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 constellation of malformations classically involving the face, eyes and ears.\(^2\)

In 1960, Gorlin and Pindborg noted that the disorder varied from mild to severe and the facial involvement occurred unilaterally as well as bilaterally. In 1963, Gorlin et al. suggested the use of the term oculoauricularvertebral dysplasia to describe the syndrome characterized by epibulbar dermoids, auricular appendages, blind-ended auricular fistulas and vertebral anomalies.\(^3\) They considered this syndrome to be a variant of this complex.

It is otherwise known as oculoauriculo-vertebral syndrome, hemifacial microsomia, first arch syndrome, first and second branchial arch syndrome, Goldenhar–Gorlin Syndrome, lateral facial dysplasia, unilateral craniofacial microsomia, otomandibular dysostosis, unilateral intrauterine facial necrosis and auriculo-branchiogenic dysplasia. Facio-auricular dysplasias represent a single disorder with great variability of expression and an isolated ear malformation may represent the mildest expression of the disorder.\(^4\)

Abnormalities are unilateral in 85% of cases and bilateral in 10–33% of the cases and the right side is more frequently affected.\(^5\) The incidence of Goldenhar Syndrome has been reported to be varying from 1:3500 to 1:5600 live births and it is present in 1:1000 children with congenital deafness with a male to female ratio of 3:2.\(^6\) The disease is seen as sporadic and its etiology is not fully understood; however, positive family histories have been reported suggesting autosomal dominant or recessive inheritance. Some researchers have suggested that multifactorial inheritance are caused by the interaction of many genes, possibly in combination with environmental factors.\(^7\)

Patients with Goldenhar Syndrome may exhibit a wide range of anomalies as described in Table 1.\(^8-22\)
A 25-year-old male patient reported to the Department of Oral Medicine and Radiology with the chief complaint of malalignment of teeth since childhood. History revealed that the patient had difficulty in hearing from the left ear since childhood.

On extra-oral examination, a transverse cleft was present in right angle of mouth, with preauricular tissue tags and incompetent lips. Bilateral asymmetry of face and macrostomia was present and inter-incisal distance was 43 mm. On intraoral examination, 16 and 47 were missing with spacing between 11 12 13 14 42 43 44, proclined 11 12 13 43, cross bite in relation to 24 34, increased overjet and high arched palate [Figures 1-5].

Correlating the history and clinical findings a provisional diagnosis of hemifacial microsomia was given. Differential diagnosis of Goldenhar Syndrome was considered but could be difficult because of the variety of clinical signs. Collins and
Wildervanck Syndrome (Syndroma cervicoocuioacusticum) must also be distinguished. Goldenhar Syndrome can be classified as a variant of hemifacial microsomia, OAV spectrum, or OAV Dysplasia by some authors.[20]

Other craniofacial anomalies such as Treacher's Collin's Syndrome, mandibulofacial dysostosis,[23] Miller Syndrome, a very rare genetic condition, often referred to as “postaxial acrofacial dysostosis” must also be ruled out. These disorders are characterized by distinctive craniofacial malformations that occur in association with limb abnormalities.[24]

Investigations were done where OPG revealed reduction in size of body of mandible, condyles, coronoid process and ramus on the right side. The border of the mandible was well defined and intact. The trabecular pattern and the course of mandibular canal were normal with normal surrounding structures. PA view of skull was taken to rule out any developmental anomaly. It revealed reduction in size of body of mandible, condyles, coronoid process and ramus on the right side of mandible with normal surrounding structures. Lateral cephalogram was taken which revealed proclined maxillary incisors with increased overjet. Impedance Audiometry revealed hearing sensitivity of right ear to be in normal range whereas severe loss of hearing was noted in left ear. Consultation with the Ophthalmologist was done, who suggested normal eye morphology with no abnormality. Ultrasonography of abdomen was done which revealed no abnormality.

Correlating the history, clinical findings and investigative findings a final diagnosis of Goldenhar Syndrome was given.

The patient was then referred to the Department of Orthodontics for his orthodontic treatment, where orthodontic brackets were fixed on his maxillary teeth with nickel-titanium wire [Figure 6].

DISCUSSION

Most of the patients reported within the OAV spectrum, a term proposed by Gorlin et al. (1990), are sporadic.[25] As similar to previously reported cases, our patient had marked right facial hypoplasia with the chin slightly deviated to the affected side.
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.[25] Congenital ophthalmoplegia associated with Goldenhar Syndrome was reported in only one instance, concerning two patients with paralysis of one or more extraocular eye movements on neurologic examination. A third patient presented with unilateral agenesis of trochlear and abducent nerves and corresponding brain stem nuclei demonstrated at autopsy.[26] Brain stem involvement detected by magnetic resonance imaging in patients with Goldenhar Syndrome has been also reported; however, the stem lesions are very severe, resulting in a poor general prognosis.[27]

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.[28]

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 corner of the mouth and the tragus.[29]

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.[30]

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. The lack of development of the jaws can cause breathing problems, as well as dental malocclusion which requires multidisciplinary approach.[5] Timing of the reconstruction plays an important role in the treatment. Primary reconstruction typically consists of a cleft repair, corrections of colobomas and ear deformities and removal of dermoids and preauricular tags.[31] 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.[23]

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.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.
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