Retinitis pigmentosa associated with ectopia lentis in a patient with Marfan syndrome

Dear Editor,

A 32-year-old, 6.2-foot man presented with complaints of defective vision since early childhood and a history of defective night vision. His father died of an unknown cause when he was 5 years old. The patient had visible arachnodactyly and hyperextensible joints [Fig. 1], but this did not affect his life. Ocular examination revealed that both lenses were dislocated superiorly. Punctate anterior and posterior “snowflake” lens opacities resembled a complicated cataract [Fig. 2]. Best-corrected visual acuities (BCVA) were 20/400 (-2.25), right, and 20/250 (-4.00), left, respectively. The anterior chamber was normal for both eyes. Intraocular pressures were 20 and 18 mmHg, respectively. We could not see the fundus clearly before the cataract surgery. Electroretinography (ERG) findings showed a barely detectable rod b-wave [Fig. 3]. The amplitudes of a- and b-waves in the mixed cone–rod signal were reduced, and the cone response was delayed, suggesting a diagnosis of retinitis pigmentosa (RP). Marfan syndrome was confirmed through his tall height, arachnodactyly,
hyperextensible joints and echocardiography [Fig. 4]. Phacoemulsification and anterior vitrectomy were performed on both eyes. Intraocular lens (IOL) was not implanted the first time. After the surgery, fundus photograph showed “bone-spicule formation” in both eyes [Fig. 5]. BCVA were 20/50 (+10.00), right, and 20/25 (+12.50), left, respectively. Scleral fixated IOL were implanted 3 months later in order to improve the patient’s quality of life. BCVA was 20/60 right and 20/50 left after the surgery.

About 50% of the patients with Marfan syndrome are diagnosed by ophthalmologists. The presence of ectopia lentis (EL) is considered a major criterion for the diagnosis of Marfan syndrome in nosology, which unequivocally diagnoses or excludes Marfan syndrome in 86% of the cases.[1]

Although the link between RP and EL has been reported before,[2-4] systemic symptoms have not been reported. The only complaint of the current case was defective vision, and EL was found during the eye examination. Considering his 6.2-foot height (taller than average Chinese man), arachnodactyly, hyperextensible joints and echocardiography, a systemic examination was performed and Marfan syndrome was then confirmed. These results suggest that a systemic examination may be necessary in tall subjects with poor vision.
To the best of our knowledge, this is the first report of bilateral spontaneous EL associated with RP in a patient with Marfan syndrome.

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