CASE REPORT

GOLDENHAR SYNDROME WITH MANDIBULAR PARTIAL AGENESIS -A CASE REPORT

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ABSTRACT

Oculo-auriculo-vertebral syndrome or Goldenhar syndrome is a rare congenital condition characterized by abnormal development of the eye, ear and spine. First documented in 1952 by Maurice Goldenhar, an ophthalmologist and general practitioner. Children are born with partially formed or totally absent ears, benign growths of the eye, and spinal deformities such as scoliosis. In most cases, the deformity only affects one side of the body. Here, we present a case report of Goldenhar syndrome in a 4-year-old male patient with partial agenesis of mandibleand symptoms affecting multiple organ systems

KEY WORDS

Oculo-auriculo-vertebral syndrome, Goldenhar syndrome, Partial agenesis of mandible

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INTRODUCTION

Goldenhar syndrome, also known as oculo-auriculo-vertebral syndrome (OAVS), is a rare congenital condition arising from defects in the first and second branchial arches. It was first described by Dr. Maurice Goldenhar in 1952[1]. The incidence of Goldenhar syndrome has been reported to be 1 in 35,000 to 1 in 56,000, with a male-to-female ratio of 3:2[1]. It consists of a triad of mandibular hypoplasia accompanied by facial asymmetry, oculo-auricular malformations, and vertebral abnormalities. Gorlin et al. found an association between this syndrome and vertebral anomalies and named it oculoauriculovertebral dysplasia[2]. In 85% of cases, it is unilateral, and in 50% it can be associated with other systemic features such as cardiac, vertebral, and central nervous system malformations[3].

The classical manifestations include the absence or incomplete development of the auricles, in conjunction with middle or inner ear abnormalities, which ultimately result in hearing loss. In addition, common features include mandibular or maxillary hypoplasia and hemifacial microsomia. Other recurring characteristics include epibulbardermoids, which may impair visual acuity, and congenital scoliosis, which affects about 50% of cases[1].

The exact etiology of this condition remains unknown. Chromosomal abnormalities, aberrant neural crest cells, and environmental factors-such as drug use during pregnancy, including cocaine, thalidomide, and retinoic acid-as well as maternal alcohol consumption, have been linked to the development of the disease.[4] Most cases are sporadic, but autosomal dominant inheritance has also been reported. There is a frequently observed variability of clinical expression within families.

Here, we present a case report of Goldenhar syndrome in a 4-year-old male patient with symptoms affecting multiple organ systems. Although diagnosis is mainly clinical, radiographic investigations help to support the clinical diagnosis. The case's phenotypic presentation is both typical and comprehensive, making it exceptionally rare. Many classical signs of the syndrome were present in the patient, along with a few rare features. The

Fig 1: Frontal view showing facial asymmetry involving left side face

Fig 2: Ocular defect showing lipodermoid cyst

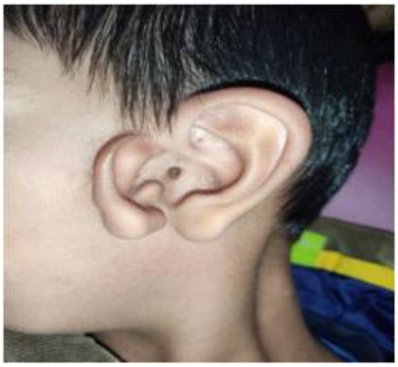

Fig 5: preauricular tag, resembling an extra auricle

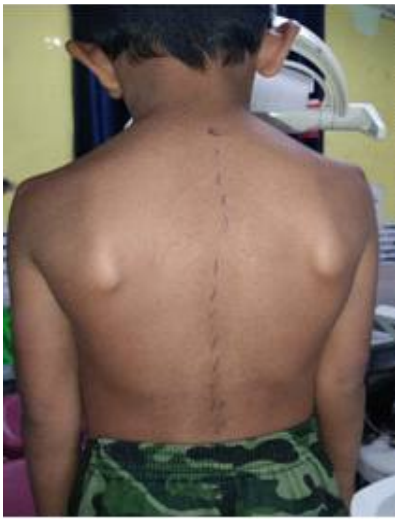

Fig 4: Mild scoliosis

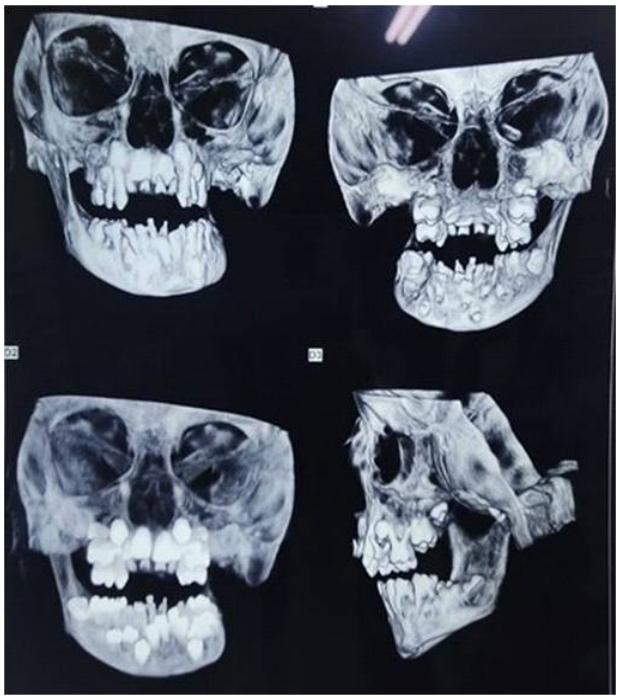

Fig 5: CBCT showing absence of the ascending ramus, condyle, and coronoid process on the left side

various aspects of this rare disease are discussed, with emphasis on timely diagnosis and a multidisciplinary approach to management.

CASE REPORT

A 4-year-old patient was referred from NRS Medical College to the Department of Pediatric Dentistry at Dr. R. Ahmed Dental College and Hospital with complaints of multiple decayed teeth. On initial observation, a pronounced asymmetry involving the left side of the face was noted. History revealed that the patient had experienced hearing difficulties in the left ear since childhood.

On extraoral examination, facial asymmetry with hypoplasia of the left side of the face was observed, involving the eyes, ears, and jaws (Fig 1). The eyes showed ptosis of the eyelids and a lipodermoid cyst, with normal vision (Fig 2). The ears exhibited a preauricular tag, resembling an extra auricle, along with hearing impairment (Fig 3). Hypoplasia of the facial bones, especially the mandible, maxilla, and zygoma, caused typical disfigurement of the affected side of the face. The mandible deviated toward the affected side upon mouth opening. On general examination, mild scoliosis also noted (Fig 4).

On intraoral examination, the patient exhibited

normal dentition with the absence of the pterygomandibular raphe on the affected side. Radiographic evaluation revealed normal dentition; however, there was typical agenesis or absence of the ascending ramus, condyle, and coronoid process on the left side (Fig 5). Additionally, disfigurement of the zygomatic bone was observed. The prognosis depends on the presenting malformations and their association with systemic defects. A multidisciplinary evaluation is essential to reach this challenging clinical diagnosis.

DISCUSSION

Goldenhar syndrome is a congenital disorder known as oculo-auricular-vertebral syndrome (OAV) with classic triad syndrome, namely mandibular hypoplasia (facial asymmetry), ear malformation (microtia/anotia and preauricular fistula) and/or eyes (bulbar dermoid cyst, microphthalmia), as well as vertebral anomalies[7].

The characteristic combination of external ear anomalies, ocular defect, vertebral defect and ipsilateral facial underdevelopment is the hallmark of Goldenhar syndrome as seen in our case. The condition is mostly unilateral in occurrence in 85% cases, with the right side more frequently affected than the left with a ratio of 3:2. In our case, unilateral facial involvement of left side was seen which made it a more rare category.

The patient's history of congenital hearing loss and the presence of an accessory auricle (preauricular tag) align with known auricular anomalies seen in Goldenhar syndrome. The lipodermoid cyst and ptosis are also common ocular findings associated with this condition, though vision remains unaffected, as observed in this case. Mandibular deviation upon mouth opening, due to hypoplasia and agenesis of the ramus, condyle, and coronoid process, further supports the diagnosis. Radiographic findings confirmed the agenesis of critical mandibular structures and deformity of the zygomatic bone. The absence of the pterygomandibular raphe on the affected side is a rare intraoral finding and may have implications for future oral function and surgical planning. The general examination also revealed mild scoliosis, which points toward possible vertebral involvement-an important systemic association found in nearly half of cases. This further underlines the necessity for comprehensive systemic evaluation, including cardiac, renal, and central nervous system assessments. Prenatal diagnosis is possible with considerable accuracy with ultrasound which may detect obvious defects[1]

Management of OAVS is complex and typically requires a multidisciplinary approach, of ophthalmology, ENT, otolaryngology, odontology, radiology and neurology contributes to the diagnosis and treatment of this case[9]. Early intervention can significantly improve function and aesthetics, and treatment planning must be individualized based on the patient's age, severity of anomalies, and associated systemic conditions. In cases of mandibular hypoplasia, reconstruction can be done with rib grafts, and underdeveloped maxilla can be lengthened by bone distraction device. Structural abnormalities of the eyes and ears are typically managed with reconstructive or plastic surgical interventions.

The prognosis varies widely and is largely dependent on the extent of craniofacial malformation and the presence of systemic defects. While this case does not currently demonstrate cardiac or CNS involvement, the presence of skeletal abnormalities like scoliosis necessitates ongoing monitoring and potential orthopedic management.The successful management of OAVS hinges on early diagnosis and a coordinated, multidisciplinary approach that addresses both functional deficits and aesthetic concerns. Individualized care plans and ongoing follow-up are essential for improving the quality of life and long-term outcomes for affected individuals.

CONCLUSION

Pediatric dentists play a central role in the early detection and ongoing care of such patients, working collaboratively with other healthcare professionals to ensure optimal functional and aesthetic outcomes. Given the variability in clinical presentation and systemic involvement, individualized treatment and long-term follow-up are essential. Early diagnosis and comprehensive treatment not only improve the patient's physical appearance and function but also significantly reduce the emotional, psychological, and financial burden on the family.

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