Bilateral retinal detachment in Goldenhar syndrome

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The ocular features of Goldenhar syndrome (GS) are typically external, like surface dermoids and lid coloboma. Retinal detachment (RD) is rare and has not been reported in absence of other concomitant predisposing congenital ocular disorders. We present a unique case of bilateral rhegmatogenous retinal detachment (RRD) with GS. To the best of our knowledge, this association of GS with RRD is novel and has not been reported earlier in ophthalmic and systemic literature on RRD.

Key words: Goldenhar syndrome, retinal detachment, vitreoretinopathy

Case Report

A 25-year-old male presented with painless loss of vision in right eye (RE) since 2 months. The left eye (LE) had low vision since 3 years. There was no family history of ocular disorders. Visual acuity was perception of light in both eyes (BE), while

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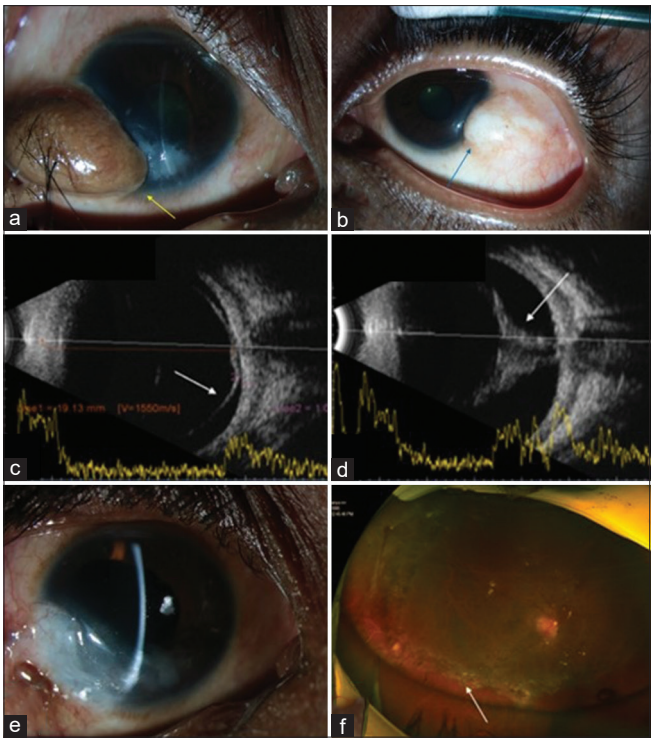

Figure 1: (a and b) Clinical photographs of the (a) right and (b) left eyes showing epi-bulbar dermoids at the inferior-temporal limbus (arrows). (c and d) Ultrasound B-scans of the (a) right and (b) left eyes showing retinal detachment (arrows). The detachment in the left eye is funnel shaped posteriorly. (e and f) Slit lamp photograph at 1 month of the right cornea showing healing and vascularization at the site of dermoid excision. Fundus photograph shows attached retina with laser marks visible at the edge of the retinectomy site (arrow)

projection of rays was inaccurate in LE. He had gross malar and mandibular hypoplasia with multiple preauricular skin tags on the left face, along with a dysmorphic right external ear. BE visual axis was involved by limbal dermoids and complicated cataract with low IOP. Ultrasound showed retinal detachment (RD) in BE, with LE having a funnel configuration [Fig. 1]. No choroidal coloboma or staphyloma was detected. Axial length was measured as 19.13 mm in RE, and 17.47 mm in LE. Further systemic work up revealed kypho-scoliosis without any visceral defects. Hence the patient was diagnosed to have Goldenhar syndrome (GS) with BE RD.

The patient underwent RE dermoid excision, pars plana vitrectomy, pars plana lensectomy and silicone oil injection. During surgery, an old total rhegmatogenous RD (RRD) with severe inferior retinal shortening was noted along with lattice degeneration and retinal hole [Fig. 1]. The vitreous cavity appeared grossly empty, even after staining with triamcinolone. Three months after surgery, the visual acuity was 20/400-N36 with attached retina and normal IOP [Fig. 1].

Discussion

External eye is involved in up to 60% of cases of GS,[1-3] but retinal involvement is rare. The pathogenesis of GS involves anomalous formation of the embryonal branchial arches which don't directly contribute to retinal development.[3-5] A case each of vitelliform maculopathy and familial exudative vitreo-retinopathy has been described with GS before, but RRD occurs only in simultaneous presence of choroidal coloboma or morning glory syndrome.[1,2,4] Further literature is required for establishing a true association, if any, between GS and RRD.

Conclusion

To the best of our knowledge, the association of GS with RRD is novel and has not been reported earlier.

Ethical approval

All procedures performed were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments.

Acknowledgment

Hyderabad Eye Research Foundation and Hyderabad Eye Institute, Hyderabad, India.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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