

# Goldenhar Syndrome: A rare case report

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## ABSTRACT

**Introduction:** Goldenhar syndrome (GS), or oculo-auriculo-vertebral syndrome, is a poly-malformative syndrome associating most often an epibulbar dermoid cyst, pretragial diverticula and vertebral anomalies. We report a rare observation of Goldenhar syndrome associating lateral labial cleft, dermoids of the ocular limb and pre-tragic excretion.

**Materials and Methods:** This is the observation of an 2 years female child presenting a Goldenhar syndrome associating lateral cleft lip, dermoids of the ocular limb, preauricular appendices and pre-auricular diverticula. No family history or other symptoms were recorded. The baby , had a normal auditory meatus without any evidence of skin abnormalities stromal .radiograph of the spine, echocardiography did not show any associated abnormalities.

**Discussion:** Goldenhar syndrome is a rare malformation syndrome related with an anomaly of development of the first gill arches. Although most cases of SG was sporadic, the modalities of autosomal recessive inheritance and the dominant have been described. Also referred to as oculo-atrial dysplasia, it is most often unilateral affecting the soft tissues and less the bone component. It usually include peribulbar dermoids or dermolipomas, pre-auricular cuticular diverticula, Abnormalities of the shape of the auricle, conductive hypo-acoustics, abnormalities of the facial area and sometimes vertebral anomalies. It can be added to These malformative alterations a coloboma of the eyelid, a Duane syndrome And very rarely microphthalmia or anophthalmia.

**Conclusion:** Through this observation, we will discuss the pathogenic aspects and clinics of this rare clinical entity and the therapeutic measures taken.

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## INTRODUCTION

Goldenhar syndrome or facio-auriculo-vertebral dysplasia is a complex syndrome characterized by a combination of maxillomandibular hypoplasia, ear deformity, ocular dermoid and vertebral abnormalities .

in the most severe forms of hemifacial microsomia.

Rare pathology, whose prevalence is estimated between 1 / 5,600 to 1 / 20,000 births. Most cases are not familial, occur sporadically.

The genetic origin of the disease is complex and heterogeneous (several genes and chromosomes would be involved).

We describe the case of a 2-year-old patient with a form considered minor of the disease.

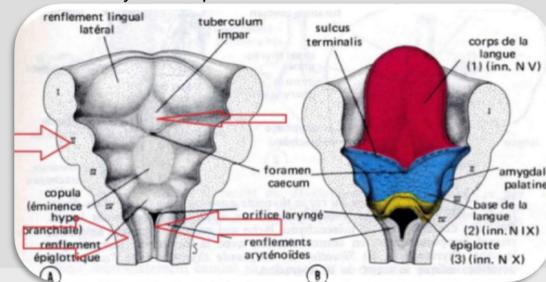


Figure 1. Clinical photo showing unilateral cleft .



Figure 2. multiple accessory tags.



Figure 3. preauricular tags .



Figure 4. eyelid coloboma of the left eye.

## RESULTS



Figure 5,6,7. Postoperative aspect

## OBSERVATION

2 years-old patient reported to the Department of pediatric surgery ,Constantine Algeria, with a transverse cleft present in the left angle of mouth, with incompetent lips epibulbar dermoid cyst, pretragial diverticula (bilateral preauricular tags) There was no history of trauma to head and neck region or maternal exposure to teratogenic agents. The remainder of the clinical examination, moreover, was without any abnormality. Echocardiography as well as the other para-clinical investigations, radio-scan; abdominopelvic ultrasound, were without abnormalities. Correlating the history and clinical findings a final diagnosis of Goldenhar Syndrome was given (Fig.1.2.3.4).

The intervention under general anesthesia was done, in which the orbicularis muscle was reconstructed by reconstruction of the angle of the lip on its mucous and cutaneous surface. Excision of pre-auricular appendicitis. the patient was referred to the ophthalmic department, where she underwent a simple excision of limbic tumors (Fig.5.6.7.8). The postoperative evolution was very favorable from an esthetic point of view, without complication ;a retreat of a year later.

## DISCUSSION

Goldenhar's syndrome, or oculo-auriculo-vertebral syndrome, affects the soft tissues more often than bone structures it usually includes cleft lip, dermoid or dermolipoma of limb, pre-auricular cutaneous diverticula, fistulas in the same area, malformations of the external ear , It can be added to these malformative changes: colobomas of the upper eyelid, conductive hypoacusis, bilateral asymmetry of face and macrostomia, abnormalities of the lower part of the facial mass (macrostomia, micrognathia, partial inoclusion of the dental arches), costal or vertebral abnormalities and finally visceral [1,2]. GS syndrome is made up of anomalies, mainly of first and second branchial arch derivatives. Characteristic features include structural malformations of the external and middle ears, face, and jaw. It has been suggested in first that hemorrhage involving the first and second branchial arches causes hypoplasia and malformation of the face and auricle, this theory cannot explain the multisystemic manifestations of this disease. The most accepted theory. This would imply a molecular mechanism involving cell adhesion molecules that unify the two disease processes and explain the multisystem anomalies of the GS. The aim of the surgical management is to create symmetrical commissure with minimal scarring[1]. The reconstruction of the commissure needs proper attention[3]. There are various techniques, like Z plasty, W plasty, simple linear flap and triangular flap .more important factors Correction are to correct different function as, speech problems and compromised chewing ability . in our patient, We have used the triangular flap technique The well-repaired orbicularis oris counters the lateral contractile force of the skin scar.

## CONCLUSIONS

Goldenhar syndrome is a rare multifactorial disease. Clinically stands as a combination of several abnormalities such as limbal tumors, malformation of the ears of different degrees, as well as cardiac and vertebral malformations, cleft lip or palate of different stages. delicate care

## REFERENCES

- [1]Goldenhar syndrome: a report of 3 cases. Gaurkar SP et al,Indian J Dermatol. 2013
- [2]Multiple accessory tragi as a clue to the diagnosis of the oculo-auriculo-vertebral (Goldenhar) syndrome.Miller TD1, Metry D.2004
- [3]The etiology research progress of oculo-auriculo-vertebral spectrum.Chinese