

Goldenhar Syndrome in a Young Girl

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ABSTRACT

Goldenhar or oculo-auriculo-vertebral (OAV) syndrome is a congenital disorder that is usually apparent at birth. Sometimes, its diagnosis can be delayed and it is first diagnosed in adolescents or adults. We present a case of an 18-year girl who presented with chief complaint of multiple carious teeth and was found to have abnormal facies. On examination, it was found that the patient had asymmetry of face due to hypoplastic maxilla and mandible on the left side. Patient also had presence of ocular dermoid in the left eye, pre-auricular tags, scoliotic posture, ankyloglossia and fissured tongue, which were in accordance with OAV syndrome. This case report unveils the need for an early diagnosis and prompt treatment of the anomalies found in this syndrome.

Key Words: *Oculo-auriculo-vertebral syndrome, Goldenhar syndrome, Hemifacial microsomia, Ocular dermoid, Scoliosis.*

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INTRODUCTION

Goldenhar syndrome is a rare genetic disorder first documented in 1952 by Maurice Goldenhar, an ophthalmologist and general practitioner.¹ It is also known as oculo-auriculo-vertebral dysplasia, as it arises from the defect in the development of first and second branchial arches of the 1st pharyngeal pouch and primordia of temporal bone.² It is a developmental disorder with a prevalence of 1:3,500 to 1:5,600 live births with male to female ratio of 3:2.³ The occurrence of this syndrome can be sporadic; but cases with autosomal dominant and recessive inheritance have been reported. There are a diverse amount of oral and systemic manifestations which occur due to this syndrome and the severity of the abnormalities and symptoms varies from individual to individual.⁴

Herein, we describe a case of Goldenhar syndrome in an 18-year girl who initially presented with the chief complaint of multiple carious teeth and was later diagnosed with the constellation of this syndrome.

CASE REPORT

An 18-year girl reported to the Department of Oral Medicine And Radiology with the chief complaint of multiple carious teeth with a request for the restoration of the same. History of the patient revealed that she was third among the four children in the family. At birth, she

was found to have a redundant ear in the pre-auricular region of left ear and presence of a growth in the left eye. Her milestones were normal throughout the growth period. The patient, however, had difficulty in speech. No history of consanguinity nor any abnormalities were noted in parents. No history of any physical abnormality was elicited in any of the siblings.

Examination of the patient revealed asymmetry of the face due to hypoplastic maxilla and mandible of the left side (Figure 1a). Pre-auricular tags were noted on the left side of the face (Figure 1b). The patient had hypertelorism with epibulbar dermoid in the left eye and mild loss of vision with respect to the same eye (Figure 1a). Patient's posture appeared to be scoliotic, and she also had flat feet (Figure 2). Intraoral examination revealed multiple carious teeth, ankyloglossia and fissured tongue (Figure 3a & b). Based on the above findings, the patient was diagnosed with oculo-auriculo-vertebral (OAV) syndrome with hemifacial microsomia, also called Goldenhar syndrome. The patient was then subjected to

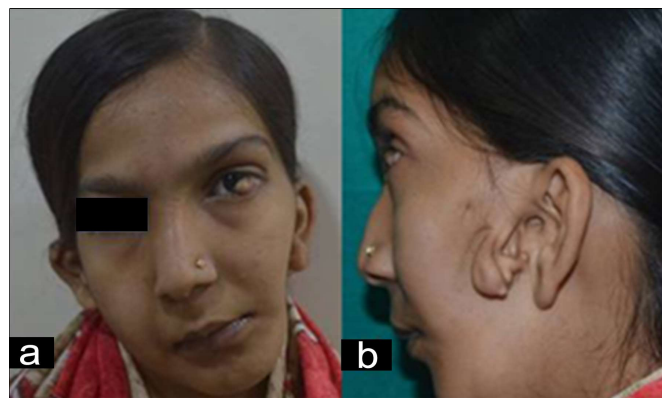


Figure 1: (a) Frontal profile of the patient showing epibulbar dermoid in the left eye. (b) Left ear showing presence of tissue tags (redundant ear).

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Figure 2: Patient presenting a scoliotic appearance on standing erect.

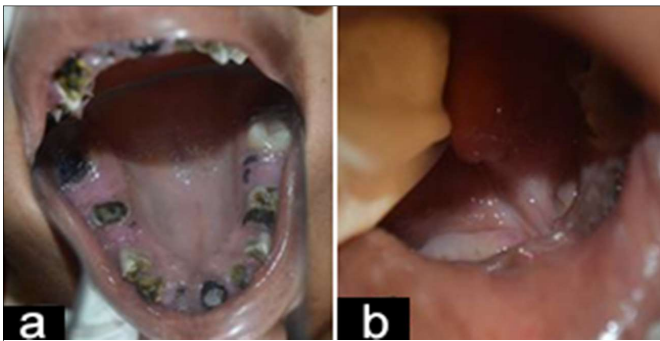


Figure 3: (a) Clinical intraoral image showing multiple carious teeth, suggestive of rampant caries. (b) High lingual frenum attachment (tongue tie).

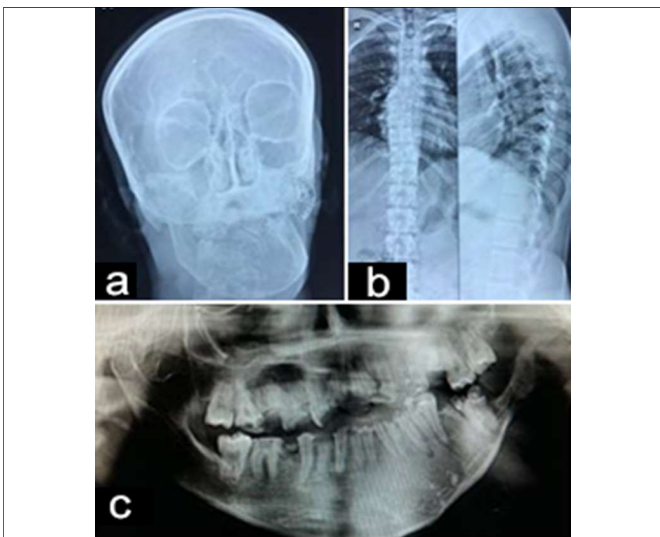


Figure 4: (a) Skull showing asymmetry of the face. (b) AP spine (black arrow) showing scoliotic spine. (c) Opg showing multiple carious teeth and hypoplastic mandible on the left side with a short ramus and hypoplastic condyle and coronoid.

radiographic evaluation. Skull radiograph revealed a left-sided mandibular deviation, suggestive of hypoplasia. Hypoplastic mandible and maxilla were further confirmed on the orthopantomogram (OPG) (Figure 4a & c). Spine radiograph revealed slight curvature of the spine to the right side (Figure 4b). Audiometric examination of the patient revealed mild hearing loss in the left ear. The patient was further subjected to karyotyping, which showed presence of translocation between chromosome 13q10 and 14q10. Thus, looking at the clinical findings, the radiographic features and karyotyping results, the patient was diagnosed with Goldenhar or OAV syndrome.

DISCUSSION

Goldenhar syndrome is a diverse and poorly understood disorder of unknown etiology. It shows many changes in the facial appearance as well as systemic changes. The patient exhibit a wide range of anomalies, such as dentofacial abnormalities with unilateral facial hypoplasia, prominent forehead, hypoplasia of the zygomatic area, maxillary and mandibular hypoplasia, lateral facial cleft involving palate, tongue cleft, posteriorly angulated ears, and bilateral pre-auricular tags. Ocular abnormalities include epibulbar dermoid (unilateral or bilateral), hypertelorism, and bilateral epicanthal folds. Vertebral column anomalies include synostosis, hemivertebrae, fused vertebral kyphoscoliosis, and missing ribs.^{5,6} Other anomalies include anomalies in the extremities, congenital heart disease, growth retardation, ectopic kidneys, and retardation of mental development.⁷ With these anomalies, no agreeable clinical diagnostic criteria have been reported in the literature. The presence of auricular abnormalities, mandibular hypoplasia, and spinal abnormalities can be put together to consider it as a case of OAV spectrum. Literature shows that most of the patients with OAV spectrum show hypoplasia of the right side which is in the ratio of 3:2. However, the present patient had a left sided deficiency with hypoplasia of both the maxilla and the mandible.

The causative factor of this spectrum still remains unknown; however, Gorlin and Pindborg, suggested that some abnormal embryological process involved the mesoblasts which affected the branchial and vertebral systems, thereby resulting in the syndrome.⁸ Poswillo suggested that hypertension in the mother or intake of anticoagulants at the time of gestation can cause hematoma in the ear and jaw region of the fetus; thus leading to improper development of the branchial arch, hence the defect.⁹ Certain chromosomal abnormalities are associated with this syndrome, namely, del (5p), monosomy 6q, trisomy 7 mosaicism, trisomy 9 mosaicism, and translocation between 13q10 and 14q10.¹⁰ A cytogenetic analysis of this patient revealed translocation between chromosome 13q10 and 14q10.

Treatment of Goldenhar syndrome varies greatly depending on the needs of the individual. In some mild

cases, no treatment is needed. Children may need to work with a hearing specialist or speech therapist for hearing issues, or may need a hearing aid. If there are vision problems, corrective surgery or glasses may be needed. Children with an intellectual disability may need to work with education specialists. Surgical removal of pre-auricular tissue tags and epibulbar dermoid are needed. Then correction of intraoral deformities should be undertaken.

Role of pedodontist is significant to ensure optimum oral healthcare for such syndromic patients from birth till adolescence, since often they have complex unmet dental needs. Pediatric dentists and pediatricians should work in collaboration to promote treatment of the affected children. However, efforts should be made by the medical and dental community to diagnose and manage this condition at the earliest, lessening the emotional, physical and financial burden of living for these special children.

CONFLICT OF INTEREST:

Authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

NK: Contributed to the conception and design of the work (evaluation of the case, detailed history taking and investigation and reporting the investigations).

SG, RM: Revised and critically analysed of the content.

MW: Finally approved the version.

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