## Goldenhar Syndrome- Case Report

*Dr.Bipin Rathod, **Dr. Sunil Mhaske, *Dr.Thaslima K, *Dr. Ninza Rawal, ***Dr. Ramesh Kothari, *Dr. Ganesh Misal

*Resident, ** Professor & Head ,***Asso. Prof.

**Corresponding Author :** Dr. Bipin Rathod

**Mail id -** bipinrathodamt@gmail.com

**Mobile No. -** 9767705959

**Address :** Department of Paediatrics,Dr.Vithalrao Vikhe Patil Medical College, Ahmednagar(MS) - 414111

### Abstract

Goldenhar Syndrome also called as oculo-auriculo-vertebral dysplasia, is a rare syndrome developing from first and second pharyngeal arches during Blastogenesis. This condition was documented in 1952 by Maurice Goldenhar. The syndrome is characterised by multiple anomalies of the ocular, auricular, cardiac, skel etal and nervous system. Pericentric inversion of chromosome 9 is one of the most common structural balanced chromosomal aberration seen in this syndrome. Multi disciplinary approach should be undertaken by several departments in evaluating such patients.

**Key Words:** Epibulbar dermoid, Micrognathia, Preauricular tags.

**Introduction :**The Goldenhar syndrome or Facioauriculovertebral sequence or FAv sequence was first described in 1952 by the French ophthalmologist Maurice Goldenhar.[1]

Prevalence is 1–9/100,000,[2] with an incidence of 1 in 25,000–45,000 births, with a male to female ratio of 3:2.[3] it is due to problems that occur when the fetus is forming within the womb of the mother, in structures known as the "first and second brachial arch". These structures will develop to form the neck and the head. The cause is still unknown.[4] Goldenhar syndrome is part of a group of conditions known as "craniofacialmicrosomia". Major manifestations of HFM are orbital distortion, mandibular hypoplasia, ear anomalies, nerve involvement and soft tissue deficiency (OMENS classification).

**Case Report :** A newborn male child born of non-consanguinous parents was delivered vaginally at full term . The antenatal, intranatal and postnatal periods were uneventful. His parents were non consanguineous, and there was no relevant family history. On examination, the child weighed 2.9 kg and showed the following abnormal features.

**Head to toe examination :**

**Facial features:** Small size of the right mandibular region, tongue tie, high arched palate, depressed frontal region above the left medial canthus, depressed bridge of the nose .

**Ears:** The child had absence of right external ear , low set ears with bilateral pre-auricular tags .

**Eyes:** Ophthalmologic examination revealed a yellowish white sub-conjunctival mass measuring 0.5 mm × 0.5 mm, located at the nasal limbus at the 9 O'clock position in the left eye. Single umlical artery was seen.

**Systemic examination**

**Cardiovascular examn :** pansystolic murmur present

Rest examination were normal

His investigations revealed, normal haemogram, normal blood urea nitrogen and serum creatinine. X-ray chest AP and Lateral view- Scoliosis of thoracic and cervical spine

X-ray abdomen normal ,X-ray skull shows asymmetric mandible.Abdominal ultrasound- Left kidney agenesis & Right kidney normal

**Fig.1-Newborn with facial dimorphism**

**Fig.2- Infant with severe capillary abnormalities & doughy skin**

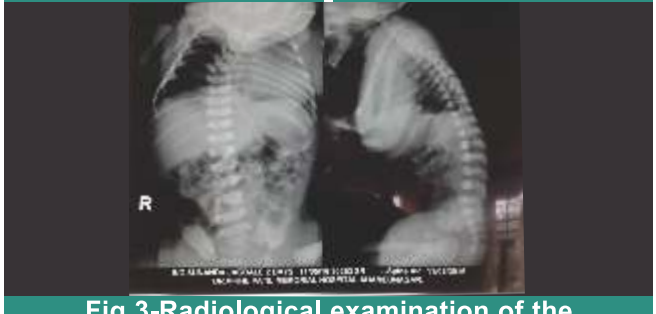


**Fig 3-Radiological examination of the skeleton showed asymmetry of the limbs.**

**Fig 4-Absent left pinna with preauricular tag**

**Fig 5-Single umblical artery**

**Discussion :** Goldenhar syndrome (oculoauriculovertebral dysplasia with hemifacial microsomia) is a rare congenital anomaly involving the first and second branchial arches. It is a disorder where the patient's facial features are incompletely developed on one side, resulting in eye, ear, and jaw abnormalities. When these facial abnormalities are associated with vertebral malformations in the spine and more severe involvement of the eyes, this collection of symptoms is called Goldenhar syndrome[1]. In 85% of patients with Goldenhar syndrome, only one side of the face is affected.The characteristic combination of external ear anomalies and ipsilateral facial underdevelopment is the hallmark of this syndrome Cervical spine vertebral deformities are part of the collection of symptoms[5].

The treatment of the disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases. Reconstruction surgeries of the external ear may be performed at the age of 6 to 8 years. In patients with milder involvement, jaw reconstruction surgeries can be done in the early teens; epibulbar dermoids should be surgically excised. [2] Structural anomalies of the eyes and ears can be corrected by plastic surgery.[2] 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.

Importance of diagnosis and consequences of avoiding treatment Severe cases of Goldenhar syndrome or hemifacial microsomia can affect many aspects of the patient's life and sometimes requires immediate intervention from birth. For example, the patient may suffer from severe obstructive sleep apnea due to airway abnormalities Jaw problems may result in a restrictive diet and malnutrition, and issues with the eyelids may lead to subsequent vision problems. Without hearing assistance or surgical reconstruction, individuals with full bilateral microtia and atresia will lead to permanent deaf mutism.

The etiology of this rare disease is not fully understood Numerous hypothesis have been proposed to explain the etiopathogenesis of this syndrome:

Gorlin and pindborg 1964,suggested that some abnormal process affects the mesoblasts embroyologically which affects branchial and vertebral systems thereby resulting in syndrome. Krause in 1970 suggested hereditary pattern to be causative agent as he described the syndrome affecting a brother and sister. Jong bloet in 1971 ,suggested that goldenhar syndrome may be result of fertilization of an overripe ovum.Baum and Feingoid in 1973,stated that goldenhar's syndrome may be sporadic event that occurs early in embryogenesis which is explained by reduced penetrance ,somatic mosaicism or epigenetic changes. Poswillo in 1976 using animal model, showed and suggested that maternal fetal hypoxia ,hypertension and anticoagulants can result into haematoma in the region of the ear and jaw which expands and causes destruction of differentiating tissue. Gomez et al in 1984 hypothesized about the role of radiological intervention like cholecystography which is practised between 4th and 6th week of pregnancy as a causative factor [6].

**Differential diagnosis**[7]

- Treacher-Collins syndrome,
- Wolf-Hirschhorn syndrome,
- Nager's acrofacial dysostosis,
- Wildervanck syndrome (cervicooculoacoustic syndrome),
- Townes-Brocks syndrome, and
- Delleman syndrome.

Treacher Collins syndrome is associated with maxillary and mandibular hypoplasia but is not associated with ocular and aural anomalies.Wolf-Hirschhorn syndrome the eyes are widely spaced and may be protruding. Other characteristic facial features include a shortened distance between the nose and upper lip (a short philtrum), a downturned mouth, a small chin (micrognathia), and poorly formed ears with small holes (pits) or flaps of skin (tags). Additionally, affected

individuals may have asymmetrical facial features and an unusually small head (microcephaly).Nager syndrome often have eyes that slant downward, absent eyelashes, and a notch in the lower eyelids called an eyelid coloboma. Many affected

individuals have small or unusually formed ears. Wildervanck syndrome is typically characterized by three primary findings (triad). These include abnormal union or fusion of two or more bones of the spinal column (vertebrae) within the neck (Klippel-Feil syndrome); impairment or absence of certain eye (ocular) movements and hearing impairment that is present at birth.

**What is new?**

1. Though earlier reports showed male preponderance lately majority female cases have been reported.
2. Some of the patients can also have high arched palate, gingival hypertrophy, malaligned teeth, bulbous nose, vitiligo as an atypical association.
3. The treatment of the disease is now possible and varies with age and systemic associations and is mainly cosmetic in uncomplicated cases.

In patients with mandibular hypoplasia, reconstruction can be done with rib grafts and an underdeveloped maxilla can be lengthened by a bone distraction device. In patients with milder involvement, jaw reconstruction surgeries can be done in the early teens; epibulbar dermoids should be surgically excised.

**Conclusion :** The rarity of Goldenhar Syndrome and its heterogeneity of spectrums demonstrate the multifactorial feature of this pathology. Having as classic triad the ocular, auricular and vertebral alterations, they present the hemifacial microsomia and the absence of right external auricular pinna as more frequent alterations. Once it is probably a multifactorial dysgenetical syndrome, the genetic counseling and the detailed study of the cause in each patient affected by this syndrome is necessary, once associated factors such as ingestion of drugs during the gestation, gestacional diabetes, alcohol ingestion during the pregnancy, amongst others, can be prevented, avoiding the appearance of new cases as much as possible.

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