Case Report

# Oculo-auricular-fronto-nasal syndrome – a new entity

**Sujata Chahande[1], Ramesh Murthy[2]**

**We describe a unique combination of fronto nasal malformations and oculo auricular vertebral syndrome without any vertebral defects and with bilateral involvement of eyes. The oculo auricular vertebral syndrome manifests with defects of the first and second branchial arches including hemifacial microsomias and Goldenhar's syndrome. We describe the clinical features in a 6-year-old male child with lid colobomas, ocular dermoids and dermolipomas, hearing defects, zygomatic hypoplasia but with bilateral involvement and absence of heart disease, absence of encephalocoele and without any maxillary or mandibular hypoplasia. We name this unique entity as Oculo-auricular-fronto-nasal syndrome.**

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***Key words*** **: Corneal dermoid, lipodermoid, preauricular skin tags, lid coloboma, zygomatic.**

Fronto-nasal malformations (FNM) is a developmental field defect representing abnormal morphogenesis of the fronto-nasal eminence while oculo-auricular-vertebral spectrum (OAVS) describes a more broader range of the first and the second branchial arch defects including hemifacial microsomias and Goldenhar's syndrome[1]. We describe a 6 year old boy with a rare combination of features of fronto-nasal malformations (FNM) and oculo-auricular vertebral syndrome (OAVS) without any vertebral defects and with a bilateral involvement. The uncommon features in our patient was the absence of heart disease, absence of encephalocoele, bilateral involvement, no maxilla or mandibular involvement with the presence of zygomatic hypoplasia.

## Case Report

A 6 year old male patient born of non- consanguineous marriage presented with congenital deformities of upper eyelids, preauricular skin tags and ocular dermoids. There was no positive family history of similar eye anomalies.

***Examinations*** — Patient had vision of finger counting more than 3m in each eye. There was bilateral total complete upper lid coloboma which was quadrangular involving medial third of upper eyelid and about one third eye lid width (Fig 1). Patient had broad dorsum of nose and hypertelorism along with telecanthus. The conjunctiva showed presence of a lipodermoid temporally in the right eye lateral fornix along with a limbal dermoid located at the inferior limbus from 5 to 7 o'clock which was confluent with the lipodermoid. In the left eye there was a limbal epithelial dermoid at the inferior limbus from 4 to 8 o'clock encroaching on the cornea upto the mid pupillary zone. There was no sign of exposure keratitis. The pupils were briskly reacting to light. Patient's intraocular pressures were found to be normal. Fundus examination revealed the presence of tilted optic disc in both eyes. The ocular motility was normal.

Patient had multiple pre auricular skin tags, four on the right side and two on the left with no external ear deformity (Fig 2). Patient had left hemi-facial microsomia with misaligned teeth along with cleft palate. There was decreased hearing due to conductive deafness. The upper and lower jaws were normal but with zygomatic hypoplasia. The patient had delayed milestones.

X-Ray of the skull confirmed the presence of bilateral zygomatic hypoplasia with normal maxillary and mandibular bones. X Ray spine did not show any vertebral anomaly. Computer aided tomography of brain was normal.

## Discussion

The minimum clinical features for the diagnostic confirmation of Goldenhar's syndrome is the presence of unilateral ear involvement (microtia) together with preauricular tags[2]. Ocular abnormalities associated with Goldenhar's syndrome include microphthalmia, anophthalmos, epibulbar dermoid and lipodermoid and strabismus[3]. Bilateral ocular involvement which was present in our patient, is rare 10%[4].

Fig 1 — Face photograph of a 6 year old boy with bilateral total complete upper lid coloboma, broad dorsum of nose and hypertelorism along with telecanthus, lipodermoid temporally in the right eye lateral fornix along with a limbal dermoid located at the inferior limbus & a limbal epithelial dermoid at the inferior limbus in the left eye & 2 preauricular skin tags on left side

Department of Ophthalmology, BJ Government Medical College & Sassoon General Hospital, Pune 411001
[1]MS, DOMS, Associate Professor and Corresponding author
[2]FRCS, Medical Director, Pediatric Ophthalmology, Strabismus and Oculoplasty, Axis Eye Clinic, Pune 411038

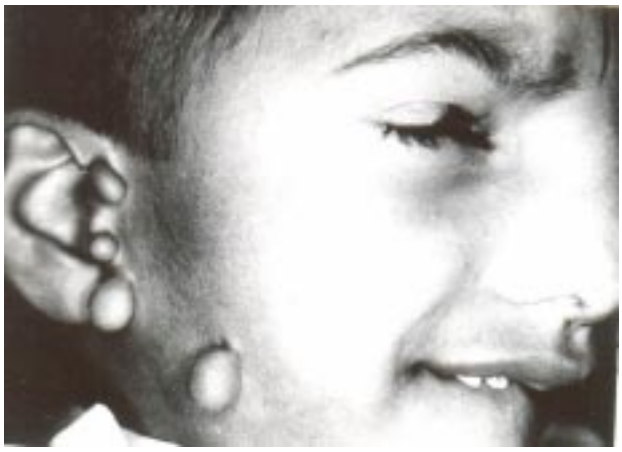

Fig 2 — Face photograph of a 6 year old boy right lateral view with multiple pre auricular skin tags, four on the right side with no external ear deformity with misaligned teeth along with cleft palate and with no corneal exposure on eyelid closure

The NORD studies suggest that in individuals with this disorder, craniofacial malformations resulting in facial asymmetry (hemifacial microsomia) is progressive and initially affects bone and soft tissue of lower jaw (mandibular hypoplasia). As the disease process progresses, asymmetry of the lower jaw becomes more pronounced and causes associated malformations of upper jaw (maxilla), nose and orbit. Our patient was unique as he had no maxilla or mandibular involvement but had zygomatic hypoplasia. Zygomatic hypoplasia has classically been described as a feature of Treacher-Collins Syndrome. In addition the child had conductive hearing loss and cleft palate which is common to both Goldenhar's syndrome and Treacher- Collins syndrome[2,5].

The absence of vertebral involvement is another unique feature of this case. The incidence of vertebral involvement in Goldenhar's Syndrome is reported to be as high as 40%[6]. However some studies mention that there was no absolute correlation between the presence of any of the formative or segmentation vertebral defects and that of other concomitant malformations and thus patients with various groupings of Goldenhar related anomalies should be considered a single entity to which Goldenhar's association could be applied[7].

## Conclusion

To conclude, though the present case does not have all the features typical of Goldenhar's Syndrome or Treacher-Collins Syndrome, it is unique in the presentation of bilateral involvement, zygomatic hypoplasia without vertebral and spinal involvement. We would therefore describe it as Goldenhar's Syndrome having a unique syndrome pattern or a distinct entity. We would like to label this entity as 'Oculo-Auriculo-Fronto-Nasal Syndrome' .

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