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Case Report

STICKLER SYNDROME WITH BILATERAL CORNEAL OPACITIES–A RARE ENTITY

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ABSTRACT
Ophthalmologic anomalies are the most characteristic and most serious manifestations of the syndrome. In the eye, abnormalities of vitreous gel architecture associated with high myopia and retinal detachment have been described. There is a substantial risk of retinal detachment. Here in we report a rare association of stickler syndrome with bilateral corneal opacities as the only ocular abnormality, which has not been described earlier.

Key Words: *Stickler Syndrome and Corneal Opacities*

INTRODUCTION
Stickler syndrome or hereditary progressive arthroophthalmopathy is an autosomal dominant condition characterized by ocular manifestations, arthritic changes, orofacial features and deafness (Stickler, 1965; Herrmann, 1975). Ophthalmologic anomalies are the most characteristic and most serious manifestations of the syndrome. High myopia and abnormalities of vitreous gel architecture are characteristically seen. There is a high risk of retinal detachment that can lead to blindness. Here in we report a case of stickler syndrome with bilateral corneal opacities as the only ocular abnormality. This to our knowledge has not been described earlier.

CASES
A, baby boy was a spontaneous vaginal delivery at full term to a 23 years old primigravida out of a non consanguineous marriage. Baby cried immediately after birth but developed respiratory distress, which settled in six hours. The birth weight was 2.3 kg, head circumference 32.5 cm and there was cleft lip on left side, cleft palate, retrognathia and bilateral corneal opacities. No other gross congenital malformations were present.Whole body skeletal survey, USG skull and eyes and echocardiography was normal. USG abdomen revealed absent left kidney. BERA revealed bilateral sensorineural deafness. Molecular genetic testing for the affected gene could not be done. Baby's father was also operated for cleft lip and cleft palate at the age of 2 years. A clinical diagnosis of Stickler syndrome was made. The course of the child during hospital stay remained uneventful and baby was discharged on 10^{th} day of life on self feeding.

DISCUSSION
Stickler syndrome is an autosomal dominant disorder with characteristic ophthalmological and orofacial features, deafness, and arthritis. In the eye, abnormalities of vitreous gel architecture associated with high myopia and retinal detachment have been described (Baraitser, 1982). There is a substantial risk of retinal detachment. Bilateral corneal opacities present in the index case have not been described earlier. Mild spondyloepiphyseal dysplasia is often apparent radiologically. Sensorineural deafness with high tone loss may be asymptomatic or mild. Occasional findings include slender extremities and long fingers. Stature and intellect are usually normal (Zlotogora, 1992). The diagnosis of Stickler syndrome is clinically based. At present, no consensus minimal clinical diagnostic criteria exist (Snead, 1999). The diagnostic criteria for Type I Stickler Syndrome include orofacial abnormalities where 2 points are for major criteria like cleft palate (open cleft, submucous cleft, or bifid uvula) and 1 point for minor criteria

like characteristic face (malar hypoplasia, broad or flat nasal bridge, and micro/retrognathia) (Peter, 2005). Similarly ocular abnormalities like vitreous changes or retinal abnormalities (lattice degeneration, retinal hole, retinal detachment or retinal tear) and auditory abnormalities like high frequency sensorineural hearing loss (Zlotogora, 1992) are considered major criteria for 2 points and Hypermobile tympanic membranes is considered as minor criteria for 1 point. Other points include skeletal abnormalities like femoral head failure (slipped epiphysis or Legg–Perthes-like disease), radiographically demonstrated osteoarthritis before age 40, Scoliosis, spondylolisthesis, or Scheuermann-like kyphotic deformity which are all awarded I point each. There is also 1 point given to independently affect 1st degree relative in a pattern consistent with autosomal dominant inheritance or presence of COL2A1, COL11A1, or COL11A2 mutation associated with Stickler syndrome .The diagnosis requires 5 or more points with at least one major 2-point manifestation and absence of features suggestive of more severe skeletal dysplasia or other syndrome. In index case, we have got orofacial abnormalities like cleft lip and palate, ocular abnormality like corneal abnormality which is not reported in literature, bilateral sensorineural deafness (Szymko-Bennett, 2001) and father of the child having cleft palate and cleft lip at birth. We couldn't convince parents for molecular analysis as it involved considerable costs which parents couldn't afford. Recognition of Stickler syndrome has important medical and personal consequences for patients and their families. Early identification of ocular and auditory abnormalities allows surveillance for and early treatment of complications. Similarly, correct diagnosis allows prognosis of and surveillance for skeletal complications and genetic counseling for affected families.

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