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# To Report a Rare Case of Goldenhar Syndrome

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**Abstract:** ***Goldenhar syndrome (GS) is a relatively common developmental disorder characterized by craniofacial anomalies in association with vertebral, cardiac, renal, and central nervous system defects. This paper describes GS features with special emphasis on bilateral involvement. The clinical features of this patient with GS aged 22 are described, and a brief review of the literature about this genetic disorder is also presented.***

**Keywords:** Goldenhar Syndrome, Bilateral, Limbal Dermoid, Preauricular Tag

## 1. Introduction

- Goldenhar syndrome (GS), also known as oculoauriculovertebral spectrum or hemi facial microsomia has a wide range of clinical manifestations, including craniofacial, vertebral, cardiac, renal, and central nervous system anomalies.(1)
- GS is a Autosomal dominant condition with a prevalence ranging from 1:5, 000 to 1:7, 000 live births, and a male-female ratio of 3:2.(2)
- Soft tissue involvement is more in this condition than bony involvement which is seen in teacher-Collins syndrome.

## 2. Case Report

A 22yr old maleborn of nonconsanguineous marriage, presented with history of whitish mass in left eye since birth. There was no increase in the size of the mass. There was no history of any scholastic backwardness, convulsions or deafness. The child was born of a full-term normal delivery and there was no history of any maternal illness during the pregnancy. All other family members including one younger sibling is normal.

**Upon Ocular examination**

**Figure 1:** Left Limbal Dermoid

| | OD | OS |
|---|---|---|
| UCVA | 6/6 | 6/36 |
| Pin Hole Improvement | - | No Improvement |
| BCVA | 6/6 | 6/36 |

**Systemic examination:**

**Figure 2:** Facial asymmetry- Mandible deviated to right side, limbal dermoid on left side (opposite)

**Figure 3:** Pre auricular tags

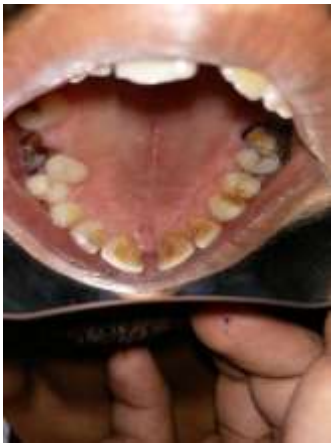

**Figure 4:** High arched palate

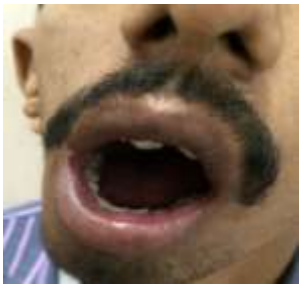

**Figure 5:** Lower midline shifted towards right

**Figure 6:** Anterior and right posterior open bite and Central diastema noted
Non palpable right condyle of mandible
Left condylar movement normal

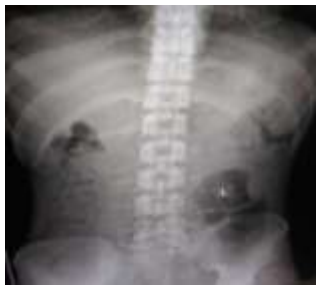

**Figure 7:** X-ray of spine- normal

In this case USG abdomen was normal

**X ray and Orthopanthomogram (Fig 8, 9, 10):**

- Gross asymmetry of right side
- Hypoplasia of malar bone
- Hypoplasia of right lower border of mandible
- Right sided condyle outside the glenoid fossa, smaller in size
- Vertical height of ramus reduced to one third
- Aplasia of right coronoid process
- Zygoma appears short, appears at higher level on right side

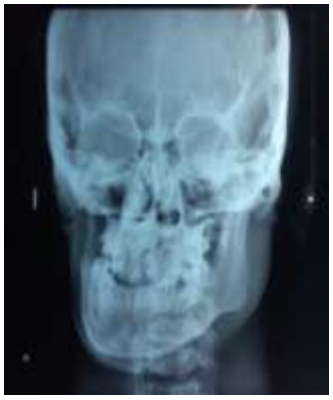

**Figure 8**

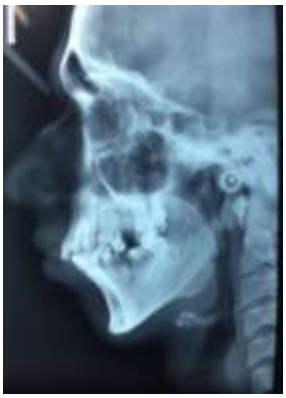

**Figure 9**

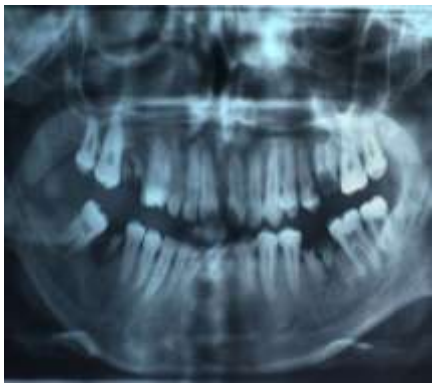

**Figure 10**

## 3. Discussion

It is a rare hereditary condition characterized by numerous anomalies affecting the first and second branchial arches of the first pharyngeal pouch, the first branchial cleft, and the primordia of the temporal bone before the end of organogenetic period (7th or 8th week of embryonic life)[(3)].The triad of anomalies comprising this syndrome include accessory auricular appendages, aural fistulae and epibulbar dermoids.[(4)]Inheritance is of Autosomal dominant type.The aetiology of GS remains unknown. Hartsfield (2007) reviewed the literature and suggested that GS is a result of some type of vascular perturbation and/or neural crestopathy during a critical time of embryogenesis. [(1)]The abnormalities are found to be unilateral in 85% of cases and bilateral in 10-33% cases. [(5)]The treatment of the disease varies with age and systemic associations .Reconstruction

surgeries of the external ear may be performed. In patients with milder involvement, jaw reconstruction surgeries can be done in the early teens; epibulbar dermoids should be surgically excised.[5, 6] Structural anomalies of the eyes and ears can be corrected by plastic surgery.[6] Prognosis of the disease is good in otherwise uncomplicated cases without any systemic associations. Successful treatment requires a multidisciplinary approach involving otolaryngologists, ophthalmologist, pediatrician, dermatologist, orthopedician.

## 4. Conclusion

This is a rare case wherein the soft tissue involvement is on one side (right) and the dermoid is on opposite side (left) which is in limbal position.

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