Golden ring in the eyes: Weill-Marchesani syndrome

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DESCRIPTION

A 35-year-old woman presented with painful decreased vision in both eye over 3 months. On examination, she had a short stature of 4 feet 4 inches, short broad head (brachycephaly; [figure 1A](#)), thickened skin with short broad fingers,

brachydactyly ([figure 1B](#)) and joint stiffness. Best corrected visual acuity (BCVA) in the right eye was 20/200 and finger counting close to face (FCCF) in the left eye. Ocular examination revealed a small globular lens, microspherophakia with stretched zonules causing anterior subluxation ([figure 1C](#))

Figure 1 (A) Showing the patient's short stature and short broad head (brachycephaly), (B) short broad stubby fingers (brachydactyly) with thickened wrinkled skin, (C) golden ring due to reflection of light from 360° periphery of small crystalline globular lens (microspherophakia) with stretched zonules causing anterior subluxation in left eye.

Figure 2 (A) Right fundus showing advanced glaucomatous optic disc with vertical cup disc ratio of 0.8:1, thin neuroretinal rim and peripapillary atrophy. (B) Left fundus with near total cupping of optic disc with total loss of neuroretinal rim and peripapillary atrophy.

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resulting in angle closure glaucoma. Intraocular pressure (IOP) was high, with 34 and 38 mm Hg in right and left eye, respectively. The fundus in both eyes showed advanced glaucomatous optic atrophy (figure 2A and B). Intralenticular lens aspiration and left aphakia were performed. Visual rehabilitation was achieved with aphakic glasses and IOP was controlled with topical antiglaucoma medication. On follow-up, the patient's BCVA was 20/120 in the right eye and the left eye had FCCF

due to advanced glaucomatous optic atrophy. IOP in the right and left eyes were 16 and 14 mm Hg, respectively, with three topical medications, latanoprost 0.005% eye drops, timolol 0.5% and brimonidine 0.2%. The patient's elder sister had a similar history and presentation.

Autosomal recessive (AR) and autosomal dominant (AD) modes of inheritance have both been described in Weill-Marchesani disease.¹ It is a multisystem disease that affects the skin, bones and joints, the heart and the eyes.¹⁻³ Early lens aspiration should be carried out in these patients when they display symptoms of decreased vision due to myopic shift, so that angle closure glaucoma can be prevented.

Learning points

- ▶ Weill-Marchesani syndrome is a multisystem disease that affects the skin, bones and joints, the heart and the eyes. So cardiac evaluation should be performed before surgery.
- ▶ Early lens aspiration should be carried out in these patients when they display symptoms of decreased vision due to myopic shift as a consequence of the globular shape of the lens, otherwise pupillary block and angle closure glaucoma develop, leading to irreversible vision loss.
- ▶ Trabeculectomy may be required to control Intraocular pressure in these patients.

Competing interests None declared.

Patient consent Obtained.

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