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Goldenhar Syndrome In A Paediatric Patient: A Case Report

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ABSTRACT:
Goldenhar syndrome, also known as oculo-auriculo-vertebral syndrome is a disorder with craniofacial morphogenesis. Goldenhar syndrome (GS) is characterized by craniofacial anomalies in association with vertebral, cardiac, renal, and central nervous defects. The syndrome is characterized by a classical triad of 1) mandibular hypoplasia resulting in facial asymmetry, 2) ocular and auricular malformations and 3) vertebral anomalies.
In this article, we report a case of goldenhar syndrome describing about clinical features and importance of early diagnosis.

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I. INTRODUCTION

Goldenhar syndrome, also known as oculo-auriculo-vertebral syndrome is a disorder with craniofacial morphogenesis. Goldenhar syndrome (GS) is characterized by craniofacial anomalies in association with vertebral, cardiac, renal, and central nervous defects. The syndrome is characterized by a classical triad of 1) mandibular hypoplasia resulting in facial asymmetry, 2) ocular and auricular malformations and 3) vertebral anomalies.

The incidence of this syndrome, first described by Maurice Goldenhar in 1952, is described between 1:3500 and 1:4500. Male-female ratio of 3:2. The etiopathogenesis is multifactorial and dependent on genetic and environmental factors but there are still many unknown aspects. Abnormalities of chromosomes, neural crest cells, environmental factors during pregnancy like ingestion of drugs, such as cocaine, thalidomide, retinoic acid, intake of alcohol by the mother were also related to development of the disease.

Diagnosis of primary clinical based on history and examination of the eye, face and skeletal system. A multidisciplinary approach is necessary for the overall well-being of the patient and the treatment protocol should be determined as early in life as possible to avoid physical difficulty and psychological stigma to the growing child.

In this article, we report a case of goldenhar syndrome describing about clinical features and importance of early diagnosis.

II. CASE

A 12-year-old patient reported to ADK Jain Eye Hospital, Khekra, UP with the chief complaint of mass in both eyes and abnormal head positioning. There was no history of trauma to head and neck region or maternal exposure to teratogenic agents. No signs of mental retardation or impairment of cognitive function were seen.

On examination, the patient had bilateral limbal dermoids in the inferotemporal quadrant, preauricular tags and torticollis due to hemivertebrae. Vision was 6/6 in both the eyes and the anterior segment and fundus findings were normal. As the limbal dermoid was away from the visual axis and not causing astigmatism or restriction of ocular motility, no surgery for removal was planned. The patient was referred to ENT specialist asked to follow up regularly at 6 monthly interval.

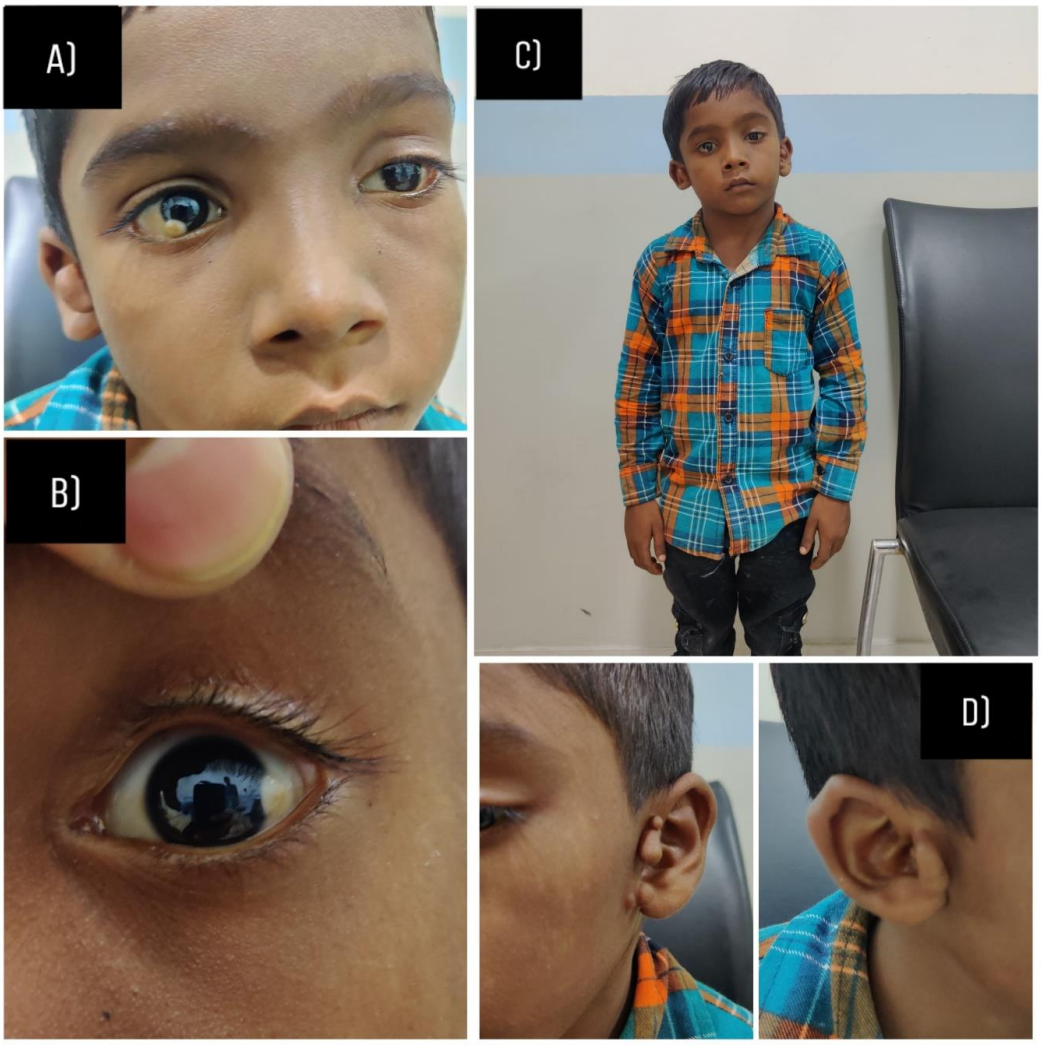

Figure 1: A) and B) showing Inferotemporal and temporal limbal dermoid, C) Hemivertebrae causing torticollis, and D)Preauricular skin tags