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Research Article

Dental Anomalies in Individuals with Treacher Collins Syndrome – Tomographic Analysis

Letícia Maria Ramos Zanchetta[1], Patrícia Jost[2], Adriano Porto Peixoto[2], Beatriz Costa[1] and Gisele da Silva Dalben[1]*

[1]Department of Pediatric Dentistry, University of São Paulo, Brazil
[2]Department of Orthodontics, University of São Paulo, Brazil

*Corresponding author
Gisele da Silva Dalben, Department of Pediatric Dentistry, Hospital for Rehabilitation of Craniofacial Anomalies, University of São Paul, Rua Silvio Marchione, 3-20 – Vila Universitária, CEP 17012-900 – Bauru – SP – Brazil, Tel: 55(14) 3235-8141; Fax: 55(14) 3234-7818; Email: gsdalben@usp.br

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Abstract

The Treacher Collins syndrome is a genetic disorder with prevalence of 1:50,000 livebirths, characterized by multiple craniofacial deformities, requiring high-complex and multidisciplinary treatment. This study analyzed the occurrence of dental anomalies in individuals with Treacher Collins syndrome, by analysis of computed tomographies obtained before orthodontic or surgical treatment, to analyze the dental findings associated with the syndrome. The study was conducted on computed tomographies of 42 individuals, among which 20 were excluded according to the study criteria, 4 had no alterations, and 18 individuals presented dental anomalies. There was high prevalence of anomalies of number and position, especially hypodontia (23.8%) and rotations (23.8%).

INTRODUCTION

The Treacher Collins syndrome (TCS) is a craniofacial developmental disturbance, first reported in the medical literature by Thomson in 1846, yet later described in more detail by Treacher Collins [1]. It is caused by abnormal development of facial structures from the first and second pharyngeal arches [2], and early diagnosis allows proper esthetic and functional multidisciplinary treatment [3].

The syndrome is clinically defined by bilaterally symmetric characteristics [4], with several oral findings and high prevalence of dental anomalies, suggesting a possible etiological relationship, not fully elucidated in the literature so far [5].

The Treacher Collins syndrome (TCS) is a rare autosomal dominant genetic disorder [6] with high penetrance and variable expressivity [7], It occurs between the 5th and 8th weeks of intrauterine life [8], and may be diagnosed prenatally by ultrasound examination, evidencing micrognathia and microtia in the fetus [5], being mainly characterized by multiple craniofacial deformities [9]. Even though Thomson was the first to describe the syndrome in 1846, Treacher Collins described its fundamental aspects in 1900. Later, in 1949, Franceschetti and Klein conducted extensive studies on the syndrome and provided more accurate description of its characteristics [10,11].

The syndrome is caused by mutations in chromosome 5 (5q32-33.1) in gene TCOF1, called 'treacle' [12], which cause a reduction in neural crest cells, which are necessary for the craniofacial embryonic development by genic transcription of DNA [13]. It has also been suggested that the disorder may also be caused by abnormalities in cell differentiation or extracellular matrix [9].

The estimated prevalence is 1 in every 50,000 live births, with 40% of cases with autosomal dominant inheritance, and the remaining 60% classified as genetic mutations [14].

Its occurrence is believed to be influenced by the increase in paternal age [15], without predilection of gender [13] or ethnicity, and the risk of transmission to the offspring is 50% [16].

The anomalies are usually bilateral and symmetric and present wide phenotypic variation, both inter- and intrafamilial, spanning from underdiagnosed cases to perinatal death, usually caused by airway collapse in more severe cases [1].

The main clinical aspects mentioned in the literature on TCS are: convex facial pattern with prominent nose and retruded chin [3], downslanting palpebral fissures, maxillary hypoplasia, upper eyelid coloboma, total or partial absence of the lower eyelashes, external ear malformations, conductive hearing loss, macrostomia, malformations of the heart, cervical vertebrae, kidney and limbs, intellectual disability (5% of cases), obstructive apnea (25% of cases) and curved lower mandibular border [8].

Concerning the oral aspects, the following characteristics may be observed: cleft palate, short soft palate, enamel hypoplasia, open bite [8] supernumerary teeth impacted at the maxillary anterior region, hypoplasia and abnormal positioning of maxillary central incisors, micrognathia, temporomandibular joint dysplasia (TMJ), limited mouth opening, midline deviation,

lack of occlusion on the right side, deep bite, Class II or III malocclusion, maxillary rotation and retrognathism in relation to the cranial base [17], xerostomia [13], hypodontia, especially of the mandibular second premolars, and ectopic eruption of the maxillary first molars [7].

A previous investigation on individuals with TCS observed anomalies of number, shape and position, mainly hypodontia in the permanent dentition [5,18].

Nearly half of individuals with TCS present dysplasia or aplasia of greater salivary glands, as observed by ultrasound imaging, which may significantly increase the caries, risk [19]. The mandible and temporomandibular joint (TMJ) may also be severely affected, and the condyle malformation may be the mandibular region most severely affected [20].

Individuals with TCS usually require multiple surgical procedures and multidisciplinary treatment throughout childhood and adolescence [20], involving craniofacial surgeons, pediatric dentists, orthodontists, otolaryngologists, ophthalmologists, pathologists, psychiatrists and psychologists [7]. It is estimated that 66% of individuals with TCS require some airway intervention, with emphasis to maintain an adequate airway in newborns with the syndrome, since dysphagia and difficult weight gain are often primary symptoms of airway involvement [10].

The dental treatment of individuals with Treacher Collins syndrome may be complex due to the difficult management of children with hearing impairment and micrognathia, often precluding the outpatient care [17], requiring general anesthesia in some cases [21]. Individuals with TCS often require orthognathic surgery with anterior maxillary impaction and maxillary extrusion, thus opening the posterior nasopharyngeal airway. They also require procedures as rhinoplasty, genioplasty and soft tissue increase, which may improve their facial esthetics and quality of life [22].

Computed tomography has been used in cases of syndromic and non-syndromic craniofacial anomalies to determine and record the abnormal anatomic structures. Tomographic images are used both for diagnosis and to observe the craniofacial bones, allowing morphological analysis of these bones in individuals with deformities [3] and for surgical planning [7].

This study analyzed the occurrence of tooth abnormalities in permanent teeth of individuals with Treacher Collins syndrome.

MATERIALS AND METHODS

This study was approved by the Institutional Review Board of HRAC–USP (protocol n. 2.096.011). This was a cross-sectional, retrospective study with quali-quantitative design.

The study was conducted at the Hospital for Rehabilitation of Craniofacial Anomalies, University of São Paulo (HRAC-USP). Computed tomographies of individuals with Treacher Collins syndrome were analyzed for observation of tooth abnormalities affecting the permanent teeth. The study considered the following inclusion criteria: presence of Treacher Collins syndrome, diagnosed by clinical examination by the Clinical Genetics sector of HRAC/USP; availability of at least one computed tomography of the face/facial sinuses/temporomandibular joints in the files of HRAC/USP, obtained before orthodontic treatment of surgical procedures to the maxillomandibular complex; and individuals aged more than 6 years. Tomographies of individuals whose information available on the records did not allow analysis of previous dental treatment were excluded.

The study included all individuals who met the inclusion/exclusion criteria described above, aiming to analyze 100% of the sample available for analysis.

The computed tomographies were analyzed in different planes and sections, to evaluate tooth abnormalities affecting the permanent teeth. The number of present teeth was also recorded, to calculate the percentage of affected teeth.

Tooth abnormalities were classified as alterations of shape, number and position [23], and as hyperplastic, hypoplastic and heterotopic disorders [24]. Data were recorded in a form especially designed for the study. The presence and type of cleft lip and palate was also recorded.

The results were analyzed by descriptive statistics. The results between individuals with different types of clefts were compared by the chi-square and Mann-Whitney tests.

RESULTS AND DISCUSSION

Computed tomographies were available from 62 individuals with Treacher Collins syndrome. Among these, 20 individuals were excluded because of previous orthodontic treatment or orthognathic surgery or images with poor quality.

Dental anomalies were observed in 38 individuals (90.5%), with predominance of anomalies of number and position, especially hypodontia and rotation, followed by root dilaceration, anomalies of shape and ectopic position, pulp nodule, microdontia and mesiodens supernumerary tooth.

Among hypoplastic anomalies of number, there was predominance of hypodontia, with 24 permanent teeth missing in 10 individuals (23.8%), primarily affecting the mandibular second premolars (n=10), followed by the maxillary second premolars (n=6), maxillary lateral incisors (n=6) and mandibular molars (n=2). One individual presented hyperplastic anomaly of number, exhibiting one mesiodens at the region of maxillary central incisors.

Concerning heterotopic alterations, rotations also presented high prevalence, being observed in 10 individuals (23.8%). Overall, 21 rotated teeth were observed, mainly affecting the mandibular lateral incisors (n=8), followed by the mandibular canines (n=6), mandibular central incisors (n=3), maxillary canines (n=2), maxillary central incisor (n=1) and mandibular first premolar (n=1). There were also two teeth in ectopic position in two individuals, being one maxillary canine and one mandibular second molar.

Concerning hypoplastic anomalies of shape and structure, root dilaceration was observed in 4 individuals (9.52%), affecting nine teeth, with higher prevalence in molars (n=5), followed by canines (n=2) and premolars (n=2). Microdontia was observed in one individual, affecting two maxillary lateral incisors. Crown shape anomaly was observed in two individuals, affecting the

maxillary central incisor and mandibular second molar.

With regard to hyperplastic anomalies of shape and structure, one individual presented two pulp nodules, both affecting the maxillary second molars.

The detailed findings for the entire sample of individuals are presented in Table 1.

Individuals with TCS have been shown to present anomalies of number, shape and position, including hypodontia,

Table 1: Distribution of dental anomalies observed per patient.

PATIENTS	DENTAL ANOMALIES									Exclusion
	Hypodontia	Supernumerary tooth	Rotation	Ectopic position	Root dilaceration	Microdontia	Crown anomaly	Pulp Nodule	No anomalies	
Patient 1										X
Patient 2										X
Patient 3			33,43,44		38					
Patient 4										X
Patient 5*										X
Patient 6										X
Patient 7	25,35 and 45									
Patient 8										X
Patient 9*	12,22,15 and 35						11			
Patient 10										X
Patient 11										X
Patient 12				23	37,47 and 45			27 and 37		
Patient 13										X
Patient 14										X
Patient 15										X
Patient 16										X
Patient 17										X
Patient 18										X
Patient 19										X
Patient 20										X
Patient 21										X
Patient 22	12,15,25 and 35		41 and 42							
Patient 23									X	
Patient 24	15,25,35 and 45			48	13, 14 and 23					
Patient 25										X
Patient 26*	22									
Patient 27									X	
Patient 28									X	
Patient 29*	12 and 22		11							
Patient 30										X
Patient 31	35	Between 11 and 21								
Patient 32			42							
Patient 33			33							
Patient 34			13 and 33							
Patient 35	47		32 and 42							
Patient 36	35 and 36				37 and 46		47			
Patient 37										X
Patient 38			42, 32 and 33							
Patient 39			13,43,32,31 and 42							
Patient 40									X	
Patient 41	35 and 45		41							
Patient 42						12 and 22				

*Individuals with complete cleft lip and palate

supernumerary teeth, ectopic eruption of the maxillary permanent first molar, rotated teeth [5], abnormal positioning of maxillary central incisors [18], besides enamel hypoplasia [8] supernumerary teeth, hypoplasia and abnormal positioning of teeth, micrognathia, midline deviation, deep bite, malocclusion [17], xerostomia [13], hypodontia and ectopic eruption [7].

According to the literature, hypodontia is the most frequent dental anomaly in individuals with the syndrome, primarily affecting the mandibular second premolars [7], followed by the maxillary second premolars, maxillary lateral incisors and maxillary canines [5]. This study observed higher frequency of hypodontia in mandibular premolars, followed by the maxillary premolars and maxillary lateral incisors, similar to the literature. Besides these, the mandibular molars were congenitally missing in two individuals, namely the mandibular right second molar in one case (patient 35) and the mandibular left first molar in another (patient 36).

Also regarding hypodontia, four individuals presented associated cleft lip and palate (CLP), bilateral in two cases and unilateral left in other two cases. Cleft lip and palate increases the frequency of hypodontia at the cleft area in 5 to 7 times [25], therefore the hypodontia of maxillary lateral incisors observed in the present individuals with TCS are possibly related with CLP, yet not strictly related, as mentioned in a previous study [17], since one individual (patient 22) exhibited isolated cleft palate and hypodontia of the maxillary right lateral incisor.

Concerning the rotated teeth, some authors [5] reported four teeth with rotation, affecting the mandibular second premolars, maxillary canines and mandibular canines, in decreasing order. This study observed 21 teeth with rotation in 10 individuals, mainly affecting the mandibular lateral incisor, followed by the mandibular canine, mandibular central incisor, maxillary canine, maxillary central incisor and mandibular first premolar. The high rate of rotated teeth, especially at the mandibular anterior region, may be correlated with lack of space due to the maxillary and mandibular hypoplasia, besides micrognathia, which are common in individuals with TCS [10].

Possibly, the observation of two teeth in ectopic position in two individuals, being one maxillary canine and one mandibular second molar, were also related with lack of space caused by the reduced bone dimensions of these individuals [10].

Some anomalies were not observed at high frequency in this sample, including root dilaceration (n=4), crown shape anomaly (n=2), microdontia (n=1) and mesiodens (n=1), thus these features are probably not strictly related with the syndrome

CONCLUSION

- Dental anomalies are frequent in TCS, with predominance of anomalies of number and position, especially hypodontia and rotation;
- Hypodontia in TCS may or may not be associated with complete cleft lip and palate;
- Most anomalies of position may be related with lack of space, due to the micrognathia observed in individuals with TCS;
- The high prevalence of dental anomalies indicates the need to enroll these individuals in regular programs for dental prevention and follow-up.

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