Atypical Goldenhar Syndrome: A Case Report

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A patient with atypical Goldenhar syndrome is reported. The characteristic features similar to the other reported cases are facial asymmetry, multiple skin tags, limbal dermoids and posteriorly angulated ear. The atypical intraoral feature is unilateral presence of multiple complex odontomes. The hypotrophy of left hemisphere with localized calcification in the occipital and parietal regions of brain is also an additional finding in this patient.

Key words: *Goldenhar syndrome, oculoauriculovertebral spectrum, dermoids, odontomes.*
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INTRODUCTION

The first observation of the oculoauriculovertebral dysplasia was reported in 1881 by Arlt F.[1] In 1952, Goldenhar collected and classified the clinical features and named the malformation complex which is known as Goldenhar syndrome.[2] Goldenhar syndrome is a sporadic and non-hereditary syndrome characterized by a triad of anomalies consisting of epibulbar dermoids, accessory auricular appendages and pretragal fistula. In 1964, Gorlin and Pindburg described hemifacial microsomia as another facial malformation.[3] However, the clinical characteristics of hemifacial microsomia and Goldenhar syndrome are similar to that of oculoauriculovertebral dysplasia. It has also been suggested that these entities represent a single disorder with great variability of expression and an isolated ear malformation may represent the mildest expression of the disorder.[4-10] In 1990, Gorlin *et al.* proposed to lump several syndromes including facioauriculovertebral syndrome, hemifacial microsomia, otomandibular dysostosis, Goldenhar syndrome, the first branchial arch anomalies and the first and second branchial arch anomalies together and to use the term "Oculoauriculovertebral spectrum."[5] Recently, Tasse *et al.* has also proposed a new classification system for oculoauriculovertebral spectrum.[11] Gorlin estimated the incidence of hemifacial microsomia to be 1 in 56000.[5] Morrison *et al.* however estimated a minimum prevalence rate of oculoauriculovertebral dysplasia is of 1:45000 among Northern Ireland population.[12] The purpose of this article is to present a Goldenhar syndrome case with an atypical dental finding.

REVIEW OF LITERATURE

Goldenhar syndrome as described by the Gorlin in 1952 is characterized by a triad consisting of epibulbar dermoids, accessory auricular appendages and pretragal fistula.[2] These clinical features are often found in combination with other malformations. A number of cases have been reported in the literature with major involvement of eye, ear and vertebra. However, very few cases among them describe the dental anomalies of Goldenhar syndrome. A summary of the characteristic ocular, auricular, vertebral, systemic and dentofacial anomalies abstracted from the literature follows.

Ocular anomalies

Typical ocular anomaly is unilateral or bilateral epibulbar dermoids,[13-16] which are benign fibrous tumors on the edge of the cornea. The main concern about epibulbar dermoids is development of corneal astigmatism which may lead to amblyopia i.e. lazy eye syndrome. Other eye anomalies include coloboma of the upper eyelids,[17] coloboma of the iris and choroids,[18] ptosis,[13] small eye with notched upper lid,[19] hypertelorism,[19] short palpebral fissure,[20] hypoplasia of the supraorbital ridges,[16] bilateral epicanthal folds,[16] prominent eyes with cloudy corneae[16] and juvenile glaucoma.[21] Sabo[22] and Thomas[23] reported lipoma of the conjuctiva in Goldenhar syndrome patients. However, it was found that epibulbar dermoids or lipodermoids are not hereditary in man either alone or in combination with Goldenhar syndrome.[24]

Auricular anomalies

Auricular anomalies include mild ear malformations as preauricular skin tags and nodes,[4,14,15,20] preauricular dimples[20] to imperforated external auditory meatus,[25] deafness,[26] atresia of the external auditory canal,[15] anomalies in the size and shape of the external auricle[15] and even to anotia.[27] Another significant ear anomaly in Goldenhar syndrome patients is posteriorly angulated ears.[14-16,28] Kobrynsky *et al.* also reported hypoplastic left auricle with absence of external auditory meatus and bilateral preauricular pits.[16]

Vertebral and other skeletal anomalies

Vertebral skeletal anomalies include occipitalization of the atlas,[29] spina bifida,[29] spondylolisthesis,[29] kypho-scoliosis,[29] kyphosis,[30] hemivertebrae,[20,23,30] butterfly vertebrae,[20,30] equinovarus deformity[14] and unfused neural arches in the thoracic region.[23] The vertebral bony anomalies are common but usually tend to be overlooked.[31] Rib anomalies may include a missing rib or reduced number of ribs.[16,20,23,30] Limb anomalies include deep-set and hypoplastic nails on all limbs,[16] hypoplastic distal phalanges,[16] clinodactyly of the 5th

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fingers,[16] hypoplasia of the left acetabulum,[23] short stubby fingers with ulnar deviation,[16] bilateral radio-ulnar synostosis[23] and club feet with prominent heel.[16] In some patients skeletal anomaly may also include short neck with anterior webbing.[16]

Systemic anomalies

General appearance of the patients is usually normal. There may be growth retardation with marked hair in the body.[20] They may have torticollis,[32] upturned nose,[15] a flat nasal bridge with broad nasal root,[16] heminostrial,[32] deep creases on the feet[20] and asymmetry of the gluteal folds with possible ortolani click.[23] Severe respiratory distress especially during sleep due to intermittent upper airway obstruction secondary to narrow upper airway is also a common finding.[14,16] Sutphen *et al.* found tracheoesophageal fistula with or without esophageal atresia in patients with oculoauriculovertebral spectrum.[33] Handa and Upadhyay reported H type tracheoesophageal fistula in Goldenhar syndrome patients.[34] Cardiac anomaly may include isolated ventricular septal defects (VSD), atrial septal defect (ASD), pulmonary stenosis (PS), tetralogy of the Fallot and complex heart disease including left superior venacava, VSD, ASD, PS and pulmonary trunk hypoplasia.[12] Ryan, Finer and Ives reported systolic murmur in Goldenhar syndrome patients.[20] Stringer *et al.* reported portal vein cavernoma in Goldenhar syndrome patients.[35] Genital anomaly may include bilateral vescicoureteral reflux[28] and bilateral ureteropelvic junction stenosis.[19] Wilson reported bilateral hydronephrosis secondary to bilateral ureteropelvic junction stenosis in Goldenhar syndrome subjects.[19] Increased arterial distensibility and renovascular hypertension in Goldenhar syndrome patients has also been reported.[36] Cranial and brain defects may include hydrocephalus with cysts in the frontal cortex,[18] plagiocephaly[32] and ipsilateral cerebellar hypoplasia.[37] Recently Michel-Adde *et al.* reported neuroblastoma in a Goldenhar syndrome patient.[38] Bilateral inguinal hernias and two vessels umbilical cord can also be found in patients with Goldenhar syndrome.[14]

Dentofacial anomalies

Facial asymmetry or hemifacial microsomia is often present in patients with Goldenhar syndrome. Unilateral facial involvement is more common. Dentofacial anomaly may include cleft lip,[20,32] bilateral cleft lip and palate,[14,16,28] submucous cleft,[28] a crease over the lateral commissure of the mouth,[16] lateral oral cleft,[14] hypoplastic anterior part of the tongue,[20] cleft in the junction of anterior and posterior part of the tongue,[20] high arch palate,[15,20] hypoplasia of the maxillary and mandibular arches,[14,28] micrognathia,[16,20] facial paresis[14] and gingival hypertrophy.[16] Barker, Acaroglus and Soykan found congenital facial nerve palsy in a Goldenhar syndrome patient.[39] Recently hypoplasia and aplasia of the trigeminal nerve were also found in a Goldenhar syndrome patient.[40]

ETIOLOGY AND INHERITANCE

The cause of oculoauriculovertebral spectrum (Goldenhar syndrome) is heterogenous. The anomalies of the syndrome may be seen in aneuploid syndromes[41-45] or as Mendelian mutations.[14,46] Isolated case reports of familial occurrence have suggested as both autosomal dominant and autosomal recessive inheritance.[47-50] However, for the large majority of the cases multifactorial etiology have been postulated.[51]

Regenbogen *et al.*[52] and other authors[53,54] found that the mode of inheritance Goldenhar anomalies is autosomal dominant. The involvement of an eye may be less marked in the dominant form.[52] Ryan *et al.* observed the Goldenhar anomalies with different severity in monozygotic twins.[20] They concluded that Goldenhar anomaly is a result of fetal hemorrhage in the region of the first and second branchial arches at the time when the blood supply of these arches switches from the stapedial artery to the external carotid artery. Soltan and Holmes, however, suggested a link between genetic cause and vascular disruption.[55]

Many studies reported monozygotic twins discordant for Goldenhar anomalies and suggested heterogenecity.[14,15,56,57,58] Jongbloet suggested that sporadically occurrence of Goldenhar syndrome is the result of "overripeness ovopathy."[59] Kelberman *et al.* however performed a genome wide search for linkage in two families with features of hemifacial microsomia.[60] The data of the one family were suggested of linkage to a region of approximately 10.7cM on chromosome 14q32 with a maximum multipoint lod score of 3.00 between microsatellite markers D14S987 and D14S65. The linkage to this region was however not found in the second family suggesting genetic heterogenecity. Kelberman *et al.* however considered the goosecoid gene to be an excellent candidate gene for hemifacial microsomia.[60] Beverly and Kaye evaluated pedigree data in 97 proposition, of whom 44 had a family history of the same or similar anomaly and concluded that the pattern of occurrence in many families suggested multifactorial determination.[61]

CASE REPORT

A 9-year old North Indian male patient with diagnosis of Goldenhar syndrome was referred from the Department of Opthalmology to the Department of Dental Surgery, All India Institute of Medical Sciences, New Delhi for dental check-up. The patient was examined clinically and a thorough family and medical history was recorded.

The family pedigree is shown in Figure 1. The patient had one elder sister who was absolutely normal. None of the family members had any similar kind of anomaly. The patients past medical and dental history suggested that the patient had spontaneous vaginal delivery. The mother had no history of any drug ingestion or systemic disease during pregnancy. The patient had a history of repeated stomatitis and chest infections since birth. He had also a history of swelling in the left side of face when he was 1 year old and an epileptic attack two years back. Patient had limbal dermoids with bony deformity in the left eye for which he had undergone surgery at the age of 1 year. There was no history of any tooth extraction or any type of dental intervention. There was also no eruption of any posterior tooth in the left quadrants since birth.

Extraoral frontal examination of the patient revealed facial asymmetry with a depressed left zygomatic arch. (Figure 2) Lips were competent. There were multiple small skin tags in the region surrounding the left corner of the mouth. There was a scar mark in the left eye. The color of the cornea in the lateroinferior aspect of the left eye was cloudy. The extraoral profile (Figure 3) examination revealed slightly convex profile with posteriorly angulated left ear and a small skin tag in the left infratemporal region. There was not any vertebral, skeletal or cardiovascular anomaly.

Intraoral examination revealed mixed dentition stage with absence of all posterior teeth in the left quadrants. (Figure 4) The shape of all the erupted teeth was apparently normal. The height of the clinical crown was average for all the erupted teeth. The anatomy of the tongue was normal. The level of attachment of the maxil-

lary and mandibular frenums was normal. The maxillary permanent left central incisor had a mesiolabial rotation. (Figure 5) The alveolar ridge area in the region of missing teeth was expanded and the upper and lower ridges were touching to each other in centric occlusion. (Figure 4) The expansion was hard and non-tender. There was no cleft lip and cleft palate. (Figure 5)

The panoramic view (Figure 6) was peculiar in this patient. There was normal dental development in the right side where as there were multiple diffuse radiopaque lesions (complex odontomes) having cotton wool appearance in the left side. The CT scan report of the brain revealed a hypotrophy of the left hemisphere with calcification in the occipital and parietal regions.

DISCUSSION

The association of epibulbar dermoids, preauricular fistulae and skin appendages and ocular malformations as a specific entity involving the first and second branchial arches was recognized by Goldenhar.[2] Epibulbar dermoids, accessory auricular appendages and pretragal fistulae are triad of Goldenhar syndrome. The diagnosis of Goldenhar syndrome is further substantiated if vertebral anomalies are present in addition to the triad. However, presence of multiple additional symptoms makes diagnosis more difficult. Feingold and Baum offered working criteria which included a lipodermoid or lipoma of the conjuctiva, an epibulbar dermoids or an upper lid coloboma and two of the following three i.e. small size or abnormal shape of the ears or preauricular skin tags or both, unilateral aplasia or hypoplasia of the ramus of the mandible and vertebral

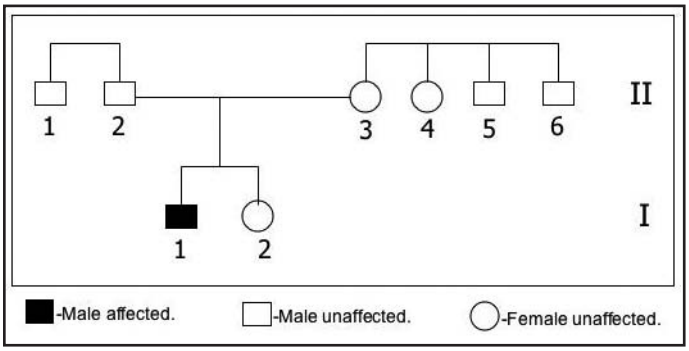

Figure 1: Pedigree analysis of two generations of the Goldenhar syndrome family revealed that the etiology does not seem to be genetic and appeared as sporadic.

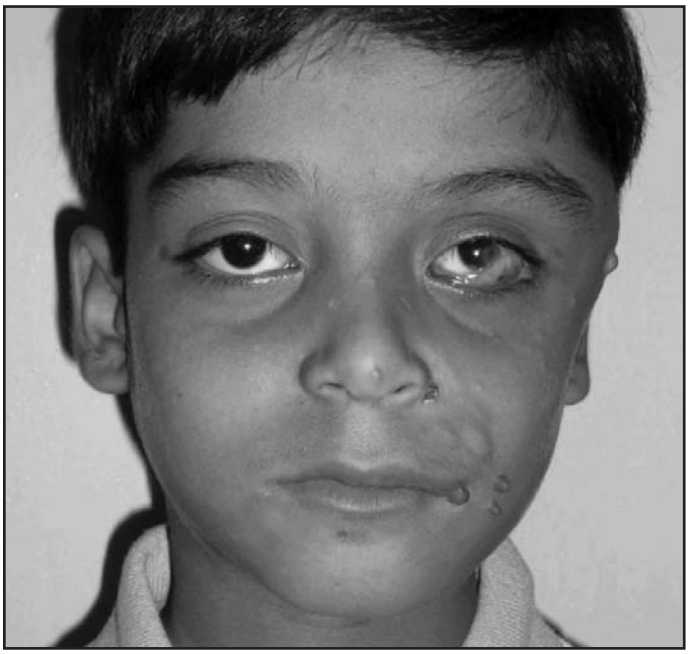

Figure 2: Extraoral frontal photograph shows facial asymmetry with depressed left zygomatic arch, posteriorly angulated left ear, scar mark in the latero-inferior aspect of the left eye and multiple small skin tags in the region surrounding the left corner of the mouth.

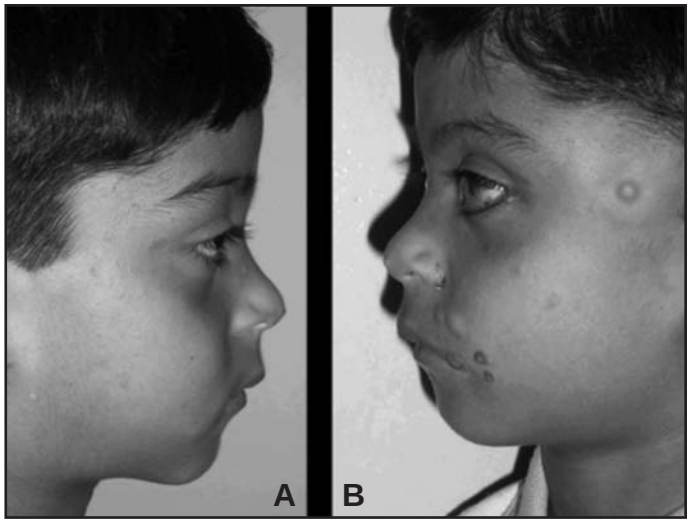

Figure 3: The extraoral profile photographs showing slightly convex profile with posteriorly angulated left ear and small skin tags surrounding the left corner of the mouth and in the left infratemporal region. A. Right lateral view, B. Left lateral view.

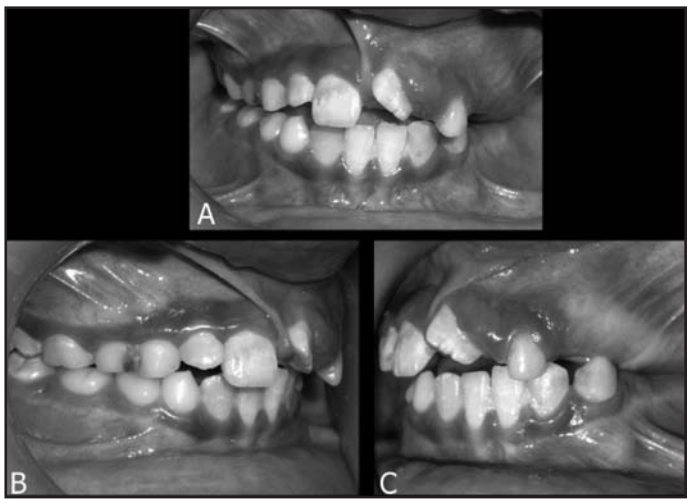

Figure 4: Intraoral photographs showing mixed dentition stage with absence of all posterior teeth in the left quadrants. A. Front view, B. Right lateral view, C. Left lateral view.

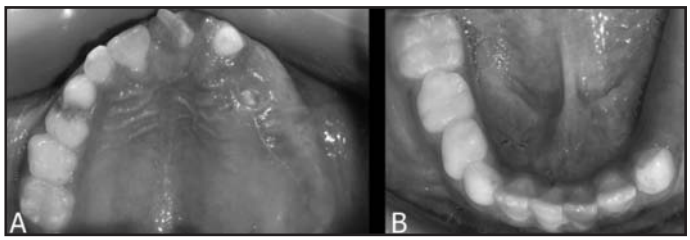

Figure 5: Intraoral occlusal photographs showing mesiolabial rotation of the maxillary permanent left central incisor and expanded upper and lower alveolar ridges in the region of missing teeth. A. Maxillary occlusal view, B. Mandibular occlusal view.

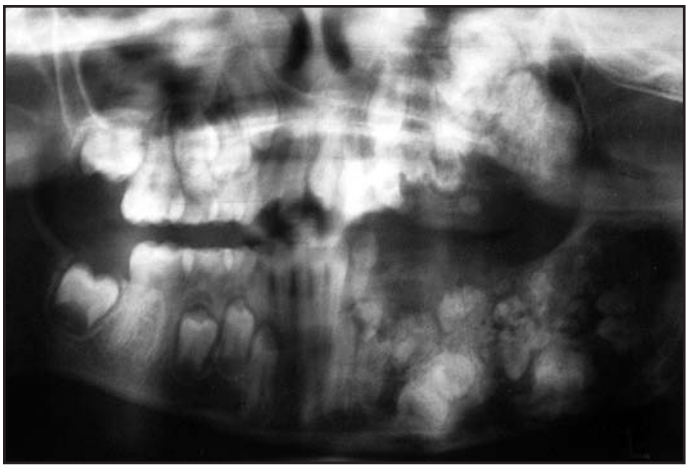

Figure 6: Panoramic radiograph showing normal development of the dentition in the right side and multiple diffuse radiopaque lesions having cotton wool appearance in the left side.

anomalies for the delineation of Goldenhar syndrome.[62]

The etiology of the Goldenhar syndrome is heterogenous. Most of the reported cases are sporadic in nature. External influences during embryogenesis have been postulated as possible etiologic factors. Fetal hemorrhage in the region of the first and second branchial arches resulted similar deformity in animal experiments.[63] Lack of concordance among monozygotic twins suggested that the disorder is the consequence of a single early localized malformation giving rise to a malformation complex rather than a genetically determined syndrome. In the present case report the etiology does not seem to be genetics and appear as sporadic.

In the present case, the patient had left side facial hypoplasia. There were multiple skin tags around the left corner of the mouth. Presence of preauricular skin tags which is a characteristic feature of Goldenhar syndrome was however absent in this case. A posteriorly angulated ear which is also a common finding of Goldenhar syndrome as reported by many authors[14-16] was present in this case. Patient had limbal dermoid for which he had undergone surgery. This finding was similar to the finding of many authors.[13-15,20] The patient had an attack of epilepsy for which he had not taken any treatment and it could be because of localized calcification in the occipital and parietal regions of the brain and hypotrophy of the left hemisphere. Martinelli *et al.* however reported ipsilateral cerebellar hypoplasia in a fetus with oculoauriculovertebral spectrum.[37] The patient had not any neurological problem at this stage.

The significant intraoral finding was unilateral presence of multiple complex odontomes in both the jaws. This finding was however atypical in Goldenhar syndrome patients. So far none of the authors in the literature has noted such findings. Whether the presence of odontomes is a component of Goldenhar syndrome or it is an independent anomaly, is not known however.

SUMMARY

Many extraoral characteristics of this case report are classical to Goldenhar syndrome. Intraoral features reported in the literature were absent in this case. Unilateral presence of multiple complex odontomes in both the jaws is a new intraoral finding in Goldenhar syndrome patients.

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