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Goldenhar Syndrome: A Rare Case Report

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Abstract

Goldenhar syndrome is a hereditary condition which is characterized by preauricular appendages, fistulas, and epibulbar dermoids. It not only involves the facial structures, but also includes renal, genitourinary, cardiac, and skeletal anomalies. This is a rare congenital anomaly. Here, we report a case of Goldenhar syndrome.

Keywords: Goldenhar, syndrome, Preauricular skin tags, Epibular dermoid

Introduction

Goldenhar syndrome is a congenital defect characterized by constellation of malformations classically involving face,eyes and ears. It is also known as oculoauriculo-vertebral dysplasia.[1] It was first observed and recorded by Carl Ferdinand von Arlt.[2] Maurice Goldenhar was the first to describe the syndrome in detail and thus the condition was called Goldenhar syndrome.[3]

The exact etiology of this rare condition is not fully understood, but various authors hypothesis have been proposed and explained the etiopathogenesis.[4] Gorlin and Pindborg 1964,suggested that some abnormal process affects the mesoblasts embryologically the branchial and vertebral systems.Also there are familial cases in successive generation having history of consanguineous marriage that requires consideration of autosomal recessive, dominant or multifactorial inheritance. [5]

The incidence has been reported 1 in 3500 live births, with a male predominance (3:2). It is unilateral in 85% of cases with the right side more frequently affected than the left with a ratio of 3:2 and bilateral in 33% cases.[6]

The characteristic clinical findings includes, Epibulbar dermoid or lipodermoid (mostly bilateral), colobomas of the upper eyelid, iris, choroidea and retina, or other eye anomalies such as microphthalmia and anophthalmia ,

Preauricular skin tags or blind fistulas; microtia, or other external ear malformations, middle and internal ear anomalies,Unilateral facial hypoplasia, prominent forehead, hypoplasia of the zygomatic area, maxillary and mandibular hypoplasia,Unilateral macrostomia .

These features are sometimes associated with various malformations such as cleft lip and palate, tongue cleft, rib anomalies, anomalies of extremities, congenital heart disease, and mental retardation.[3]

Here we report a rare case of a child diagnosed with Goldenhar syndrome.

Case Report

A 6 years old female patient was referred from Department of Opthalmology ,HSK Hospital ,Bagalkot to the Department of Pediatric and Preventive Dentistry, P.M.N.M Dental college and Hospital Bagalkot, for oral examination and checkup.

On general examination the patient was conscious and cooperative; there were no signs of pallor, clubbing, cyanosis, icterus and lymphadenopathy.

On extraoral examination, facial asymmetry was present with deviation of angle of mouth toward the left side. There was flattening of left side of face with loss of left malar prominence. Facial profile was concave due to midface deficiency. Microtia along with preauricular tags was present on the left side [Fig 1,2,3,4].

On Intraoral examination revealed High arched palate, class 1 molar relation with respect to the right side, on the left side, teeth were out of occlusion. Deep occlusal caries with respect to 54,64,74,75 and proximal caries with respect to 51,52 ,61,62. [Fig 5,6,7]

Further investigations were not performed as the parents were not willing to take the treatment.

Figure1: Frontal View –Facial Asymmetry

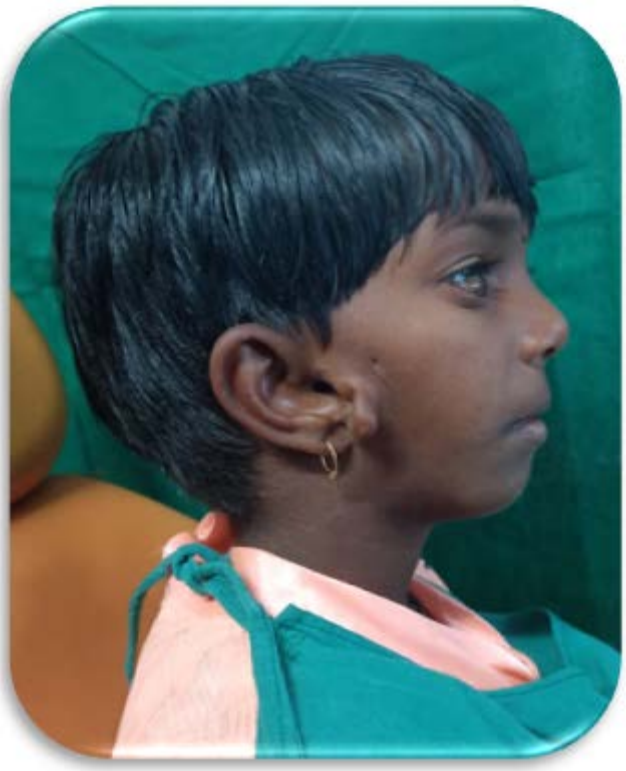

Figure 2: Lateral View –Mid Face Hypoplasia

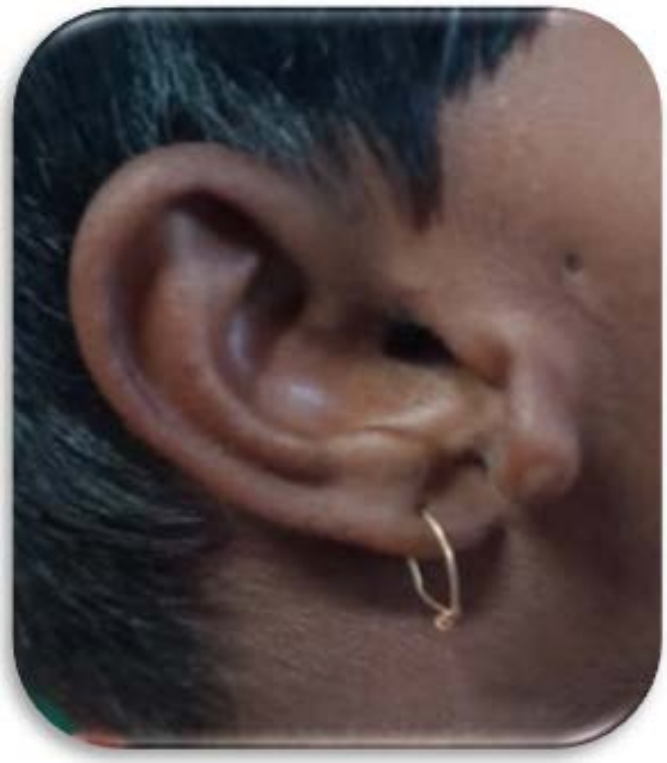

Figure 3: Ear Malformation -Preauricular Tag

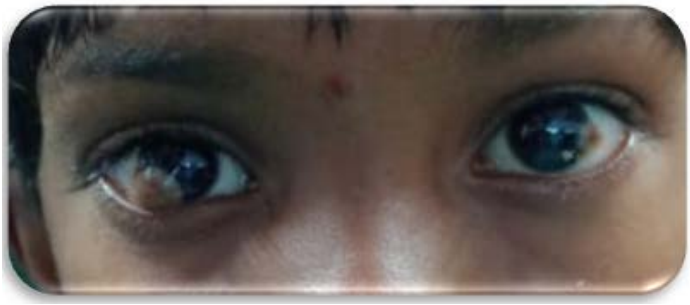

Figure 4: Eye Abnormality – Epibulbar Dermoid Tumor

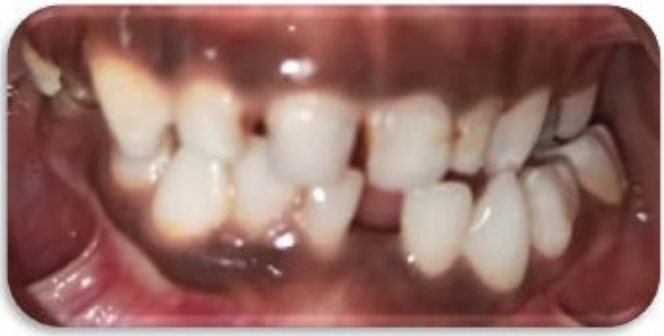

Figure 5: occlusal view

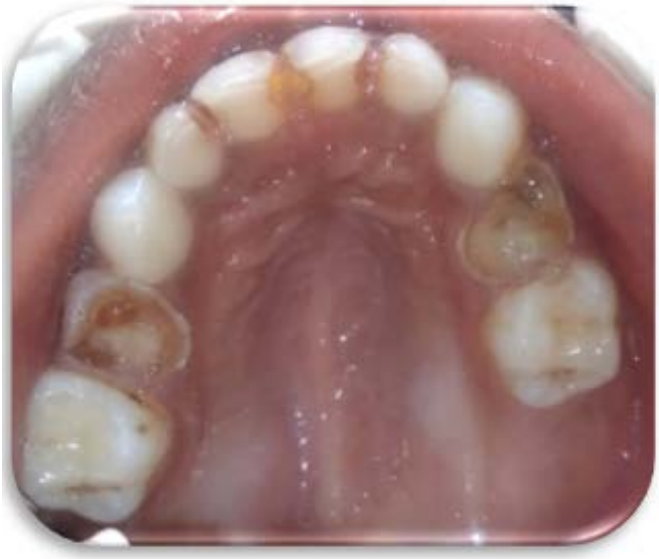

Figure 6: Maxillary Arch

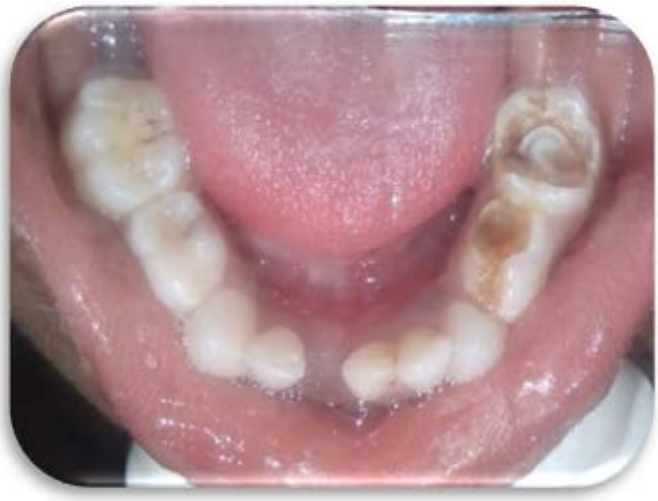

Figure 7: Mandibular Arch

Discussion

Goldenhar syndrome is characterized by a triad of accessory tragi, mandibular hypoplasia, and ocular dermoids.[7]

There are certain clinical findings which are typical to Goldenhar syndrome are in Eyes, Epibulbar dermoids occurs in about 35% of cases. These may be unilateral or bilateral. They appear as solid yellowish or pinkish white ovoid masses which vary in size from that of a pinhead to 8–10 mm in diameter. They usually occur at the infer temporal quadrant at the limbus. The surface is usually smooth and frequently has fine hairs. They can occur at

any location on the globe or in the orbit and can be dermoid or dermis like or complex. [8,9]

In Ears, Anotia to an ill-defined mass of tissue that is displaced anteriorly and inferiorly to a mildly dysmorphic ear may be seen. Supernumerary ear tags, unilateral and bilateral preauricular tags of skin and cartilage, along with blind fistulas and sinuses, are extremely common.[3,9]

In Face, Marked facial asymmetry with unilateral macrostomia may be seen due to displacement and abnormality of the pinna and other underlying abnormalities of the skeleton. Aplasia or hypoplasia of the mandibular ramus and condyle may be seen. This in turn may lead to reduction in the size of maxillary, temporal, and malar bones. Hypoplasia of the zygomatic area may also be seen. [3,9]

In Skeletal alterations, Defects of the skull such as cranium bifidum, microcephaly, dolichocephaly, and plagiocephaly have been noted. Spina bifida, hemivertebrae, butterfly, fused, and hypoplastic vertebrae like vertebral anomalies will be seen. [3,9]

Oral manifestations include cleft lip and palate,tongue cleft, unilateral tongue hypoplasia, a highly arched palate, hypoplasia of the maxillary and mandibular arches, micrognathia, gingival hypertrophy, supernumerary teeth, enamel and dentin malformations, and delayed tooth development are common.[3,9]

The present case had the features such as facial asymmetry, epibulbar dermoids affecting both the eyes, preauricular tags and hypoplasia of mandible and high arched palate.

Various diagnostic aids such as ultrasonography, computed tomography and radiographic analysis should be done to rule out the syndrome. Ultrasonography is done during pregnancy and can rule out severe hypoplasia of mandible, severe abnormality of the auricle and cleft lip and/or cleft palate. Computed tomography is done for the evaluation of hearing to see the middle ear bones and to rule out skeletal findings radiographic analysis can be carried out. [10]

The treatment of the patient depends on the age and systemic condition. Management is usually cosmetic. Reconstructions can be done using rib bone grafts in patients with hypoplastic mandible. Bone distraction and osteogenesis may be used to lengthen the underdeveloped maxilla. In cases with cleft lip and palate, surgical corrections can be done followed by orthodontic correction on completion of jaw growth. In patients with malformed or deformed external ear, reconstructive surgeries may be performed at an age of 6–8 years. Epibulbar dermoids present in the eyes may be surgically excised. Plastic surgery can be performed for structural deformities of the eyes and ear. The prognosis for this condition is good in patients with no systemic complications. [5,8]

Conclusion

Severe cases of Goldenhar syndrome can affect the routine and social life of the patient. Early detection can help avoid complications at a later stage of life as they will have an increased risk for psychosocial difficulties. The Role of a Pedodontist is significant to ensure optimum oral health care for such syndromic patients from birth till adolescence since often they have complex unmet dental needs. Pediatric dentists and Pediatricians should work in collaboration with sound referral services for prompt treatment of the affected children. 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 in these special children.

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