

Weill-Marchesani syndrome associated with retinitis pigmentosa

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Retinitis pigmentosa (RP) is associated with a wide variety of ocular and systemic disorders. The Weill-Marchesani syndrome is a multi-system disorder with microspherophakia as one of the common manifestations. A 14-year-old girl presented with short stature, short and stubby fingers, hypodontia and low-set ears. Slit-lamp examination revealed microspherophakia, with shallow anterior chambers with irido and phacodonesis. Ultrasonographic biomicroscopy confirmed the clinical findings and revealed hypoplastic ciliary body. Electroretinogram confirmed the diagnosis of RP. Though RP has been associated with ectopia lentis in earlier reports, this is, to the best of our knowledge, the first case report describing the association of RP and Weill-Marchesani syndrome.

Key words: Ectopia lentis, retinitis pigmentosa, Weill-Marchesani.

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Retinitis pigmentosa (RP) is a condition characterized by degeneration of the retinal pigment epithelial (RPE) cells. It has a wide variety of association with ocular and systemic syndromes. The Weill-Marchesani syndrome is a systemic syndrome characterized by short stature, short and stubby fingers, microspherophakia of the lens leading to lenticular myopia and pupillary block glaucoma due to anterior subluxation. Though association of ectopia lentis with RP has been reported, association of the Weill-Marchesani syndrome with RP has never been reported.¹⁻³ We report a case of RP with bilateral microspherophakia and other features of the Weill-Marchesani syndrome.

Case Report

A 14-year-old female presented to us with complaints of defective vision since early childhood. She had history of defective night vision. She was the third child of a second-degree consanguineous marriage, full-term normal delivery with normal developmental milestones. There was no suggestive family history. On systemic examination, she had normal development and mental makeup without any neurological deficit. She was short (height 115 cm) with short and stubby fingers in both the limbs [Fig. 1a]. She also had contracture of hands, low-set ears and hypodontia. Ocular examination revealed hypertelorism, shallow anterior chamber, microspherophakia with a few lens opacities,

iridodonesis and phacodonesis [Fig. 1b]. Refraction was grossly myopic -17.0 diopter sphere (DS) / -2.0 diopter cylinder (DC) at 180° in right eye (RE) and -20.0 DS / -2.0 DC at 180° in left eye (LE). The best corrected visual acuity was 20/80 in RE and 17/200 in LE. Intraocular pressure was 22 mm of Hg in RE and 20 mm of Hg in LE. Axial length was



Figure 1a: Clinical photograph showing short stubby fingers in upper limb



Figure 1b: Slit-lamp photograph showing microspherophakia

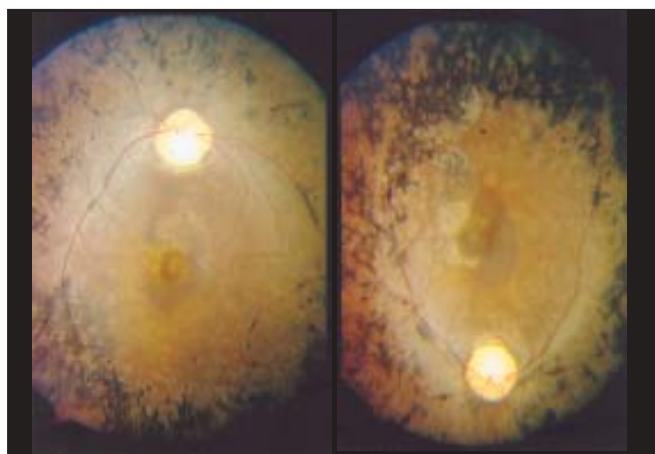


Figure 2a: Fundus photographs of both the eyes showing disc pallor and pigmentation suggestive of retinitis pigmentosa

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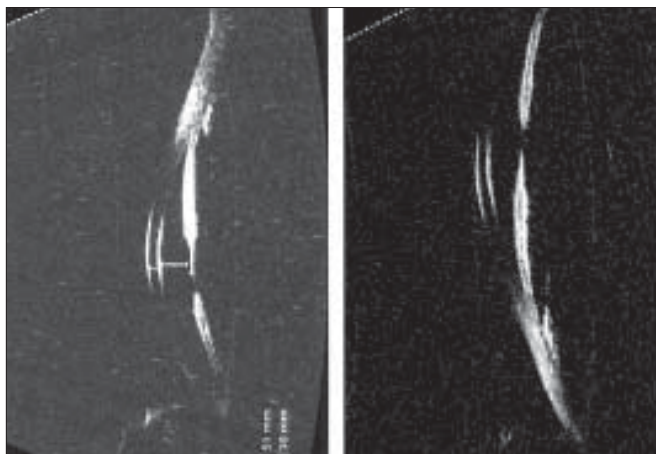


Figure 2b: UBM of right eye showing spherophakia, shallow anterior chamber with hypoplastic ciliary body

21.24 mm in RE and 21.57 mm in LE. Mean keratometric values were 44.5 D and 45.5 D for RE and LE, respectively. Pachymetry showed central corneal thickness to be 0.51 mm in RE and 0.52 mm in LE (HF35-50 High Frequency Ultrasound, OTI, Toronto, Canada). In the RE, the lens thickness was measured to be 4.98 mm and in the LE, it was 4.32 mm (A-scan, OTI, Toronto, Canada). Fundus examination showed the presence of multiple bony spicules characteristic of RP with disc pallor in both eyes [Fig. 2a].

Electroretinogram (ERG) revealed flat waveforms for both scotopic and photopic conditions which further confirmed the diagnosis of RP.

On ultrasonic biomicroscopy (UBM) with a 50Hz probe (HF35-50 High Frequency Ultrasound, OTI, Toronto, Canada) we found decreased anterior chamber depth, narrow angle of the anterior chamber, increased lenticular sphericity, elongated zonules and ciliary body hypoplasia [Fig. 2b]. Prophylactic YAG laser iridotomies were done in both eyes. The findings are similar to previously reported findings of UBM on the Weill-Marchesani syndrome (disagreeing with Marchesani's view of ciliary body hyperplasia).^{4,5}

Laboratory investigations revealed anemia, absence of homocystine in the urine (negative sodium nitroprusside test), absence of sulfur-containing amino acids and lysine in the urine, normal level of phytanic acid in the serum. All family members of the child underwent complete ophthalmological examination and were found to be normal

Discussion

An association of RP with lens dislocation has been reported rarely.² Available literature suggests sparse association of dislocated lens with RP, but the dislocations reported were spontaneous as all systemic syndromes were ruled out.^{2,3} Derigs,

in 1882, reported two cases of congenital ectopia lentis in a series of 60 RP patients and Herlinger in 1899, noted one out of 92 patients of RP to have a subluxated lens.² Pifarreti studied the correlations between ocular malformations in 51,000 patients seen at the University of Lausanne² and found one patient with lens dislocation out of 42 patients of RP. The coexistence of Marfan-like syndrome and RP has been reported earlier.¹

Our patient was diagnosed to be having the Weill-Marchesani syndrome (presence of microspherophakia with axial diameter of 4.98 and 4.32 mm (mean saggital diameter of spherophakic lenses, 4.5 to 4.9 mm)), shallow anterior chamber, hypoplastic ciliary body (on UBM), raised intraocular pressure and systemic features such as short stature and short and stubby fingers.^{4,6} Other causes of lens subluxation were ruled out owing to a normal set of laboratory investigations, normal skeletal features and absence of mental retardation. RP was diagnosed from the fundus picture as well as the flat ERG waveforms.

To the best of our knowledge, such an association has never been reported earlier in literature (Medline search on pubmed). Ectopia lentis, though reported in literature to be associated with RP in nearly six cases, was never secondary to the Weill-Marchesani syndrome as in our case.¹⁻³ Although there was no family history, there was a history of consanguinity which suggests that it could have been a recessive disorder with variability in penetrance. However, in the absence of chromosomal and genetic analysis,³ we would like to suggest that this may be a new clinical entity for which sub-clinical and genotypic classifications will be necessary.

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