

# Goldenhar syndrome

## Abstract

Goldenhar syndrome is a hereditary condition which is characterized by preauricular appendages, epibulbar dermoids, along with anomalies of the heart, kidney, and vertebral, central nervous and gastrointestinal system. We report a case of Goldenhar syndrome in a 7-years-old male patient who presented with limbal dermoid, pre auricular tag, microtia and mandibular hypoplasia. We AIM to report this case, given the rarity of this syndrome, and classical signs seen in our patient.

**Keywords:** goldenhar, ocular, ear

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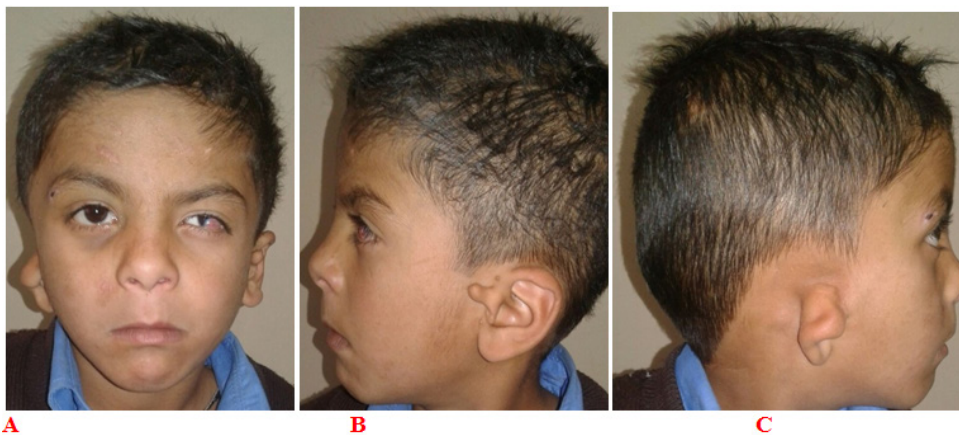
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## Case report

A 7 year old boy was brought by his grandfather (farmer by occupation) to our outpatient department. The only complaint put forward by the grandfather was that the boy had defective vision and hearing and he had come only for getting a disability certificate for the child. On a first hand gross examination of the child, the child had limbal dermoid in the left eye, pre auricular tag (left side), right sided microtia and mandibular hypoplasia (Figure 1) consistent with the diagnosis of Goldenhar syndrome. Rest no other gross anomaly

was seen. We explained about the disease to the grandfather in great detail and further stressed the need of a complete history plus a multispeciality examination of the child (besides detailed ocular examination) as various organs can be involved in this syndrome before a disability certificate can be issued plus other asymptomatic disease could be diagnosed. We also made it clear that all the process may take a few days. But as the grandfather was only interested in getting the disability certificate for the child and not in the child's health, he thanked us and left. We never had a follow up with the child again.



**Figure 1** On a first hand gross examination of the child, the child had limbal dermoid in the left eye, pre auricular tag (left side), right sided microtia and mandibular hypoplasia (A,B&C) consistent with the diagnosis of Goldenhar syndrome.

## Discussion

Goldenhar syndrome was documented in 1952 by Maurice Goldenhar. Males are affected more than female (3:2) and its incidence has been reported to be around 1:3000 to 1:5600. It is also called as oculo-auriculo-vertebral syndrome because of the association of eye anomalies with or without vertebral and ear anomalies. The aetiology is thought to be an abnormal embryonic vascular supply, disrupted mesodermal migration leading to the defective formation

of the branchial and vertebral systems.<sup>1</sup> Ingestion of drugs such as thalidomide, retinoic acid, tamoxifen, and cocaine by the pregnant mother plus maternal diseases like diabetes, rubella, and influenza have also been implicated in causing this syndrome.<sup>2</sup>

Clinical features include eye abnormalities in the form of microphthalmia, eyelid and iris coloboma, epibulbar limbal dermoids, squint, and retinal abnormalities. Ear abnormalities are in the form of acrotia, microtia, preauricular tag, auricular fistula and conductive

hearing loss. Orofacial manifestations include malocclusion to complete absence of the mandibular branch and temporomandibular joint, micrognathia, macrostomia, cleft lip, cleft palate, fissured tongue, and delayed tooth eruption, supernumerary teeth and facial asymmetry. The vertebral abnormalities are supernumerary vertebrae, hemivertebrae, fused vertebrae, rib anomalies, scoliosis, kyphosis, and skull abnormalities. Goldenhar syndrome may be associated with cardiovascular, pulmonary, renal, genital, central nervous system and gastrointestinal abnormalities.<sup>3</sup>

For diagnosis to be confirmed, the subject should at least have microtia and preauricular, or auricular abnormalities.<sup>4</sup> According to another study, at least two of the following findings must be present for diagnosis of this syndrome: unilateral microtia, unilateral mandibular hypoplasia, epibulbar dermoid cysts (unilateral or bilateral) or vertebral malformations.<sup>5</sup> Although diagnosis is mainly clinical, radiographic investigations help to support the clinical diagnosis. Prenatal diagnosis is possible with considerable accuracy with ultrasound which may detect obvious defects. Since no specific genes have been linked to this syndrome, prenatal deoxyribonucleic acid testing cannot be used to diagnose the condition.<sup>6</sup> The treatment is usually surgical and the modalities are jaw distraction/bone graft, ocular dermoid removal, repairing cleft palate/lip, cardiac surgery, spinal surgery, prescribing hearing aid etc.<sup>7</sup>

## Conclusion

Goldenhar syndrome is a rare abnormality affecting the head and neck region with multisystem involvement. The early recognition of this syndrome by a doctor of any speciality increases the chances of an early systemic workup in such cases so that any underlying non symptomatic drastic disease may be treated at the earliest.

## Conflicts of interest

The author declares that there is no competing interest.

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