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GOLDENHAR SYNDROME –A CASE REPORT

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ABSTRACT Goldenhar Syndrome or Oculoauriculovertebral syndrome is a complex syndrome characterized by an association of maxillomandibular hypoplasia, deformity of the ear, ocular dermoid and vertebral anomalies. Here, we describe a 26-year-old male patient with ocular dermoid and preauricular ear tags.

KEYWORDS : Ocular dermoid, preauricular tags, vertebral anomalies

INTRODUCTION

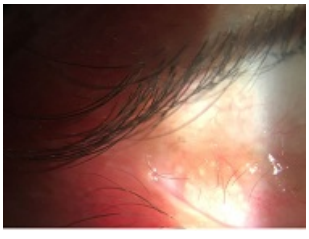
In 1881, the first observation of oculo-auriculo-vertebral (OAV) dysplasia was reported by Von Arlt (1) and in 1952, Dr. Maurice Goldenhar, a renowned Swiss ophthalmologist classified the clinical features and named the malformation complex as Goldenhar Syndrome and described it as a congenital defect characterized by group of malformations classically involving the face, eyes and ears. (2)

The incidence of Goldenhar syndrome has been reported to be 1:35,000-1:56,000 with a male to female ratio of 3:2. Etiology of this condition is not yet fully established. Abnormalities of chromosomes, neural crest cells, environmental factors during pregnancy like ingestion of drugs, such as cocaine, thalidomide, retinoic acid, intake of alcohol by the mother were also related to the development of the disease. Clinically, the patient may present with a variety of features ranging from facial abnormalities, ear abnormalities, eye abnormalities, vertebral defects, and congenital heart problems.

In this article, we report a case of Goldenhar syndrome along with discussion on clinical features, importance of early diagnosis, and interdisciplinary approach to manage it.

CASE REPORT

A 26-year-old patient reported to the Department of Ophthalmology with the chief complaint of mass growing in his left eye. There was history of lower back ache and repeated episodes of pneumonia. No signs of mental retardation or impairment of cognitive function were seen. There was no history of maternal exposure to teratogenic agents. On ocular examination dermoid was seen with hairs on its surface on the temporal bulbar conjunctiva (Fig1) preauricular tags were present on the right side (Fig 2). Orthopedic reference was done as patient also complaints of repeated episodes of lower back ache.



(Fig 1)

(Fig 2)

DISCUSSION

Most of the patients reported within the OAV spectrum, a term proposed by Gorlin *et al*. (1990) are sporadic.(3) The association of epibulbar dermoids, preauricular fistulae, abnormalities of skin appendages and ocular malformations as a specific entity involving the first and second branchial arches was recognized by Goldenhar (2). Pretragal fistulae, epibulbar dermoids and accessory auricular appendages are triad of Goldenhar Syndrome. The diagnosis is further substantiated if vertebral anomalies are present in addition to the triad. The diagnosis becomes more difficult if additional symptoms are present.

Cranial nerve involvement has been considered part of the OAV spectrum and the facial nerve involvement is cited most commonly.(4) Some other etiologic factors include maternal vasoactive medication use (especially in conjunction with smoking) in the first 10 weeks of gestation, primidone, retinoic acid and thalidomide embryopathy and maternal (preexisting or gestational) diabetic embryopathy.(5)

In the 2nd month of embryonic development, aberrant fusion of the lateral portions of the maxillary and mandibular swellings results in macrostomia, usually associated with skin tags and pits between the comer of the mouth and the tragus.(6)

The prevalence of Goldenhar Syndrome in Indian population is very low. A study was taken up to understand the prevalence of this syndrome in children below the age of 14 years with hearing loss. Out of 1073 children, this syndrome was observed only in 1 (0.09%) case.(7)

The effect of Goldenhar Syndrome is more evident as the child grows, because of delays in the growth and the development of the affected areas. Primary reconstruction typically consists of a cleft repair, corrections of colobomas and ear deformities and removal of dermoids and preauricular tags.(8) The complex treatment is focused not only on dental care, articulation and hearing but also on the prevention and treatment of the psychosocial aspects of the malformation. Treatment requires constant follow-up and reassessment of the results.

The study of this condition is still controversial because the symptoms and the physical features may vary greatly in range and severity from case to case.

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