CASE REPORT

Dental manifestations of a paediatric patient with Goldenhar syndrome: A case report

Sara Ayid Al Ghamdi

Abstract

Goldenhar syndrome (GS) is a rare congenital disorder. It arises from the first pharyngeal pouch, first branchial cleft, first and second branchial arches, and primordia of the temporal bone. It mainly involves abnormalities in the ear, mandibular, and maxillary arches, and is associated with variable clinical features such as skeletal, cardiac, and renal systems. The presence of extra teeth in the dental arch is called supernumerary teeth, and hypodontia refers to congenitally missing teeth. The occurrence of both these anomalies in the same patient is called concomitant hypohyperdontia. However, the GS itself is not very rare, though the presence of concomitant hypohyperdontia has not been reported. The purpose of the present case report is to describe the first case from Saudi Arabia with a characteristic combination of rare findings in a seven-year-old child with comprehensive oral rehabilitation.

Keywords: Goldenhar syndrome, anomalies, dental treatment.

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Introduction

Goldenhar syndrome (GS) is a rare congenital complex disorder that arises from defects in the first and second branchial arches.[1] GS is also known as oculo-auriculo-vertebral syndrome, and the typical features of this disorder include abnormality of the ear, ocular dermoid and vertebral disorders, mandibular hypoplasia, and hemifacial macrosomia.[2] The prevalence of GS varies from 1 in 3,500 to 1 in 5,600 children, and is more commonly reported in males than females (3:2).[3] GS has multi-factorial inheritance by many gene interactions combined with environmental factors, such as ingestion of drugs (retinoic acid, thalidomide) and alcohol intake by the mother.[4] The diagnosis of GS involves clinical and radiologic features and laboratory results.[1] The anomalies can affect many organs in the body, including ear defects (preauricular skin tags and microtia), eye defects (colobomas of the upper eyelid, retina, Iris, and epibulbar dermoid), and cardiac, vertebral, and renal anomalies.[2,4] The occurrence of facial asymmetry, cleft lip and/or palate, and dental abnormalities is also widespread in individuals affected by GS. The occurrence of more teeth than usual is called supernumerary teeth and congenitally missing teeth is called hypodontia.[5] The occurrence of these numerical anomalies in the same individual is sporadic, and is known as concomitant occurrence of hypohyperdontia. This anomaly has not been reported in GS. Furthermore, no single report on the dental manifestations of GS from Saudi Arabia has yet been reported. Therefore, the purpose of the manuscript is to report on the dental rehabilitation of a 7-years-7-month-old male child with GS from Saudi Arabia with extremely rare findings.

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Case Report

A 7-years-7-month-old male child was reported to the dental outpatient clinic, at King Faisal Hospital, Riyadh Saudi Arabia, in December 2018, with a chief complaint of caries in all posterior teeth. He was born to a non-consanguineous couple, and was diagnosed with GS. There were neither signs of developmental and mental delay nor inheritance from the family. The medical history revealed that he had been hospitalised many times and underwent multiple surgical procedures (facial cleft repair, bilateral cleft lip repair, removal of the right corneal band, and repair of the left facial cleft with buccinator flap). He was fearful and apprehensive, with negative behaviour during the oral examination. Extra-oral examination revealed a straight facial profile (Figure-1a and 1b) and facial asymmetry with a deviation of the mandible towards the right side, repaired

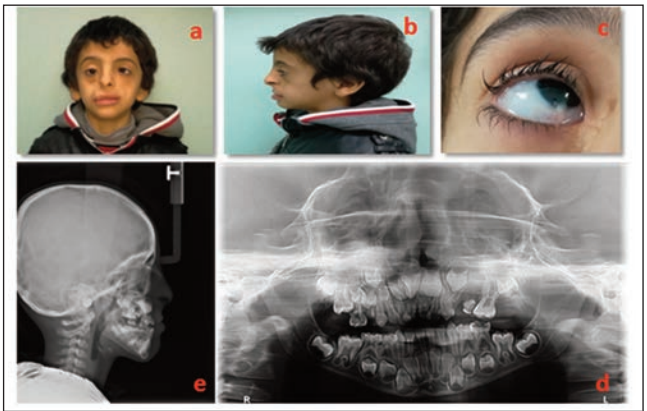

Figure-1: Extra-oral pictures (a) straight, (b) profile view, (c) coloboma of the eye, (d) orthopantomogram, and (e) lateral cephalogram of a child with Goldenhar Syndrome.

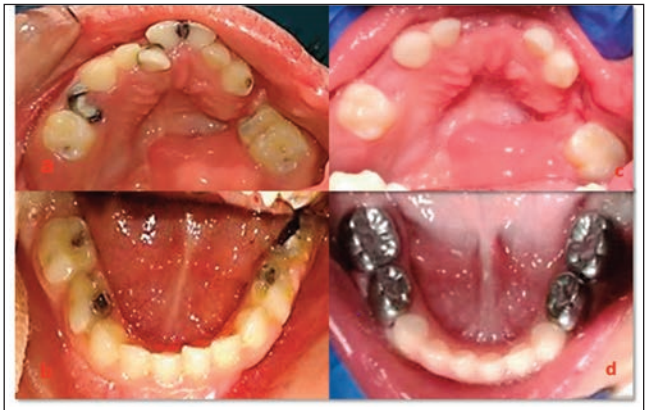

Figure-2: Intraoral clinical pictures pre-operative (a) maxillary arch, and (b) mandibular arches, and post-operative (c) maxillary arch, and (d) mandibular arches..

facial cleft (Tessir-3,4)[6] and deviation of the nasal septum to the right side. Mandibular hypoplasia and loss of malar prominence on the affected side and congenital nevus in the right eye with retinal coloboma (Figure-1c) were observed in the child. Intra-oral examination showed scar tissue in the internal buccal mucosa of the upper left corner of the mouth, repaired left unilateral cleft lip and palate (Figure-2 a). He was in the mixed dentition stage, and his oral hygiene was inadequate. He presented with a V-shaped maxillary arch and, a U-shaped mandibular arch with mesial step occlusion on the right side and class I on the left side with a 2 mm midline shift in the mandibular arch.

From the oral health aspect, the child was considered a high-risk patient with poor oral hygiene with a plaque score of 30, and no oral habit was noticed. On radiographic examination, the orthopantomography revealed (Figure-1d) hypoplasia of the mandible, ramus, and condyle. A supernumerary tooth was presented as mesial to tooth 13 and teeth 22, 24 and 25 were congenitally missing. A high mandibular angle with clockwise rotation was observed on the lateral cephalogram (Figure-1e). Deep carious lesions were present in teeth 52, 51, 61, 65, 74, 75, 84, and 85 (Figure-2a and b). The treatment plan was discussed with the parents, and informed consent was obtained. The dental treatment was performed under general anaesthesia upon consultation with a paediatrician, oral maxillofacial surgeon, and orthodontist. Under adequate nasal intubation, a throat pack was placed, and then dental prophylaxis was carried on. Composite restoration was done for teeth 55 and 26 occlusal surfaces, and proximal composite restoration for teeth 53, and 63 (Figure-2c). Pulpotomy carried out and stainless steel crown were placed in relation to teeth 74, 75, 84, and 85 (Figure-2d). Extraction of teeth 54, 65, 61, and 51, and supernumerary tooth in relation to tooth 13 was performed. A 6-month recall visit was advised to the patient, and on recent appointment, he had no complaint from the previous treatment, and topical fluoride application was performed.

Discussion

Goldenhar syndrome is a rare hereditary disorder described by Dr Goldenhar in 1952.[1] The majority of the reported cases of this syndrome were sporadic with an unknown aetiology[7] as the aetiology of GS has not been clearly documented in published literature. In the present case, the child was born to a non-consanguineous couple without any genic and family history of the disorder. However, some researchers have suggested a combination of several gene interactions and environmental factors as playing a role in the aetiology.[4,7] Foetal haemorrhage and abnormal vascular supply in the first and second arches region were also suggested as an aetiology for this syndrome.[7]

Goldenhar syndrome is characterised by various anomalies affecting body structures such as ear (92%), hemifacial microsomia (90%), eye dermoid (29%), and vertebra (20%). Abnormalities frequently are unilateral occurrence in GS in around 85% of the cases, especially the right side and rarely reported with bilateral occurrence.[1,2,4] Nonetheless, the present case is also a typical example of bilateral occurrence; the right side was severely affected than that of the left side. Hereditary heart disease is widespread with patients affected by GS. However, in the present case, the boy did not report any cardiac problems. Ear anomalies, in particular, blind fistulas with aplasia, or preauricular skin tags and external auditory meatal artesia are prevalent in GS. No ear anomaly was evident in the present case, which was rarely reported in the published literature.[1,2,4,6] High prevalence of eye abnormalities, such as upper subconjunctival dermoids, eyelid colobomas, and epibulbar choristoma, can be seen.6 In the present case, an eye dermoid and the coloboma of the right eye were observed.

Different Craniofacial anomalies were reported, such as facial asymmetry and hypoplasia of the mandible, maxilla, zygomatic bone, and asymmetric development of masticatory muscles in the affected side.[7,8] Various craniofacial anomalies were reported in the present case, including hemifacial microsomia with malar, maxillary, and mandibular hypoplasia on the right side. Similarly, oral manifestation including repaired cleft lip and palate, agenesis of the first and second premolars in the cleft side, and severe anterior malocclusion and crowding were evident in the present case. In such cases, appearance is a significant concern, and typically, the timing of the reconstruction has a vital part in the prognosis of the

treatment in GS.[3,4,7]

Dental treatment under general anaesthesia is very safe and effective for patients with multiple treatment needs and uncompromised behaviour.[9] In the present case, the patient was very uncooperative, and multiple treatments were required; hence, comprehensive patient rehabilitation was performed under general anaesthesia. The occurrence of both extra and missing teeth in an individual are extremely rare, and this concomitant numerical anomaly has not been reported in individuals with GS. To the best of the author's knowledge, this is the first such case. Al-johar and co-workers performed a comprehensive study among 447 craniofacial patients, and the authors found that GS accounts for 17% of all craniofacial anomalies in Saudi Arabia.[3] Not a single case on dental findings from Saudi Arabia has been reported in the literature. Orthodontic therapy, preferably recommended in the late mixed dentition, and various surgical methods (alveolar bone graft, surgical distraction, and orthognathic surgery), are the treatment options to manage GS with severe asymmetry cases and transverse skeletal discrepancies.[2,10] The GS prognosis is good. For the present case, three-month follow-up was done and presently the patient is under regular review. Periodic dental visits that include topical fluoride application and dental prophylaxis are recommended to prevent future dental disease and improve oral hygiene practice.[10]

Conclusion

Basic features of GS syndrome include facial asymmetry, skeletal anomalies, eye, facial asymmetry, cleft face and cleft lip, and palate; intra-orally, crowding and midline deviation, and hypodontia which are evident in majority of the cases. The present case is the first known report of Goldenhar syndrome in an Arabian subject and the only case with hypohyperdonia.

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