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IP International Journal of Ocular Oncology and Oculoplasty

Journal homepage: https://ijooo.org/

Case Report

Oculo auriculo vertebral dysplasia; Goldenhar Syndrome

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ARTICLE INFO

Article history:
Received 25-04-2022
Accepted 03-05-2022
Available online 09-05-2022

Keywords:
Goldenhar's syndrome
Treacher–Collins syndrome

ABSTRACT

Goldenhar's syndrome is a rare condition which was described at first in the early 1950's. which was characterized by a combination of anomalies such as dermal epibulbar cysts, auricular appendices and malformation of the ears. In 1963, the name oculo-auriculovertebral (OAV) dysplasia for this condition was suggested by gorlin and he also incorporated vertebral anomalies as one of the signs of syndrome. Here, we describe a case of 7 month old male patient with preauricular ear tags, limbal dermoid, cleft palate with tongue tie.

1. Introduction

Goldenhar syndrome is a congenital anomaly with very rare occurrence and involves the first and second branchial arches, also known as oculoauriculovertebral dysplasia. Maurice Goldenhar, the French ophthalmologist first described the syndrome in 1952.[1] Its prevalence ranges from 1:3,500 to 1:7,000 live births with male-female ratio of 3:2. Although majority of cases are sporadic, familial occurrences mainly autosomal dominant pattern have been observed.[2]

The exact aetiology of disease is unknown. It is believed to be an abnormality of embryonic vascular supply, disrupted mesoblastic development which affects the formation of branchial and vertebral systems.[3] The diagnosis of Goldenhar syndrome is clinical, however, there are some diagnostic tests that can be helpful. Prenatal ultrasonic diagnosis may theoretically be done at the 11th–15th week in utero.[4] The classical features of Goldenhar syndrome include ocular anomalies - epibulbar dermoids, coloboma and microphthalmia, ENT features such as preauricular tragi, low implantation of the auricular pavilion, hearing loss, micrognathia, and vertebral anomalies such as scoliosis or hemivertebrae. The abnormalities are unilateral in around 85% of the cases. Ocular features, especially bilateral dermoids are seen in 60% of the cases.[5]

2. Case Report

A 7-months old male patient was bought to ophthalmology OPD with chief complaint of mass on left eye since birth. Child was conceived of non-consanguineous marriage and as expressed by parents had difficulty in sucking of milk which was also sometimes occasionally associated with regurgitation of milk from nose. The child was born of a full-term normal delivery and there was no history of any maternal illness during the pregnancy. But the child was cyanotic at the time of birth. Other family members do not have any anomaly. Personal history of child showed normal bowel and bladder habits, undisturbed sleep. Informed consent was taken by patient. The study was approved by institutional ethical committee. On general examination

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https://doi.org/10.18231/j.ijooo.2022.018

child was conscious and cooperative. All the vital signs of the child were within normal limits. There was alteration in the posture as the shoulder levels were not at same level. On facial examination deformed left ear and preauricular tags were seen on the left side. On external oral examination, cleft palate with tongue tie was noted. Ocular examination revealed whitish area near temporal limbus of left side measuring 1*1 cm suggestive of limbal dermoid. There was no history of respiratory infections, chest was clear and on auscultation normal heart sounds were present. We diagnosed the child as a case of Goldenhar syndrome on the basis of preauricular tag, limbal dermoid and cleft palate with tongue tie. X-ray of vertebral column showed scoliosis. Patient was also referred to otorhinolologist, odontologist and paediatrician for expert opinion and further management.

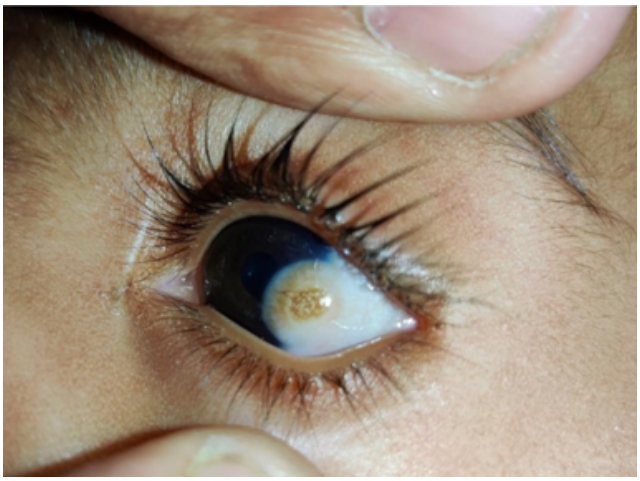

Fig. 1: Clinical photograph showing limbal dermoid

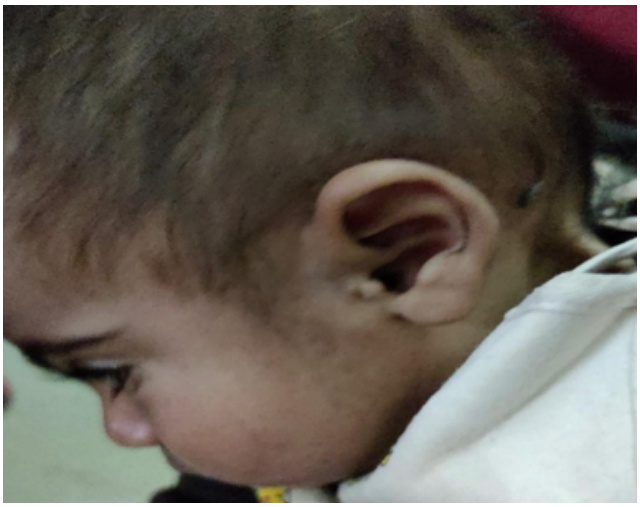

Fig. 2: Photograph showing pre auricular tag

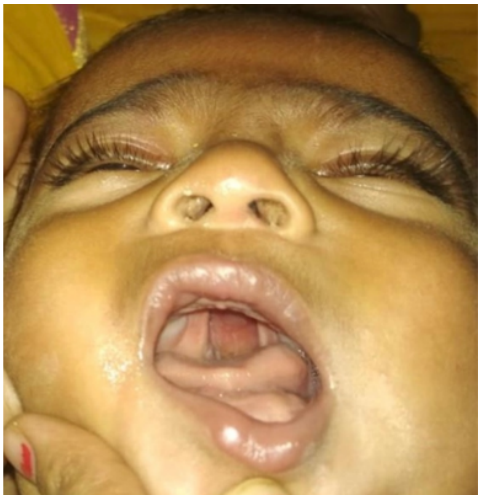

Fig. 3: Photograph showing Cleft palate with tongue tie

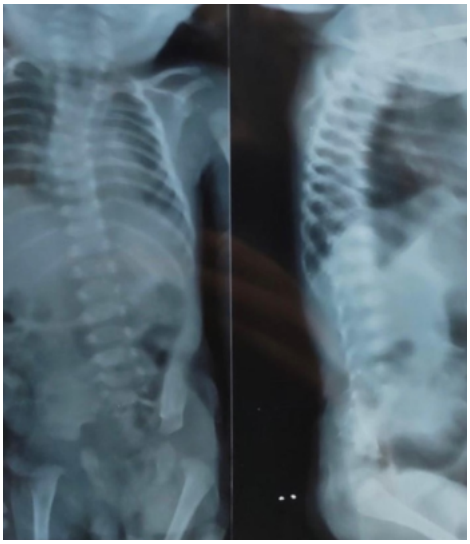

Fig. 4: X-ray image of child showing Scoliosis

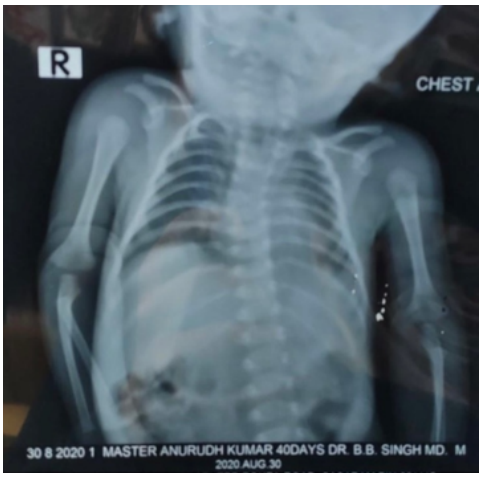

Fig. 5: X-ray image of child showing Scoliosis

3. Discussion

Goldenhar syndrome is a rare congenital defect. The spectrum of observed symptoms and their severity differs among affected patients. The combination of external ear anomalies and ipsilateral facial underdevelopment which includes limbal dermoid and tongue tie is the hallmark of Goldenhar syndrome as seen in our case.

The Differential diagnosis for Goldenhar syndrome includes other congenital anomalies such as CHARGE Syndrome, Parry Romberg Syndrome, or Treacher Collins Syndrome.[6] Treacher–Collins syndrome usually have features which are similar to Goldenhar syndrome but patients usually have a bilateral presentation and mutation in TCOF1 gene on human chromosome 5q31-34 is specifically linked to TCS and helps in its final diagnosis.[6]

The management of the disease varies according to age and systemic associations and the reason for treatment in uncomplicated cases are mainly cosmetic. The external ear reconstruction surgeries can be performed at the age of 6 to 8 years. In patients with milder involvement, jaw reconstruction surgeries are usually done in the early teens whereas epibulbar dermoids should be surgically excised if it is symptomatic or child's parents want cosmetic correction.[7] Current standard medical treatment for grade I paediatric limbal dermoids where only superficial corneal involvement is there is initially conservative. In stages II where it is affecting the full thickness of the cornea with/without endothelial involvement and III where involvement of entire cornea and anterior chamber, a combination of excision, lamellar keratoplasty, and amniotic membrane and limbal stem cell transplantation are advocated.[5,8] The purpose of treatment is to prevent amblyopia (caused by induced astigmatism or obstruction of the visual axis) or aesthetic considerations.[5] Other ocular anomalies are rare but include anophthalmos, microphthalmos, microcornea, strabismus, blepharoptosis, palpebral fissure, iris atrophy, eyelid colobomas, iris and choroid colobomas, motility disorders, polar cataract, anomalous lacrimal drainage system, and retinal and optic nerve anomalies.[9]

Systemic involvement in Goldenhar syndrome can vary widely too. Among cardiovascular anomalies, tetralogy of Fallot and ventricular septal defects are most commonly associated with OAVS.[10] Patient may also suffer from severe obstructive sleep apnoea caused by airway abnormalities and jaw growth deficiency, which could lead to restrictive diet and malnutrition.[11] In cases with systemic involvement prognosis is usually guarded, but it is good in otherwise uncomplicated cases without any systemic associations.[12]

4. Conclusion

Goldenhar syndrome is a relatively rare condition, easy to diagnose based on the clinical appearance without excluding further investigations. The management of golden har syndrome requires a multidisciplinary approach including experts from various specialities such as ophthalmologist, plastic surgeon, otolaryngologist, odontologist.

5. Conflict of Interest

The authors declare that there are no conflicts of interest in this paper.

6. Source of Funding

None.

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Cite this article: Patel R, Maurya RP, Yadav VK, Kumar A, Singh V, Saxena S, Maurya KS. Oculo auriculo vertebral dysplasia; Goldenhar Syndrome. *IP Int J Ocul Oncol Oculoplasty* 2022;8(1):80-83.