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GOLDENHAR SYNDROME

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Abstract: Goldenhar also known as Oculoauriculovertebral dysplasia is a triad of epibulbar dermoids, accessory auricular appendages and vertebral anomalies. The following case is reported for its rarity and bilateral presentation of preauricular appendages, dermoid in one eye and dermolipoma in the other.

Keywords: Goldenhar, dermoid, dermolipoma, coloboma, congenital anomalies

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INTRODUCTION

A 10 month old male child presented to our out patient department with an opacity in the left eye and a growth in the outer angle of the right eye which were present since birth. Antenatal and birth history was uneventful. Developmental milestones were normal till date.

Ocular findings: A yellowish white swelling in the right outer canthus extending from superior to inferior fornix. The posterior border was not visualized and the swelling was extending out of sight between the lid and the globe. Diagnosis of dermolipoma was suspected.

Fig1: Dermolipoma

There was a coloboma of the upper eyelid in the left eye and a limbal dermoid was seen in the inferior part. On examination, both the fundi were normal.

Auricular findings: Bilateral preauricular accessory appendages were present. Pinnae and external auditory canals were normal and there was no associated clinical deafness.

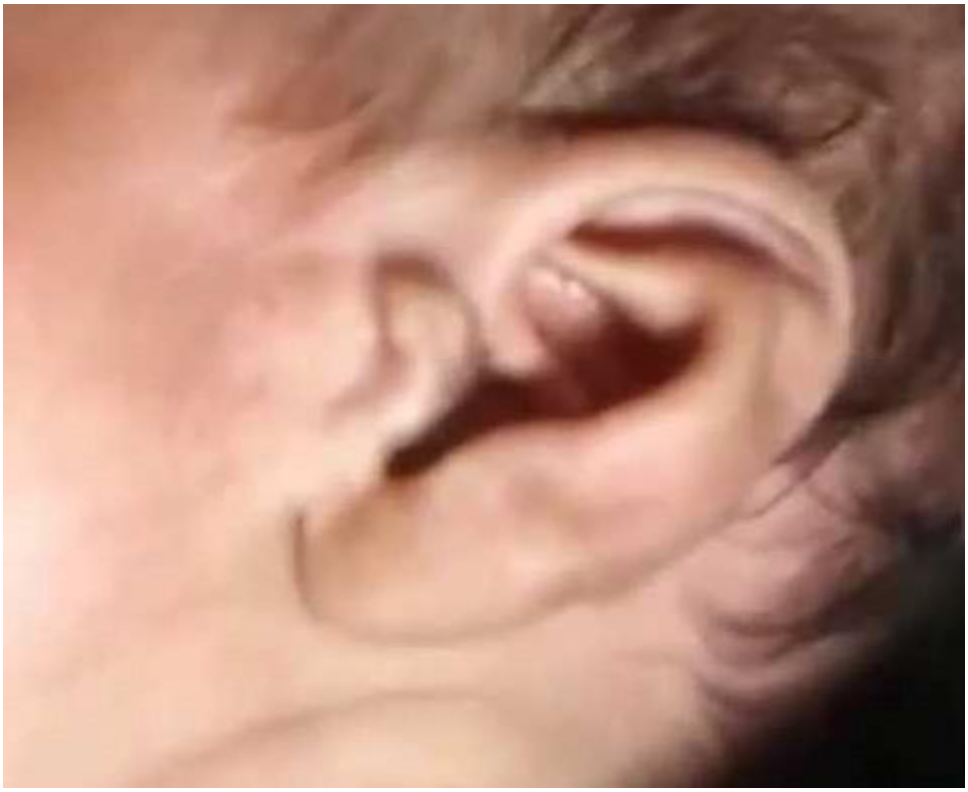

Fig2: Pre auricular skin appendages.

Oral findings: The child had macrostomia and high arched palate.

Skeletal findings: There was no clinical evidence of any skeletal anomalies.

The case reported as both features of Goldenhar and Franceschetti syndromes. Goldenhar syndrome is a congenital anomaly involving the first and second brachial arches and the differentiating features are unilateral ear deformity, coloboma involving upper lid, presence of epibulbar dermoid and vertebral anomalies. In Franceschetti syndrome, the features are bilateral ear deformity, coloboma involving lower lid. Bilateral flame shaped projection of hair extending in a line from ear to angle of mouth and lying over the temporal bone is characteristic of this syndrome.

From genetic point of view, Franceschetti syndrome appears to be irregularly dominant whereas there is no known hereditary pattern for Goldenhar syndrome. In our case there was no family history of consanguinity and also chromosomal studies showed normal karyotype and count favouring Goldenhar syndrome.

Features of Goldenhar in our case included upperlid coloboma, limbal dermoid, dermolipoma and accessory auricular appendages and those of Franceschetti syndrome were macrostomia and high arched palate.

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