Photo Essay

Stickler syndrome

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Key words: Cleft palate, radial lattices, retinal detachment, Stickler syndrome

Stickler syndrome is a well-described but uncommon clinical entity. It is a hereditary connective tissue disorder of fibrillar collagen with autosomal dominant inheritance. Phenotype manifests as four different features that is, ocular, orofacial, auditory, and musculoskeletal.

Case Description

A 17-year-old male reported with gradual, progressive, painless diminution of vision in both eyes since 3 months. Family history was unremarkable. The best corrected visual acuity was the perception of light+ in right eye and 20/400 in the left eye. The anterior segment showed dense cataract in the right and posterior chamber intraocular lens in the left eye [Fig. 1]. Intraocular pressure was 4 mmHg in the right and 16 mmHg in the left eye. Left eye fundus revealed multiple radial perivascular lattices, vitreous condensation, tessellations, and posterior staphyloma [Fig. 2], and an axial length of 26.3 mm. The left eye was amblyopic. Ultrasound B-scan of the right eye revealed closed funnel retinal detachment (RD) and reduced axial length (21.2 mm) [Fig. 3]. Electroretinogram showed normal responses in the left and expected nonrecordable responses in the right eye [Fig. 4]. Systemic examination revealed speech abnormality (nasal twang), large cleft palate [Fig. 5], and mild hearing defect.

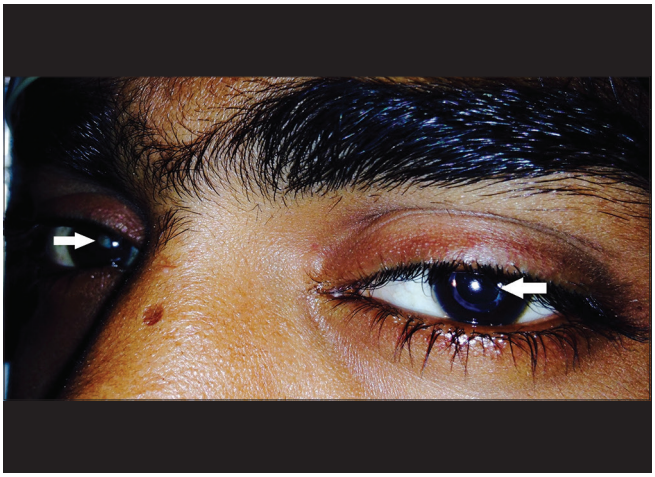

Figure 1: External (slit-lamp) photo reveals cataract in the right eye (right arrow) and posterior chamber intraocular lens implant in the left eye (left arrow)

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	DOI: 10.4103/0301-4738.167114

Discussion

Stickler syndrome was first reported in 1965 by Stickler *et al*.[1] as hereditary arthro-ophthalmopathy. It is now divided into subgroups depending on the clinical

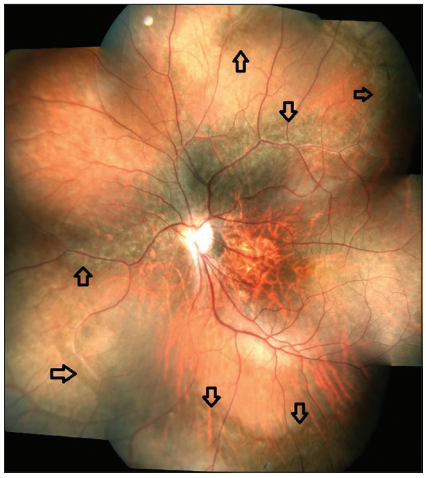

Figure 2: Left eye color fundus montage showing multiple radial, perivascular lattices (arrows), tessellated fundus, and posterior staphyloma

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Manuscript received: 11.01.15; Revision accepted: 15.05.15

Cite this article as: Rishi P, Maheshwari A, Rishi E. Stickler syndrome. Indian J Ophthalmol 2015;63:614-5.

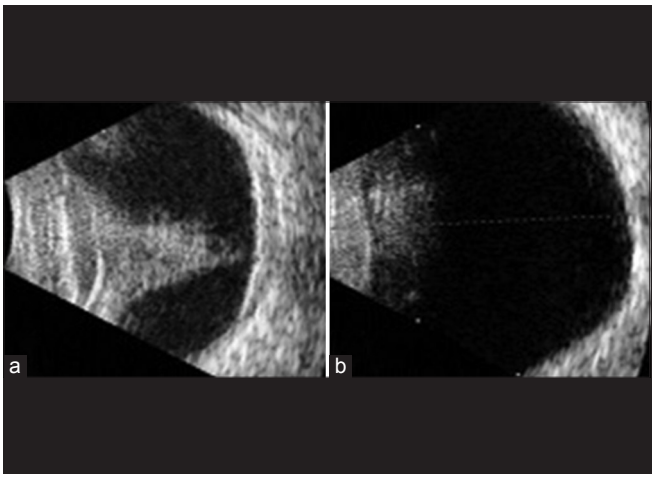

Figure 3: Ultrasonography B-scan showing closed funnel retinal detachment with axial length of 21.2 mm (a) in right eye and axial length of 26.3 mm in left eye (b)

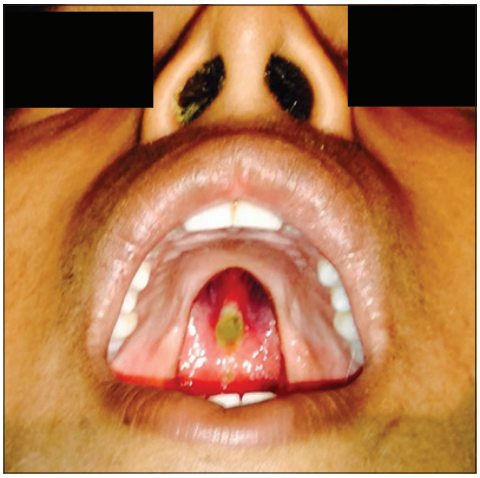

Figure 5: Oral examination reveals large cleft palate

manifestations. Vitreous assessment is the diagnostic criteria[2] and guides to molecular genetic analysis.[3] Complications such as RD (70%), cataract (49%), and ocular hypertension (10%)[4] are progressive and can lead to blindness. A multidisciplinary approach is required. Ocular rehabilitation includes spectacles/contact lenses, frequent retinal examinations, cataract surgery, and prophylactic retinal laser photocoagulation. Meticulous systemic examination and genetic counseling help in identifying the disorder in newborns and preventing complications. Mutations in the *COL2A1*, *COL11A1*, *COL11A2*, *COL9A1*, and *COL9A2* genes can cause Stickler syndrome, Types I to V. Our patient had characteristic features of Type I Stickler

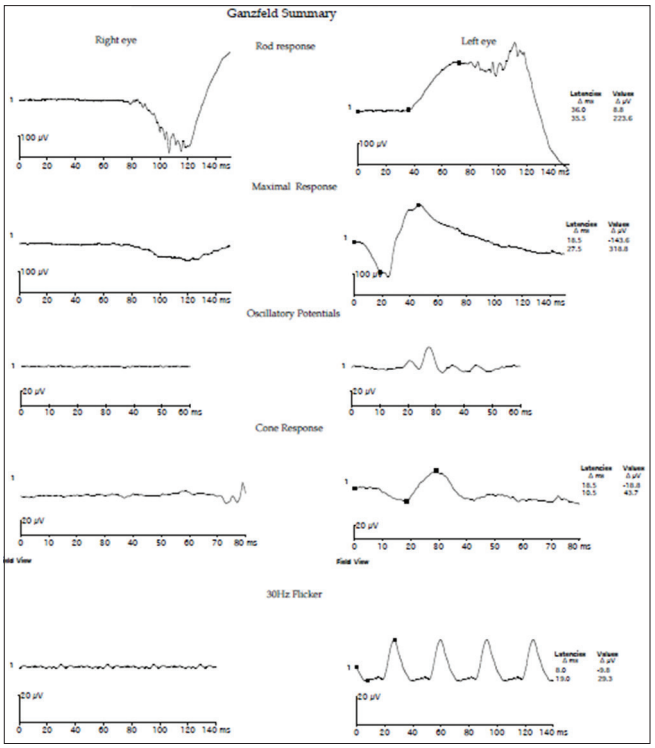

Figure 4: Electroretinogram showing nonrecordable photopic and scotopic responses in oculus dexter and normal responses in left eye

syndrome, such as cleft palate and hearing deficit, except a positive family history. Genetic testing was offered but declined by the patient. Differentials include multiple epiphyseal dysgenesis, Kniest dysplasia (musculoskeletal involvement), Knobloch (encephalocele), and Wagner syndrome (ocular involvement only).

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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