Interim obturator in an infant with Treacher Collins syndrome: Review and chairside modification in impression making

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Abstract

Treacher Collins syndrome has been described as a syndrome involving 1st and 2nd branchial arches, affecting various organs in the craniofacial region. Affected infants report with nasal regurgitation and minimal dietary intake due to cleft palate, consequently show delayed and retarded growth. The situation is further complicated when the repair of the palatal defect is postponed due to delayed milestones. At this juncture, it is of paramount importance to intervene prosthetically and close the defect with the aid of an interim obturator. Herein we describe a simple, yet successful, chairside approach to make an impression of an infant without the aid of any kind of anesthesia.

Keywords: Branchial arches, interim obturator, treacher collins syndrome

Introduction

Treacher Collins syndrome (TCS) or mandibulofacial dysostosis is a congenital, complex craniofacial developmental disorder with an autosomal dominant mode of inheritance. It presents as a peculiar facial phenotype associated with variable clinical presentation and genetic heterogeneity.[1-7]

The first historical reference to the condition was published in 1889 by Berry,[2] who reported the deformity in a mother, her daughter and also in mother's brother. Treacher Collins[2] was apparently the first to emphasize the chief features of the syndrome: hypoplasia or underdevelopment of the zygomatic bone giving a flattened appearance to the cheeks. Elder[2] reported that symmetrical coloboma in the lateral portion of the eyelid is an almost constant feature of the syndrome. Franceschetti *et al*.[2,5] included deformities of the ear and mandible, and named it as mandibulofacial dysostosis. Individuals affected by the condition show a strong, almost familial resemblance.

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The gene responsible for causing TCS is *TCOF1* and maps to long arm of chromosome 5 (gene map locus 5q32-q33.1) and encodes a simple nucleolar phosphoprotein called Treacle.[3,4] TCS be diagnosed during the prenatal period (not until the second trimester of pregnancy) by fetoscopy or an ultrasound examination. However, mild manifestations may go unnoticed. Molecular diagnosis by the identification of defective *TCOF1* can be done in the 1st trimester of pregnancy.[7] The degree of malformation present at birth is believed to be relatively stable and non-progressive with age.

The TCS involves 1st and 2nd branchial arches and is generally characterized by bilaterally symmetrical abnormalities involving the craniofacial region. The external ears may be absent, malformed or malposed. The conductive hearing is impaired (due to variable degree of hypoplasia of the external auditory canal and ossicles of the middle ear), but mixed or sensorineural hearing loss is rare.[7] The size and shape of the nose may present as narrow, deviated, hooked or kyphotic. Blind dimple may be observed on the cheeks and "tongue-shaped" processes of the hair frequently extend into the preauricular region.

The wide buccal commissures (macrostomia) are a frequent feature with the syndrome. In some cases, narrow arch, high palate and cleft palate with or without cleft lip may be present. Mandible and the body of the maxilla are reported to be hypoplastic (small) even though they appear normal. In many cases, vertebral malformation may accompany the complex.

Radiographically, mandible, maxilla and zygomatic bones are underdeveloped. The defects in the maxilla include its generalized hypoplasia with shortened posterior margin and thin orbital floor associated with small antra. The mastoid processes are small and may not have any air cells. The auditory ossicles are often absent or malformed and the inner ear may be sclerotic.[8]

Myriad of dental anomalies like tooth agenesis (single or multiple), supernumerary teeth, T-shaped teeth, enamel hypoplasia, microdontia, tooth rotation, ectopic tooth positioning and crowding due to small jaws are associated with the TCS patients.[9]

Case Report

A 10-month-old male infant was referred to the Department of Prosthodontics, Goa Dental College and Hospital, India, with the chief complaint of relative inability to drink mother's milk and milk from the feeding bottle. Parents reported frequent nasal regurgitation of milk and water since birth.

Extraoral examination showed that both the eyes had an antimongoloid palpebral slant with coloboma or notching of the lateral part of the right lower eyelid and absence of eyelashes on the same eyelid [Figure 1]. The cheeks did not show any fullness; they appeared flat due to the absence of zygomatic buttress. Another feature observed was the bilateral presence of atypical tongue-shaped projections (sideburns) of the hairline extending toward the cheeks. Both the ears were malformed and hypoplastic [Figure 1], but the child still responded to sound, although of a higher intensity. The mandible was underdeveloped and retruded. Combination of prominent dorsum of the nose and retruded mandible with decreased frontonasal angle gave the patient a convex profile [Figure 2]. Macrostomia with wide buccal commissures was a prominent feature [Figure 3].

Intraorally, the child was completely edentulous. The midline cleft of hard and soft palate was seen; however, the premaxilla was prominent and intact. No abnormality was found on the oral mucosa and tongue.

Radiographic evaluation revealed that the teeth were in the developing stage, there was complete absence of the zygomatic process of the temporal bone and absence of the lateral wall of the orbit with floor of the orbit sloping downward. Angle of the mandible was more obtuse with prominent antegonial notch. The coronoid process and the condyle were flattened or aplastic. Both maxilla and mandible were underdeveloped and hypoplastic [Figures 4 and 5].

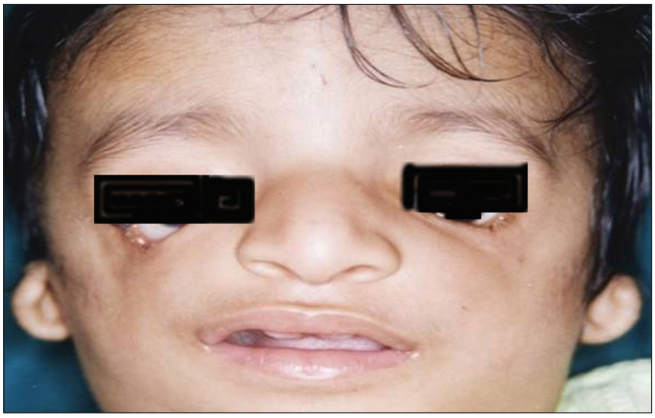

Figure 1: Frontal view showing antimongoloid palpebral slant and coloboma (notching) and missing eyelashes of the right lower eyelid, bilaterally malformed and hypoplastic ears

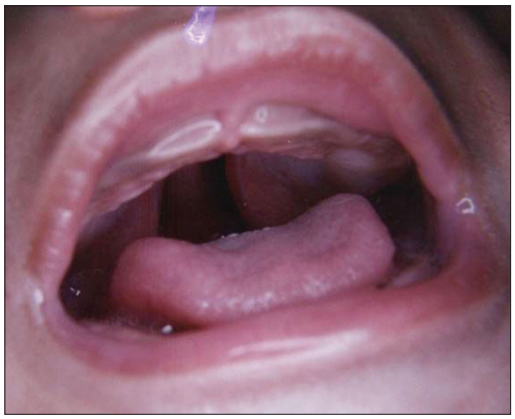

Figure 3: Midline cleft of the hard and soft palate. Intact premaxilla

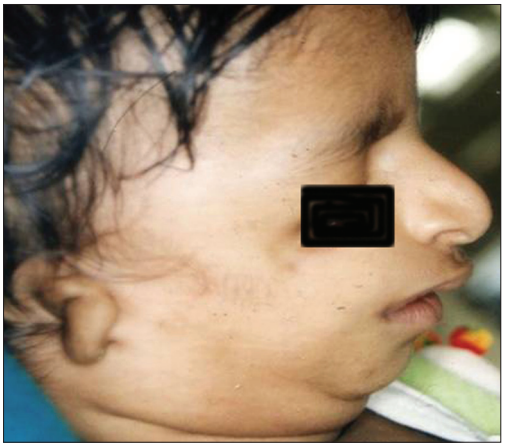

Figure 2: Presentation of sideburns of the hairline, malformed ear on the right side and convex facial profile

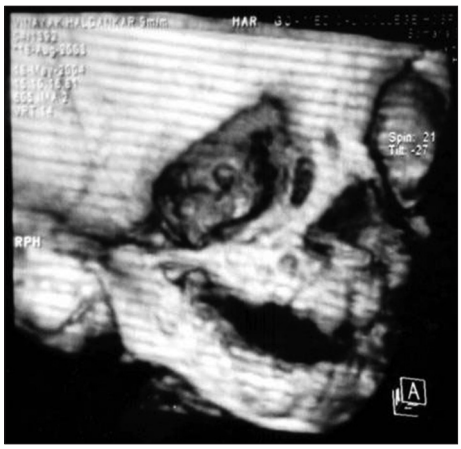

Figure 4: CT scan showing absence of zygomatic process, hypoplastic condyle and coronoid process. Lateral slant of the right orbital floor

The infant had typical features of TCS with a non-contributory family history.

The child was reported to have delayed milestones since birth. He was found to be underweight for the cleft palate surgery; therefore, it was advised to make an obturator for the child so as to improve the dietary intake.

Impression making

Due to the age factor, it was decided to complete the impression in minimal time with the intention of recording the maximum area possible within the limits of oral health and function.

Initially, "impression compound" (Y-Dents, MDM Corporation, New Delhi, India) was softened and tempered to make sure that the temperature of the impression material was well tolerated by the child. The softened compound was taken on the index finger and placed in the child's mouth. However, due to the disagreeable taste of the impression compound, the child repeatedly refused to place the material in the mouth.

Making the impression under anesthesia was ruled out due to the associated difficulties and complications in patients of TCS.[10,11] Also, molding of the material was dependent on the sucking movement of the child to record the peripheral limit of the prosthesis. To overcome this problem, the procedure was repeated in a concentrated sugar solution for both softening and tempering the impression compound. The patient then allowed to place the compound inside his mouth and even did sucking over the material. This preliminary impression was then used as the tray for border molding with "low fusing impression compound" (Aslate, Asian Acrylate, Mumbai, India), where tempering was again done in the warm sugar solution. There was an adequate retention and stability of the compound tray. Finally, a wash impression of the arch was made in the "light body polyvinyl siloxane" (Flexitime, HereusKulzer, 300HeraeusWay, SouthBend, USA) [Figure 6].

Two stainless steel wires were inserted in the wax framework so as to hold the prosthesis from outside during feeding. Obturator was then made in "heat-cure denture base acrylic resin" (DPI, Mumbai, India). The prosthesis was trimmed and polished, taking extra care to remove sharp borders and blebs from the intaglio surface of the plate. The ends of both the wires were bent and covered with "self-cure acrylic resin" (DPI) pads to prevent any injury to the facial skin during its use.

The feeding plate was tried in the mouth after applying honey on both outer and inner surfaces. It was adequately retentive [Figure 7]. Parents were instructed to apply honey on both internal and external surfaces of the feeding plate for the first few days to make the child adapt to it. Thorough cleaning of the obturator as well as oral cavity was advised after each feeding session. It was advised to keep the child in sitting posture while feeding and the same was demonstrated.

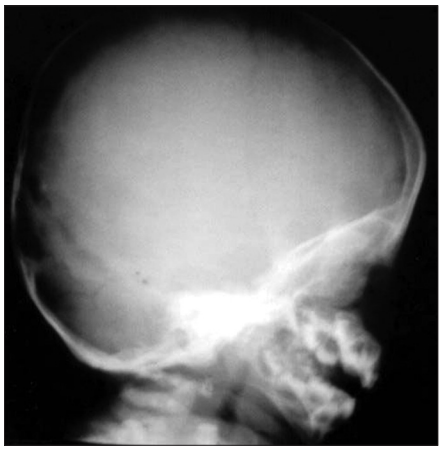

Figure 5: Radiograph showing teeth in developing stage and prominent antegonial notch

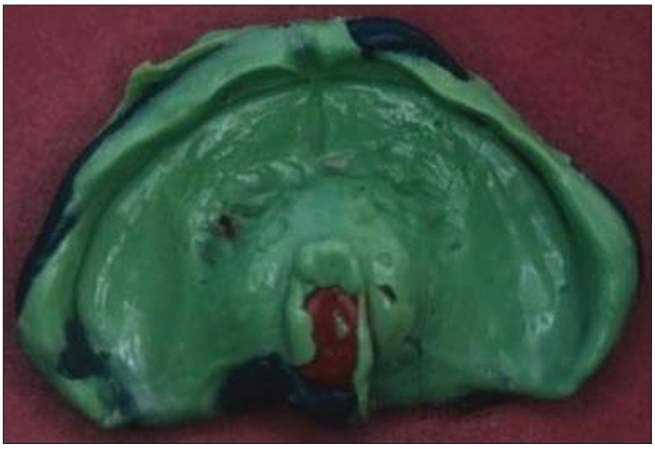

Figure 6: Final impression in light body additional silicone

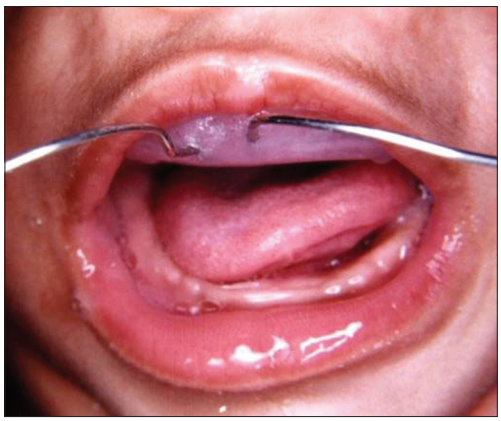

Figure 7: Adequate retention of the prosthesis, macrostomia, completely edentulous mandibular arch

After 1 week, there was reported decrease in regurgitation of the liquids and the child even started to have milk in the feeding bottle, although with great difficulty.

Discussion

Reported incidence of TCS syndrome is 1 in 50,000 live births, with no sex predilection. Racial predisposition among the Negroes, Chinese, Japanese and Asiatic Indians has been observed.[7,12]

Heredity plays a major role in the expression of the disorder and inheritance follows an autosomal dominant pattern. Suggestions have been made that the female of the affected line may genetically transmit the syndrome,[12] influenced by increase in the paternal age.[13,14]

Although in the reported case there has been no positive family history and none of the other three siblings were affected with the disorder, it is reported that 60% of the cases affected with TCS occur due to spontaneous mutation.[7]

TCS has been classified[2] depending upon the number of deformities expressed in a case into the following:

1. Complete form: The deformity in this form is severe and all the features of the syndrome can be seen.
2. Incomplete form: The patient expresses less severe and less extensive deformity.
3. Abortive form: In this, only the eyelid abnormalities are usually present.
4. Unilateral form: In this group, the deformities observed are confined to only one side of the face.
5. Atypical form: In this form of the syndrome, the principal characteristics of the complete, fully developed syndrome are missing.

The case reported here is of complete form.

In this patient, it was very important to have immediate prosthodontic intervention as reconstructive surgery was postponed indefinitely. Considering the child's age, small, retrognathic mandible and insufficient oropharyngeal space, it was critical to complete the impression in minimal time. Prosthesis adaptation was a concern at such young age, but the feeding patterns improved in 4–5 days.

Differential Diagnosis

Goldenhar syndrome[15] (hemifacial microsomia) is confused with TCS because of common clinical features, but it is more sporadic in vast majority of the cases as compared to TCS which is often hereditary. Its presentation is unilateral and affects the upper eyelid, whereas typical TCS presents as a bilateral anomaly and affects the lower eyelids.

Clinical presentation of TCS and Nager syndrome[16,17] may overlap, but the clinical features are less severe in the latter. Nager syndrome also includes characteristic limb anomalies (preaxial or thumb/radial anomalies). Sporadic and autosomal recessive pattern of occurrence have been reported. Females are affected more than males (63% females), and gene mutation for this syndrome might be associated with chromosome 9q.[17]

Miller syndrome[18] (postaxial acrofacial dysostosis) comprises craniofacial abnormalities similar to those of the TCS but associated with upper and lower limb defects mainly on the postaxial side of the limb.

Management

It depends upon the age of the patient and degree of the deformities. The current approach to correct the malformations associated with TCS is aimed at coinciding the reconstruction of the missing or deficient elements of the facial skeleton with the facial growth patterns, visceral function and psychological development. The prognosis of the patient is reported to be good and the patient can lead a normal life.[12,15]

Impression making for such patients without anesthesia is very challenging. The described chairside modification worked well for this infant. Although the feeding plate needs to be refabricated regularly due to the overall slow but constant growth, it offers an interim treatment option till the palatal defect is closed surgically.

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