J Turgut Ozal Med Cent
ISSN 1300-1744 2017;(24:2):207-9
Submit a Manuscript: http://my.ejmanager.com/totm/ DOI: 10.5455/jtomc.2016.10.112

CASE REPORT

Goldenhar syndrome-a case report

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Abstract
Goldenhar syndrome is a rare developmental disorder affecting first and second branchial arches with its manifestations in multiple systems of the body. The aetiology of the syndrome is not clear however many theories have been proposed and described. It is characterised by presence of epibulbar dermoids, fistulas and preauricular appendages. In addition to facial defects the syndrome also manifests abnormalities in renal, genitourinary, cardiac and skeletal systems. Asymmetry in Goldenhar syndrome is usually obvious at the time of birth and increases with age, particularly at puberty. The ear defects can be both unilateral and bilateral but involvement of right ear is more common. To confirm diagnosis, it is imperative to have at least microtia and preauricular or auricular defects. The prognosis of the condition is good in patients who have no complicated systemic associations. A multidisciplinary team of doctors is required for successful treatment of the syndrome. We present here a case of Goldenhar syndrome in an 8-year-old girl with classic manifestations.

Keywords: Goldenhar; Syndrome; Hemifacial Microsomia; Mandible.

INTRODUCTION

Goldenhar syndrome was witnessed first by Canton in 1861 followed by Von Arlt, in 1881, but the syndrome went unnoticed (1,2). Maurice Goldenhar, the Swiss ophthalmologist in 1952 described the syndrome in detail when he recorded three fresh cases in addition to sixteen earlier reported cases, thus the syndrome has retained the eponym of Goldenhar syndrome till date (3). Goldenhar syndrome consists of developmental anomalies of first and second branchial arch structures (4). Goldenhar syndrome also known as oculoauriculovertebral dysplasia or hemifacial microsomia has an incidence of 1 in 3,500 to 1 in 5,600 live births (5,6). Males have predilection over females with a ratio of 3:2 (5-7). The clinical presentation of this syndrome ranges from slight asymmetry of face to severe deficiency in facial development unilaterally, with deformation of orbit and microtia, or at times a total absence of the ear (6). Goldenhar syndrome has a triad of epibulbar dermoids, accessory auricular appendages, and pretragal fistulae (8).

Received: 25.10.2016
Accepted: 15.12.2016

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Microtia and/or auricular tags are present in 100% of cases, with a number of cases often with other organ defects, such as cardiac, renal, and central nervous system disturbances and vertebral and other skeletal anomalies. (9). We present a case report of Goldenhar syndrome in an eight-year-old girl with review of literature.

CASE REPORT

An 8-year-old girl was admitted to department of oral medicine & radiology with a chief complaint of defect in face since her birth. She underwent surgery for right limbal dermoid 7 days before admission to our department. Her parents were non-consanguineously married and her sibling had no sign of any anomalies. There was no relevant family history. The patient reported decreased vision in right eye from the last 5 months and decreased hearing ability on right side since early childhood.

On clinical examination the lower half of the patient's face was asymmetrical with deviation of chin towards right side, mandibular hypoplasia and loss of malar prominence on the affected side. (Figure1a). The patient showed convex profile due to micrognathia and preauricular tag with malformed pinna on the affected side (Figure 1b). The orbit of right eye was smaller than the left one with micro cornea and surgical scar of dermoid excision on right eye could also be seen (Figure 2). On intraoral examination, a V-shaped high palatal arch was seen (Figure 3a). The patient had mixed dentition with multiple decayed teeth and no sign of any developmental anomaly was seen (Figure 3a,3b).

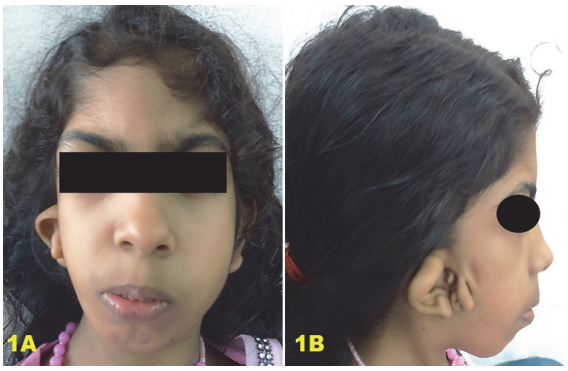

Figure 1A&B. Clinical photograph shows facial asymmetry, deviation of chin towards right side,mandibular hypoplasia with loss of malar prominence,preauricular tag and malformed pinna on affcted(right) side.

Panoramic radiograph showed aplasia of right condyle and coronoid process with decreased ramal height on right side (Figure 4). Postero Anterior view showed deficient mandible with midline shift towards the right side, deviated nasal septum to right side and scoliosis of cervical vertebrae was also evident (Figure 5). The patient was referred for an ENT and cardiac consultation. Echocardiography and Doppler studies showed our patient to be having mesocardia and trivial tricuspid regurgitation with good biventricular function. No atrial or ventricular septal defect was reported. Patent ductus arteriosus and coarctation of aorta were negative as well. Cervical spine x-ray showed signs of scoliosis. Audiological evaluation revealed moderately severe to severe mixed hearing loss on affected(right) side with hearing sensitivity within normal limits on unaffected (left) side.

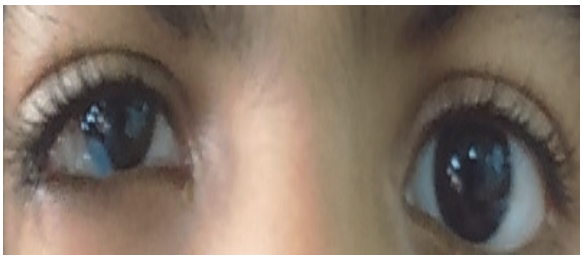

Figure 2. Right eye with micro cornea and surgical scar of dermoid excision.

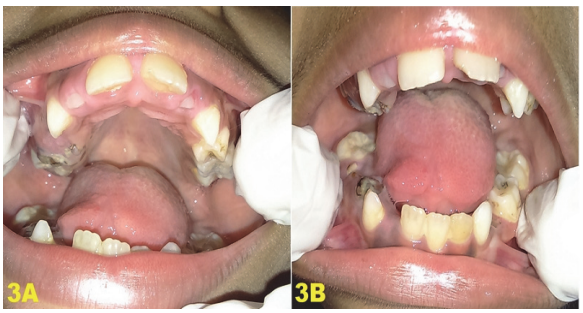

Figure 3A&B. Intraoral photograph showing V-shaped high palatal arch and mixed dentition with multiple decayed teeth.

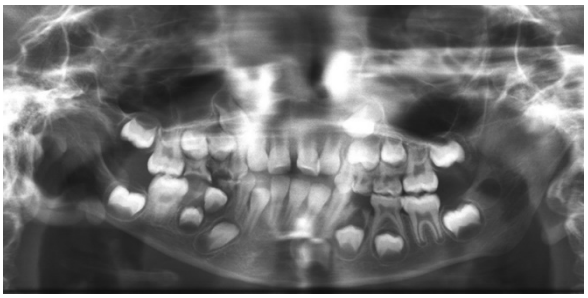

Figure 4. Panoramic radiograph showing aplasia of right condyle and coronoid process with decreased ramal height on right side.

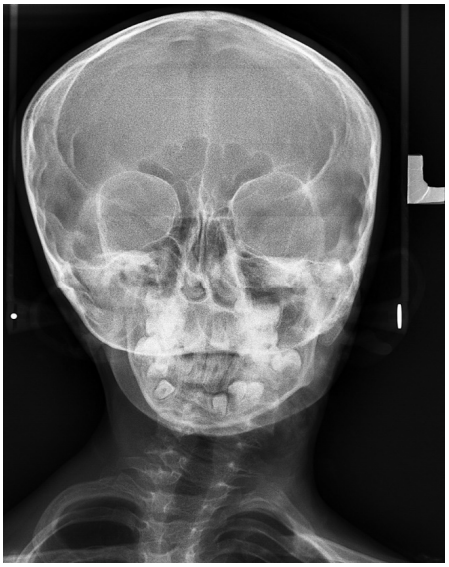

Figure 5. Postero Anterior view shows hypoplastic mandible with midline shift towards the right side, deviated nasal septum to right side and scoliosis of cervical vertebrae was also evident.

Extraction of grossly decayed teeth and oral prophylaxis was advised. The patient was also advised correction of malocclusion, corrective surgery, ear prostheses for aesthetic correction and hearing aids to improve hearing disability.

DISCUSSION

Goldenhar syndrome typically has hemifacial microsomia, microtia, dermoids, and vertebral anomalies (10). 85% of patients with Goldenhar syndrome are affected unilaterally where as it manifests bilaterally in 10% to 33% of cases (6). Goldenhar syndrome manifests with a number of inconsistencies in structures that ascend from the first and second branchial arches, first pharyngeal pouch, the first branchial cleft and the temporal bone primordia (5). Goldenhar syndrome is an outcome of diverse etiology. A type of vascular disturbance and/or neural crest pathology that leads to defective development of certain regions of the embryo which form the involved structures during the stages of embryogenesis could be a constant factor (11). The abnormality can also arise due to haemorrhage of foetus

at the time of transfer of blood supply to external carotid artery from stapedial artery during shifting of first and second arch blood supply (12). The consumption of certain drugs by mother during pregnancy such as, retinoic acid, thalidomide, cocaine and tamoxifen could lead to the development of Goldenhar syndrome. (13). Diabetes during pregnancy has also been suggested as one of the underlying factors of the syndrome (14). The history of any teratogenic exposures or any illnesses during pregnancy were unremarkable in our case and there was no history of any drug exposure during pregnancy that could have been the reason for the development of abnormality. The patient's family history was non-contributory with both the parents with no craniofacial defects.

Abnormalities of external ear and hypoplasia of facial structures ipsilaterally are considered to be as typical features associated with Goldenhar syndrome (4). Feingold and Baum have put up certain criteria for allocation of Goldenhar syndrome that include an epibulbar dermoid or an upper eye lid coloboma, lipoma of the conjunctiva or a lipodermoid and two of the following three; unilateral aplasia or hypoplasia of the ramus of the mandible, small size or abnormal shape of the ears or preauricular skin tags or both and vertebral anomalies (8), the features matching with those of our case. Mesocardia having a prevalence of 0.2 per 10000 deliveries and with a strong male predilection of 15:2 (15) was an additional feature in our case other than trivial tricuspid regurgitation.

The characteristics of Goldenhar syndrome become more marked as the child grows due to inability of involved areas to keep pace with the development of the child hence manifests in the form of abnormal occlusion, breathing problems and cosmetic complications, delays in the growth and development of the affected areas (5). The functional incapacities that are of concern at the time of birth in Goldenhar syndrome include airway sufficiency and its patency, ability at feeding and swallowing, vision, hearing and any other developmental defects that can lead to systemic problems (16). The above mentioned problems can be taken care of by infant positioning, connecting infant tongue to lower lip, placement of nasopharyngeal airway, distraction osteogenesis to advance the mandible or tracheotomy. For the maintenance of proper feeding gastrostomy tube or nasogastric feeds are used allowing sufficient oxygen supply and maintaining a positive nitrogen balance as well. According to Posnick when surgery is carried out in conjunction with a well synchronised orthodontic therapy at a time nearing the maturity of skeletal framework, the best results can be achieved (16).

Removable orthodontic appliances are used in the initial stages and later on switched to fixed orthodontic therapy during the permanent dentition stage to correct malalignment of teeth and to generate harmonious occlusion (6). Various surgical methods include costochondral rib grafts osteotomy and distraction procedure (9). If a number of organs are involved, the management of Goldenhar syndrome becomes challenging as surgical procedures are unable to treat multiple deformities with precision (5,17).

CONCLUSION

As it involves multiple organ systems of the body, Goldenhar syndrome demands a multidisciplinary effort in its management and a regular long term follow up is vital to monitor the growth and development of patients.

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