









GOLDENHAR SYNDROME –A CASE REPORT

ABSTRACT

Goldenhar Syndrome also called as facio-auriculo-vertebral dysplasia, is a rare syndrome developing from first and second pharyngeal arches during blastogenesis. It was described by Maurice Goldenhar in 1952. It is characterised by presence of epibulbar dermoids, ear malformations, vertebral anomalies, unilateral facial hypoplasia, and sometimes internal systemic complications. As the molecular basis for Goldenhar Syndrome is unclear, early detection and screening for complications would help the patients to have a normal life. This is a case report of a case of 15 year old male patient with Goldenhar syndrome with epibulbar dermoids, microtia, syndactyly and micrognathia.

Key words: Goldenhar Syndrome, dermoids, microtia, mandible

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INTRODUCTION

Goldenhar syndrome (GHS) was first seen by Canton in 1861 and Von Arlt, in 1881 but however it went unnoticed.^{1,2} Maurice Goldenhar, the Swiss ophthalmologist in 1952 recorded three new cases in addition to the sixteen cases which were previously recorded and later first precisely described the syndrome in detail. Thus, it came to be known as Goldenhar syndrome.³ It consisted of preauricular appendages, fistulas, and epibulbar dermoids and a variant of hemifacial microsomia which used to affect aural, oral, and mandibular development having both unilateral and bilateral involvement.^{1,4} In 1963, vertebral anomalies were also included by Gorlin et al as one of the manifestations of this syndrome and thus suggested the name oculoauriculo vertebral (OAV) dysplasia.^{4,5} Smith in 1978, used the term facio-auriculo-vertebral sequence to include both Goldenhar syndrome and Hemifacial microsomia.⁴ It is also known as Goldenhar- Gorlin syndrome, facio-auriculo-vertebral dysplasia, unilateral craniofacial microsomia, first arch syndrome, first and second branchial arch syndrome, lateral facial dysplasia, velo-cardio-facial syndrome, otomandibular dysostosis, unilateral mandibulo facial dysostosis, unilateral intrauterine facial necrosis, auriculo-branchiogenic dysplasia, facio-auriculo vertebral malformation complex.^{1,4} In this article we report a case of 15 year old male patient with GHS.

CASE REPORT

A 15 year old male patient reported to our dental clinic with a complaint of multiple decayed teeth since 6 months as shown in Figure 1. The patient gave a medical history of reduced hearing, delayed speech since birth, diminution of vision since 4 years. The family history revealed that the mother has syndactyly in both upper and lower limbs. On examination, the patient's head was dolicocephalic, had a convex facial profile and incompetent lips as seen in Figure 1.



Figure 1. Absence of gross facial asymmetry

The patient had solitary round soft-tissue mass was seen in the right eye indicative of epibulbar dermoids seen in Figure 2, small prominent right ear or microtia seen in Figure 3 and syndactyly of the lower limbs as seen in Figure 4.



Figure 2. Epibulbar dermoids



Figure 3. Microtia with hypoplastic mandible



Figure 4. Syndactyly of the limbs

On intraoral examination, hypoplastic mandible, bimaxillary protrusion with multiple decayed teeth were present.

Based on the history and examination, a provisional diagnosis of Goldenhar Syndrome (GHS) was given, and Treacher-Collins syndrome and Townes–Brocks syndrome was considered as differential diagnosis.

On radiological investigation, the orthopantomogram, revealed bilateral absence of mandibular condyle, coronoid process in Figure 5 and lateral cephalogram revealed a hypoplastic mandible with bimaxillary protrusion in Figure 6.



Figure 5. OPG reveals bilateral absence of condyle and coronoid and presence of multiple decayed teeth.



Figure 6. Lateral cephalogram reveals hypoplastic mandible

DISCUSSION

The incidence of GHS is rare, with a male predominance.⁶ GHS is an inherited condition, which has a multifactorial etiology along with various nutritional and environmental factors that can cause disturbances of blastogenesis.⁷ Family history shows autosomal dominant or recessive inheritance.⁸ Gorlin and Pindborg in 1964 suggested that mesoblasts are affected by some abnormal process embryologically which, in turn, affects the branchial and vertebral systems, thereby resulting in the syndrome. It has also been suggested that there is a defect in branchial arch development late in the first trimester.^{8, 10} Hereditary pattern was thought to be the causative agent by Krause. In the year 1971, Jong Bloet suggested that this condition might be a result of fertilization of an over ripe ovum. Goldenhar syndrome may be a sporadic event that occurs early in embryogenesis as stated by Baum and Feingold which may be explained by reduced penetrance, somatic mosaicism, or epigenetic

change.⁹ Familial cases in successive generations having a history of consanguineous marriages have also been reported. Gomez et al. in 1984 speculated about the role of radiologic intervention (cholecystography practiced between the fourth and sixth weeks of pregnancy) as an etiologic mark of the syndrome.⁴ The ingestion of some drugs such as cocaine, thalidomide, retinoic acid and tamoxifen by the mother and maternal diabetes were also related to the development of the disease.⁸

In a classic Goldenhar syndrome patient shows characteristic ocular, auricular facial and vertebral features as given in Table 1.⁴

Table 1. Characteristics of Goldenhar Syndrome

Ocular manifestations	Epibulbar dermoid or lipodermoid (mostly bilateral); colobomas of the upper eyelid, iris, chorioidea, and retina, or other eye anomalies (e.g. microphthalmia, anophthalmia, cataract, astigmatism, antimongoloid obliquity of palpebral fissures, and blepharophimosis)
Auricular manifestations	Preauricular skin tags or blind fistulas; microtia, or other external ear malformations (dysplasias, asymmetries, aplasias, and atresias of the external meatus); middle and internal ear anomalies.
Facial and oral manifestations	Unilateral facial hypoplasia, prominent forehead, hypoplasia of the zygomatic area, and maxillary and mandibular hypoplasia. Unilateral macrostomia (lateral facial cleft). Principal deformities of the Goldenhar syndrome are often combined with various malformations, such as: Cleft lip and/or palate, tongue cleft, and parotid gland aplasia.
Vertebral anomalies	Vertebral column anomalies (atlas occipitalization, synostosis, hemivertebrae, fused vertebrae, scoliosis, and bifid spine). Rib anomalies and anomalies of the extremities.
Miscellaneous anomalies	Congenital heart disease (ventricular septal defects), anomalies of the urogenital and gastrointestinal system (ectopic kidneys, ureteropelvic junction obstruction, and imperforate anus), anomalies of the central nervous system (occipital encephalocele), and anomalies of the larynx and lungs (tracheoesophageal fistula, esophageal atresia). Complex retardation of mental development.

In this case of GHS, unilateral epibulbar dermoids and microtia were present. The patient presented with hearing, speech deficiency and diminution of vision. There was bilateral absence of the condylar, coronoid process with hypoplastic mandible, bimaxillary protrusion, syndactyly of the limbs were present. However, there were

absence of other clinical features such as facial asymmetry, absence of spinal anomalies or systemic abnormalities.

OAV represents the mildest form of the disorder, GHS presents as most severe form and hemifacial microstomia appears to be an intermediate form. 10-30% have bilateral manifestations.¹¹ The phenotypic findings of this syndrome are variable due to heterogenous etiology. Preauricular skin tags seen in 90% of cases, microtia in 52%, hemifacial microstomia in 77%, epibulbar dermoid in 39% of cases. Vertebral anomalies were noted in 7% of cases. Cardiac manifestation are found in 39% of cases, while genitourinary anomaly was noted in 23% and various central nervous system anomalies are seen in 47% of cases.⁸ The present case may represent a mild form of GHS.

There is an overall consensus that the diagnosis of this disease must not be only based upon radiologic or laboratory results but the clinical aspect as well.⁸ Most authors consider the presence of anomalies of the ear (microtia) lower set than on the contralateral side and of appendices on the ear necessary for diagnosis.¹¹ The ossicles of the ear are derived from Meckel's cartilage the precursor of the mandible. Patients who have GHS routinely have hearing deficiencies on the affected side due to under development of the osseous components of the auditory system and a diminished or absent external auditory meatus.¹²

Radiographic examination of zygomatic bones shows a macroscopic deficiency and developmental symmetry. There is also a possibility of agenesis of these bones with lack of fusion of the zygomatic arch and agenesis of the palatine bones. Palatal cleft may be observed radiographically. Ophthalmologic and otorhinolaryngologic examination are also important for the final diagnosis.¹¹

Syndromes considered for differential diagnosis for GHS are Treacher-Collins syndrome and Townes-Brocks syndrome. The presence of facial asymmetry and far less hypoplasia of the malar bones in GHS are important features to differentiate it from Treacher-Collins syndrome.

The Treacher-Collins syndrome affected patients presented downward and mandibular hypoplasia, partial absence of the lower eyelid cilia, and abnormalities of the ears. Townes–Brocks syndrome shows additional thumb anomalies, anal defects, and renal anomalies which are not seen in GHS.

The treatment of the patients with GHS depends on the age and presence of systemic complications. 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 followed by orthodontic correction on completion of jaw growth. In patients with malformed or deformed external ear, reconstructive surgeries maybe performed at an age of 6–8 years. Epibulbar dermoids present in the eyes may be surgically excised. The prognosis for this condition is good in patients with no systemic complications.^{12,13}

CONCLUSIONS

In summary, the molecular basis of GHS is still is unavailable. Patients with GHS can have multiple congenital anomalies, and they need particular attention to internal abnormalities and therefore should be duly diagnosed and screened for presence of any kind of systemic complications.^{9,10} These patients may face difficulties in their regular social activities due to their hearing or speech deficiencies. The families of such patients should give moral support and counselling for accepting their kins and relatives with such conditions.

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None

CONFLICTS OF INTEREST

None

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