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Case Report

Goldenhar Syndrome: Case Report with Review of Literature

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ABSTRACT

Goldenhar syndrome is a rare congenital anomaly which consists of a triad of an ocular dermoid cyst, preauricular skin tags and vertebral dysplasia. We report a case of Goldenhar syndrome, diagnosed in a 9 month old baby boy.

Key words: Goldenhar syndrome, Pediatrics.

INTRODUCTION

Goldenhar syndrome is a rare congenital anomaly consisting of a triad of an ocular dermoidcyst, preauricular skin tags and vertebral dysplasia. [1,2]

Early diagnosis and treatment of this tumor prevents the development of secondary amblyopia. We present a case of a 9 month male infant with classical features of this rare condition i.e. Pre Auricular Tags, Limbal Dermoid, Tracheo Oesophageal Fistula with Oesophageal Atresia, Mild Facial Asymmetry and Left Renal Agenesis.

CASE REPORT

A 9 Month Old Male infant, born of a non consanguineous marriage presented to our department being referred from ophthalmology for evaluation of the congenital lesions of the eye and face. There was neither history of any maternal illness during pregnancy nor any similar history in family. Baby was operated on 3rd day of life for Tracheo Oesophegeal Fistula With Oesophageal Atresia. The Infant on ocular examination had limbaldermoid, located on the inferotemporal side of limbus of the right eye (Figure 1). Bulbar conjunctival dermoid tumor invading the inferotemporal side of the limbus. There was neither evidence of any coloboma nor microphthalmia, and ocular motility was normal.

On facial examination, there were two right sided preauricular tags along the line joining the tragus with the angle of the mouth (Figure 2). *Fig. 2:* skin coloured papules in the preauricular area and mild right sided facial asymmetry.

Post op scar was noted on right thorax. Ultrasound abdomen revealed normal solid organs except for left renal agenesis. Infantogram revealed normal study. CT temporal bone was near normal.

The laboratory investigations were within normal limit. ECG - normal, X-ray chest (postero anterior view) showed vertebral dysplasia. Complete excision of

the dermoid cyst was achieved and graft was placed and general anesthesia. The postoperative course was uneventful.

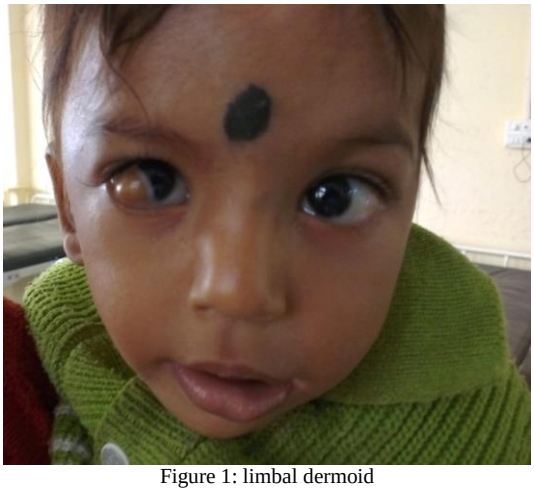

Figure 1: limbal dermoid

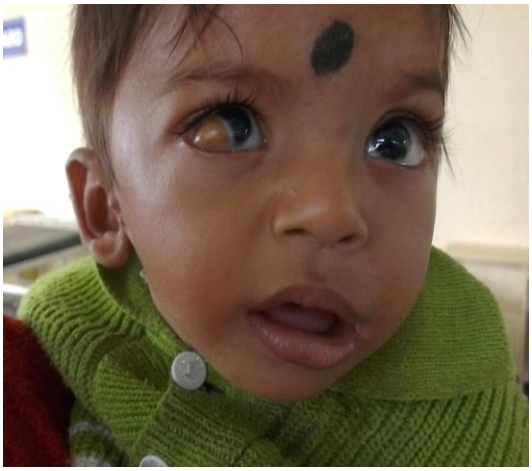

Figure 2: mild right sided facial asymmetry.

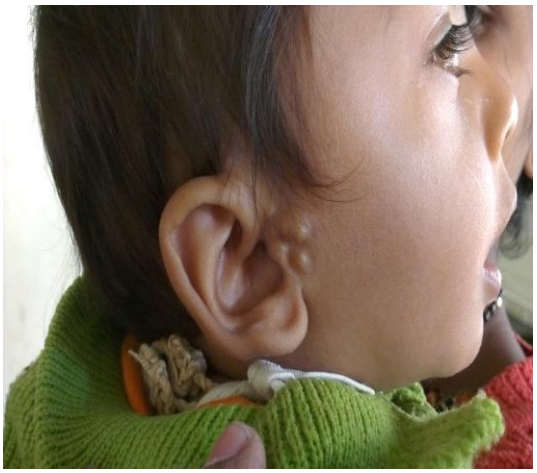

Figure 3: preauricular tags.

DISCUSSION

Goldenhar syndrome, also known as the Oculo-Auriculo-Vertebral syndrome (OAVS), is a rare congenital malformation involving the first and second branchial arches. [3] It was first described by Goldenhar in 1952.

It is one of the variants of craniofacial anomalies. It is unilateral in 70-80% of the cases. [4]

The syndrome complex includes limbaldermoid or lipodermoid, pre-auricular tags, hemifacial asymmetry and vertebral anomalies.

Age of onset being during neonatal & infancy. Prevalence rate is in 1-9/100000, [5,6] incidence rate is 1 in 25000-45000 births. [7]

Males are more commonly affected than the females (ratio 2:1). Most of the cases of OAV are sporadic, autosomal dominant transmission is reported for 1% - 2% of the cases. [7]

Aetiology of the syndrome remains unclear. There is no evidence of a clear inheritance pattern and no chromosomal anomalies have been described up to now. [8] Currently a deficiency in mesodermal formation or defective interaction between neural crest or mesoderm is suggested as possible aetiology. [7]

Ocular anomalies occur about 50% of the case of OAV. [9] Auricular defects are reported in 65% of the cases and include pre-auricular tags, microtia, anotia& conductive hearing loss. Vertebral anomalies are combination of hemivertebra, fused ribs, kyphosis and scoliosis. Additional features this syndrome can be associated with cardiac, pulmonary, renal and skeletal malformations in 50% of cases. An association of intracranial dermoid cyst and Goldenhar syndrome has been reported. [10] Cardiopulmonary distress within the few months of life is relatively common life threatening complication.

The Following Differential Diagnosis should be considered for facial anomalies

- Treacher Collins Syndrome
- Romberg Disease
- Craniosynostosis
- Hemifacial Microsomia

Differential Diagnosis For Limbal Dermoid

- Juvenile Xanthogranuloma
- Corneal Scar
- Sclerocornea
- Atypical Pterygium,
- Foreign Body Granuloma

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