Goldenhar Syndrome with Tessier's 7 Cleft: Report of a Case

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Abstract Goldenhar syndrome, a variant of hemifacial microsomia, is a well-known developmental anomaly of maxillofacial skeleton that is apparent at birth. The first and second branchial arch involvement during early embryonic development results in a wide spectrum of anomalies that may include macrostomia and lateral facial clefts. Though clefts of the orofacial region are among the most common congenital facial defects, the occurrence of lateral facial clefts (Tessier 7 cleft) in conditions such as the Goldenhar syndrome, is very rare (<5%). The lateral facial cleft, which results because of improper development of the perioral muscles of the face, gives an appearance of macrostomia giving rise to potential psychological, aesthetic and feeding problems. This clinical report describes the closure of a Tessier 7 cleft and the use of distraction osteogenesis to treat mandibular asymmetry in an 11-year-old female patient with Goldenhar's syndrome.

Keywords Goldenhar syndrome · Tessier 7 cleft · Distraction osteogenesis

Introduction

Goldenhar syndrome, a term that is often used synonymously with "Oculo–Auriculo–Vertebral (OAV) Spectrum," is a well-known developmental anomaly of maxillofacial skeleton and hemi-facial soft tissue that is apparent at birth [1]. This disorder is characterized by a wide spectrum of

symptoms and physical features that may vary greatly in range and severity from case to case [1].

Goldenhar syndrome is a variant of hemifacial microsomia. Its severity can vary and the effects can be unilateral or bilateral. The physical manifestations of this disorder match those of hemifacial microsomia with the addition of several anomalies such as dermal epibulbar tumors, periauricular appendices and microtia along with disturbances of the central nervous system and other visceral anomalies [2]. The first and second branchial arch involvement during early embryonic development results in a wide spectrum of anomalies that may include macrostomia and lateral facial clefts [3]. Though clefts of the orofacial region are among the most common congenital facial defects, the occurrence of lateral facial clefts (Tessier 7 cleft) in conditions such as the Goldenhar syndrome, is very rare (<5%) [4]. The lateral facial cleft, which results because of incomplete development of the perioral muscles of the face gives an appearance of macrostomia giving rise to potential psychological, aesthetic and feeding problems. The incidence of Goldenhar syndrome has been reported to be between 1:3,500 and 1:5,600, with a male: female ratio of 3:2 the incidence is higher, about 1:1,000 in children with congenital deafness. The exact etiology is not known. However, it is possible that abnormal embryonic vascular supply, disrupted mesodermal migration or some other factor leads to defective formation of branchial and vertebral systems. Most of the cases have been sporadic. Autosomal dominant autosomal recessive and multifactorial modes of inheritance have also been suggested. Chromosomal studies have not revealed any abnormality. Ingestion of drugs such as thalidomide, retinoic acid, tamoxifen and cocaine by the pregnant mother may be related to the development of this syndrome. Maternal diabetes, rubella and influenza have also been suggested as etiologic factors [5].

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Although the Goldenhar syndrome in itself is not very rare, [5] and the Tessier 7 cleft is the most commonly seen atypical craniofacial cleft [6], the occurrence of Goldenhar syndrome with a Tessier 7 cleft, which is a rare association, prompted the report of this case. The surgical rehabilitation of the case is also discussed.

Case Report

A 11-year-old female presented to the department of oral and maxillofacial surgery with the chief complaint of facial asymmetry and macrostomia. An epibulbar dermoid tumor on the right eye and a Tessier 7 cleft (Figs. 1, 2) with an associated lateral cleft of the palate on the left side were found during the initial examination. Speech impairment was evident and the patient was blind by her right eye. Hearing was normal and the patient did not exhibit any periauricular appendices. Orthopantomographic examination detected a Pruzansky type IIA hypoplasia [7] of the coronoid process, condyle and the ramus (Fig. 3). Polydontia and a lateral cleft of the palate in the left upper quadrant were also evident in the orthopantomograph. A diagnosis of Goldenhar syndrome was made as the patient exhibited clinical characteristics of OAV syndrome as described previously including facial asymmetry, hypoplasia of the mandible and epibulbar dermoid tumor on the right eye.

Since the patient felt that she faced rejection from her peers because of her “disfigurement”, the closure of the cleft was given priority over other planned surgical procedures. A Pfeiffer wavy incision [8] technique was used for Tessier 7 cleft closure (Fig. 4). After marking the proposed oral commissure, the vermillion turnover flap was raised and the upper and lower muscle bundles were skeletonized and divided. The upper bundle was sutured over lower one in a crisscross manner to create the modiolus and a straight-line three-layer skin closure was obtained. Sutures were removed after a week and healing was satisfactory (Fig. 5). The lateral cleft palate was operated at the same time.

To correct the functional and anatomical impairments caused because of hemifacial microsomia in this syndrome, distraction osteogenesis was considered as growth modification with orthodontics generally produces no more than 5 mm differential growth and conventional orthognathic procedures become more difficult and less predictable when greater than 8–10 mm advancement is needed [7]. The distraction as a second stage procedure was done after 4 months after the lateral cleft closure. The buccal plate was corticotomised with a surgical drill under copious irrigation, and particular attention was paid to the lingual cortex. The upper part of the lingual plate was sectioned

Fig. 1 Pre-operative view. Facial asymmetry and a Tessier 7 cleft on the left side along with an epibulbar dermoid tumor involving the right eye

Fig. 2 Pre-operative view of the Tessier 7 cleft on the left side of the face

with a drill, and the remaining portion fractured using an osteotome after securing a custom made intra-oral sub-mucosal distractor at the proposed site in the proposed

Fig. 3 Orthopantomograph showing polydontia, hypoplasia of the ramus and the coronoid and the condylar processes of the mandible on the left side. A lateral cleft of the palate is also visible

Fig. 4 Proposed outline for the lateral cleft closure with the Pfeiffer wavy incision technique. The lateral cleft of the palate can also be seen

tangential vector. A custom made intra-oral submucosal distractor was placed over the corticotomy site, in an oblique vector. The activation screw was exteriorized through a retromandibular stab incision to facilitate the activation of the appliance (Fig. 6). After a latency period of 7 days the distractor screw was turned daily, on an out-patient basis. This was designed to result in 1 mm per day distraction (0.5 mm activation twice a day) of the bony segments and was continued until an advancement of 2.0 cm was achieved (Fig. 7). The distractor was left in situ for (for consolidation of the distracted callus) 8-week period of fixation, after which it was removed. Following treatment, the patient had an elongated facial contour and corrected occlusal plane (Figs. 8, 9). The patient is

Fig. 5 Post-operative view after the lateral cleft closure

Fig. 6 The custom-made distractor with the activation screw exteriorized through a retromandibular stab incision to facilitate the activation of the appliance

Fig. 7 Distraction osteogenesis after 6 weeks showing bone formation of about 2.5 cm

currently under observation and future plans include orthodontic therapy directed towards the comprehensive oral rehabilitation of the patient.

Fig. 8 Post-operative view: The patient after the completion of distraction osteogenesis

Fig. 9 Profile view of the patient after the completion of distraction osteogenesis

Discussion

There is an overall consensus that the diagnosis of this disease must not be only based upon radiologic or laboratory results. Facial asymmetry or facial and/or mandibular hypoplasia, dermal epibulbar tumors, palpebral alterations, vertebral anomalies, lateral facial clefts, and renal problems are frequently seen in this condition [9]. Additionally both laboratory and image tests are important for the diagnosis of the disease because anomalies of the skeletal or facial bones can be diagnosed by means of several types of imaging available today.

In this case, the patient showed a prominent lateral cleft; an alteration which is observed in just about 5% of the cases. Macrostomia is generally found in the autosomal dominant form of the Goldenhar syndrome within the spectrum [10]. The patient also exhibited facial asymmetry, hypoplasia of the mandible and an epibulbar dermoid tumor on the left eye. Facial asymmetry and hypoplasia of the mandible are typical features of OAV syndrome though the presence of epibulbar dermoid tumor is highly variable [1, 11, 4].

The classical features of this syndrome include ocular changes such as microphthalmia, epibulbar dermoids, lipodermoids, coloboma; aural features such as preauricular tragi, hearing loss and microtia; and vertebral anomalies such as scoliosis, hemivertebrae and cervical fusion. The abnormalities are found to be unilateral in 85% cases and bilateral in 10–33% cases. In Goldenhar syndrome ocular anomalies especially bilateral dermoids are seen in 60% of the cases, vertebral anomalies also in 40% of the cases [5, 6]. Other systemic features are found in about 50% of the patients. Tetralogy of fallot and ventricular septal defect are the most common cardiovascular anomalies associated with OAVS. Cleft lip and palate, macrostomia, micrognathia, webbing of neck, short neck, tracheoesophageal fistula, and abnormalities of sternocleidomastoid muscle, umbilical hernia, inguinal hernia, urological abnormalities, hypoplastic vagina and anal abnormalities may be associated. Anophthalmos, facial palsy, calcification of falx cerebri, undescended testes and association of Goldenhar syndrome with Turner's syndrome and glaucoma are rarer reported associations.

The treatment of the disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases. In patients with mandibular hypoplasia, reconstruction can be done with rib grafts and an underdeveloped maxilla can be lengthened by a bone distraction device. Reconstruction surgeries of the external ear may be performed at the age of 6–8 years. In patients with milder involvement, jaw reconstruction surgeries can be done in the early teens; epibulbar dermoids should be surgically excised. Structural anomalies of eyes and ears can be

corrected by plastic surgery. Prognosis of the disease is good in other wise uncomplicated cases without any systemic associations [7–9].

In patients beyond 6 years, orthodontic treatment, facial soft tissue augmentation, limited bone augmentation of the deficient portions of the craniofacial skeleton are the important points to be considered [12]. Distraction osteogenesis is an accepted treatment modality in the treatment of congenital and acquired deformities [13], in osseous defects caused by trauma and in severe alveolar deficits secondary to periodontal disease [14]. Furthermore, in the last few years, this surgical technique has been successfully used in the treatment of pediatric deformities such as hemifacial microsomia, some severe class II skeletal deformities, and some syndromic cases, such as the treatment of TMJ ankylosis outcomes [14].

Apart from orthodontic treatment, ophthalmic and psychological concerns must also be addressed. As the surgery may have a significant impact on communication and swallowing, speech therapy may be indispensable. In conclusion, this clinical report describes the closure of a Tessier 7 cleft and the use of distraction osteogenesis to treat mandibular asymmetry in a patient with Goldenhar syndrome. As the hemifacial soft tissue manifestations in this case was different from other cases of Goldenhar syndrome, this case illustrates the importance of having a proper approach best suited to the clinical situation of the patient.

Conflict of Interest None declared.

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