

Treacher Collins syndrome with microcornea and retinal detachment

Abijith Holla,¹ Sarita R J Gonsalves,² Geover Joslen Lobo³

¹Department of Ophthalmology, Sagarmatha Choudry Eye Hospital, Lahan, Nepal

²Department of Ophthalmology, Kasturba Medical College, Manipal, Karnataka, India

³Department of Neurosurgery, Kasturba Medical College, Udupi, Karnataka, India

Correspondence to

Dr Sarita RJ Gonsalves, gonsalvessarita@gmail.com

DESCRIPTION

A 15-year-old girl was referred to our ophthalmology unit for poor vision in the left eye. She was a diagnosed case of Treacher Collin syndrome (figure 1) and was surgically treated for microtia of the left ear (figure 2). On referral she reported of progressive loss of vision in the left eye for 8 years, no history of trauma, redness, pain or ophthalmic surgical intervention. On examination of the right eye her visual acuity was 6/6. There was antimongoloid slant, lower lid coloboma with deficient cilia medial to coloboma (figure 3). Conjunctiva showed Bitot's spots (figure 4), the rest of the anterior segment and fundus was normal. The intraocular pressure was 16 mm Hg. In the left eye, the patient had only perception of light. Projection of rays was inaccurate in all quadrants. Antimongoloid slant, lower lid coloboma with deficient cilia medial to coloboma was present. Anterior segment evaluation: conjunctiva showed Bitot's spots, horizontal corneal diameter of 9 mm, non-reacting pupil, cataractous lens. Examination of the fundus was not



Figure 3 Showing downsloping palpebral fissure and coloboma of both lids with microcornea.

possible due to media opacity. Intraocular pressure was 8 mm Hg.

B-SCAN of the left eye showed retinal detachment (RD) with subretinal cysts (figures 5). Axial length of the left eye was 20.5 mm as calculated by vector A-SCAN. The patient was advised surgical intervention for RD under guarded visual prognosis, she was not willing for the same hence it was deferred. The common features of Treacher Collin syndrome are craniofacial mal-development, antimongoloid slant of the eyes, micrognathia,



Figure 1 Treacher Collin syndrome in an adult showing facial dysmorphism and lid abnormalities.

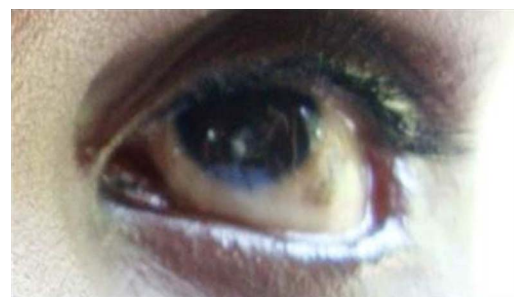


Figure 4 Anterior segment photograph showing microcornea and Bitot's spots.



Figure 2 Surgically corrected malformed pinna.



Figure 5 B-SCAN showing retinal detachment with cysts in the left eye.



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microtia.^{1 2} The association with microcornea and RD is rare and hence the case report.

Learning points

- ▶ Craniofacial anomalies and fusion defects are a part of the syndromic associations of Treacher Collins syndrome. However only few cases with microcornea and retinal detachment have been reported so far.
- ▶ Management of Treacher Collin syndrome requires a multidisciplinary approach.

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Competing interests None.

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