR LS 900 (Haag Streit, USA) revealed that the axial length of his eyeball was 22.04 and 21.96 mm in the right eye (RE) and left eye (LE), however, the anterior chamber depth (ACD) was 1.06 and 1.12 mm for the RE and LE, suggesting of the shallow anterior chamber. Mean keratometry values were 45.3 D and 44.30 D for RE and LE, respectively (Pentacam, Oculus Inc. Arlington, WA). Additional informed consent was obtained from the patient for which identifying information is included in this article.

Ocular examination revealed bilateral small spherical crystalline lens (microspherophakia) with stretched zonules causing anterior subluxation of the crystalline lens resulting pupillary angle-closure glaucoma (figure 2). Pentacam scheimpflug image (figure 3) measurement confirmed



Figure 1: (A) Showing the patient's short stature and short broadhead (brachycephaly), (B) short broad fingers and toes (brachydactyly)

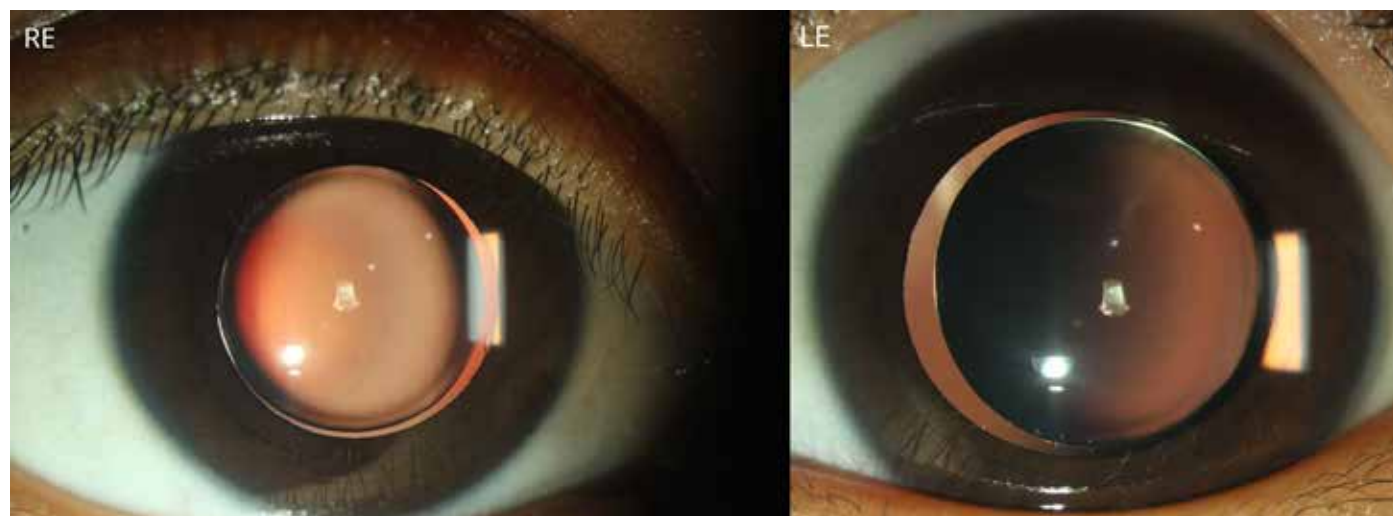


Figure 2: Slit-lamp bio-microscopy photo showing bilateral small spherical crystalline lens (microspherophakia) with stretched zonules causing anterior subluxation of the crystalline RE=right eye, LE=left eye

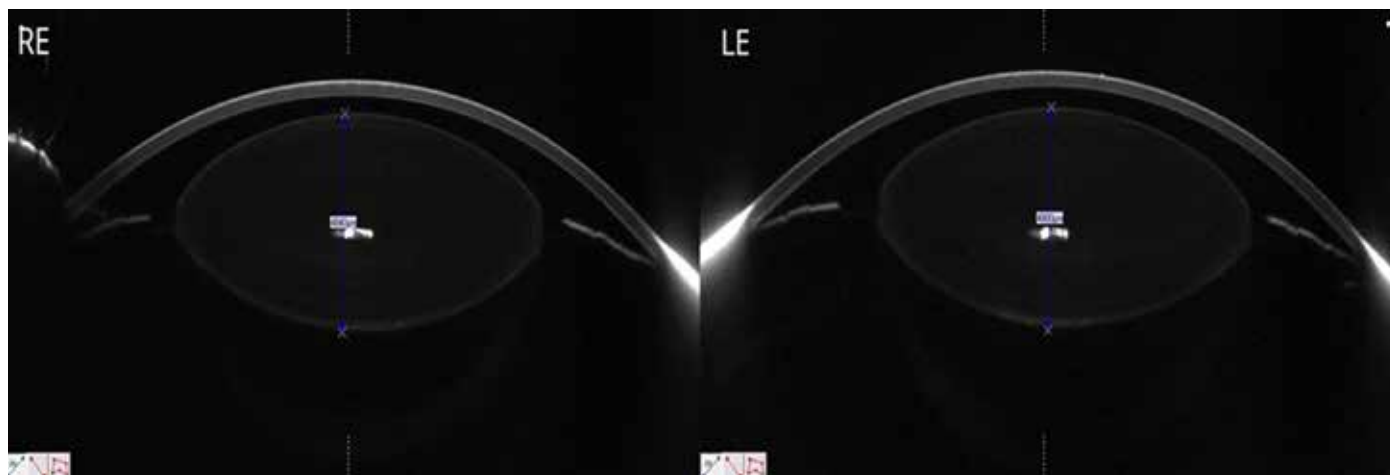


Figure 3: Pentacam (Scheimpflug image) showing microspherophakia; increased anteroposterior thickness (RE 4.83 mm, LE 4.88 mm), decreased equatorial diameter (RE 7.0 mm, LE 6.84 mm). RE=right eye, LE= left eye

the presence of microspherophakia crystalline lens. The anterior chamber were relatively shallow in the central portion compared to the periphery. The undilated Intraocular pressure (IOP) by Goldmann applanation tonometry was high, 38 and 29 mm Hg in RE and LE respectively which is reduced to 18 and 10 mm Hg in RE and LE post dilated. Dilated fundus examination revealed a glaucomatous disc in both eyes. Pars plana vitrectomy, pars plana lensectomy with glued intraocular lens were performed in both eyes. On a postoperative follow-up visit, the patient BCVA was 20/125 and 20/200 in RE and LE. The IOP in RE and LE were 16 and 18 mm Hg respectively with two anti-glaucoma medications, timolol 0.5% and brimonidine 0.2%.

Discussion

Both autosomal recessive and autosomal dominant modes of inheritance have been described in Weill-Marchesani disease.¹ It is a rare connective tissue disorder characterized by short height, brachydactyly, joint stiffness and lens abnormalities. Microspherophakia can be seen in Weill Marchesani syndrome (WMS).^{2,3} The characteristics of microspherophakia are: bilateral, small lens diameter, increased anteroposterior diameter making the lens relatively spherical, the eye is highly myopic and pupillary block glaucoma.⁴ Early surgical intervention should be carried out to restore the vision in these patients, when they present symptoms of decreased vision due to myopic shift.

References

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