ORIGINAL PAPER DOI: 10.5935/0946-5448.20160006

International Tinnitus Journal. 2016;20(1):31-35.

Speech language pathology findings in a Treacher Collins syndrome patient

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Abstract

Background: Treacher Collins syndrome (TCS) is a rare disorder with different levels of severity due to congenital head and face abnormalities which affect language, orofacial musculature, voice and breadth, suction, chewing and swallowing functions. **Aims:** This paper objectifies to report a Treacher Collins syndrome patient, describing phonoaudiological changes that can be found in individuals suffering from this syndrome. **Methods & Procedures:** The study was carried out with a male child at a teaching Speech Language Pathology clinic in Southern Brazil. The patient underwent audiological assessment, as well as orofacial motricity, swallowing, voice and speech assessments. **Outcomes & Results:** By means of the assessments, it was possible to perceive that the patient presents moderate hearing loss, inadequacy of phonoarticulatory organs, base of the tongue going backwards and epiglottis collapse over the larynx. In spite of evidencing significant difficulty in producing speech sounds, due to musculoskeletal changes, he effectively interacts in communicative situations, besides understanding sentences and narrated stories. **Conclusions & Implications:** The screened case report evidences that an interdisciplinary follow-up is indispensable for this patient suffering from Treacher Collins syndrome, which implies not only speech language therapeutic follow-up, but also in ear, nose and throat and orthodontics areas.

Keywords: mandibulofacial dysostosis, speech, maxillofacial abnormalities, speech therapy.

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Paper submitted to the RBCMS-SGP (Publishing Management System) on February 25, 2016;
and accepted on March 22, 2016. cod. 226

INTRODUCTION

Treacher Collins syndrome (TCS), also known as Mandibulofacial dysostosis features different levels of severity according to Pollo Medina et al.[1], due to congenital head-face abnormalities[2]. This syndrome was identified and described its essential components in 1900 by British researcher E. Treacher Collins, that is why it takes his name within most literature[3].

From a genetic point of view, TCS is classified as a rare disorder, average incidence of one case out of 55,000 living newborn[4]. About 60% of the cases occur from genetic mutation within the patient, without transmission of the defective gene by their parents, and other 40% of cases comprise autosomal dominant genetic inheritance[5]. The first syndrome-related gene, TCOF1, was only identified in 1996[6]. Mutations in this gene are responsible for about 90% of TCS cases[7]. Over 130 pathogenic mutations have already been identified in TCOF1 gene[5]. In 2011, two other syndrome-related genes were identified: POLR1C and POLR1D[8].

TCS is characterized by hypoplasia of cheek and jaw bones, external ear abnormalities, lower-lid coloboma and absence of lower eyelashes. About 40% to 50% TCS individuals feature conductive hearing loss attributed to the small bones malformation and hypoplasia of the middle ear cavities. Inner ear structures are usually normal. Other not-so-common changes entail cleft palate with or without cleft lip, stenosis or unilateral or bilateral choanal atresia[9].

TCS treatment must be planned according to each patient's specific needs and conducted by a multidisciplinary or interdisciplinary team[10]. In the speech language therapeutics scope, work with patients featuring head and face deformities must be extended from birth until adulthood, implying speech areas, orofacial musculature, voice, breadth, suction, chewing and swallowing functions[10].

According to Cassab et al.[8], clinical speech language therapeutic procedures to patients with TCS are relevant once they present impairments in several functions related to speech language pathological aspects, such as hearing loss and changes in orofacial motricity with significant impairment in feeding and production of speech sounds.

In this regard, Leyva and Malarino Restrepo[11] claim that aspects related to feeding of patients suffering from TCS must be considered, mainly during childhood, once stomatognathic system malformation and its association with obstructive apnea often cause major difficulties for these patients' breathing and swallowing.

Thus, keeping in mind that TCS is a rare condition featuring an array of orofacial anomalies which are studied, assessed, diagnosed and treated under the scope of speech language therapy, the current study aims to report the case of a patient suffering from Treacher Collins syndrome, describing speech language pathological changes that can be observed in individuals with that condition.

PRESENTATION OF THE CLINICAL CASE

The study was carried out at a speech language pathology teaching clinic located in Southern Brazil and accredited by the Unified Health System as delivering high complexity service. The research was approved by the Institutional Ethical Board under number CEP-UTP047/2009, with the participation of a patient identified by the fictitious name João, a boy born on 09/23/2010 and diagnosed with TCS soon after his birth.

João's parents, responsible for the case, when they searched for speech language therapeutic care in May of 2012, were informed about the goals and procedures of this research, thus complying with the free informed consent. Therefore, after agreeing with their child's participation, they signed the Free Consent Form, according to the Resolution 466/2012 of Conselho Nacional de Saúde - National Health Board[12].

Aiming at reporting the case, describing its speech language pathological aspects, semi-structured interviews were carried out with João's parents so that his personal, family and clinical data were unveiled. Being a rare case which demanded interdisciplinary speech language therapeutic care, professionals specifically from audiology, speech, oral motricity and dysphagia areas met in order to proceed a deep and detailed evaluation of the patient, considering all speech language pathological aspects involved.

Thus, a basic audiological exam was performed by means of behavioral observation and free-field pure tone audiometry, besides the analysis of the speech recognition index. Meanwhile, clinical assessments of oral motricity, swallowing, voice and speech were performed.

In the semi-structured interview, João's parents reported that he was born with severe craniofacial anomalies, characteristic of the Treacher Collins syndrome. Among such anomalies, it was observed jawbone hypoplasia, cheek and zigomatic prominences, besides bilateral absence of the ear pavilion, signs which remained until data collection.

His parents also reported that João had three cardiorespiratory arrests soon after his birth, and had to be submitted to an emergency tracheotomy. Moreover, he had to use a nasogastric feeding tube, remaining at an Intensive Care Hospital Unit until two months old. At eighteen months old, he had to undergo a surgical intervention for palate closure, and another gastrostomy for feeding tube replacement. After that, João has already been submitted to more fifteen craniofacial reconstructions,

besides making recurrent use of mandibular advancement device in order to lengthen the jawbone gradually.

As for the family dynamics, João lives with his parents and a brother, two years older than him. His relationship to his parents and brother is good, and João takes part in all the family activities, including going out and visits to friends and relatives. In this sense, his parents state that João is treated like any other children, and although he can't have oral feeding, he takes part in all family meals, sitting at the table with his parents and brother. He plays and communicates by means of gestures, and his parents report that they promote several recreational activities with João and his brother.

João cannot attend a regular school because he makes use of a gastric feeding tube, and tracheostomy to breathe. In spite of being three years of age, therefore he is old enough to attend a school, he needs a health professional to be with him during the school hours. About this specific situation, his mother reported to be seeking help at the Prosecutor's Office in order for her son to be able to attend school regularly with the required professional.

Regarding the audiological assessment, it was held in two distinctive ways. One of them was performed without the hearing aid, while the other was carried out with the bone-anchored hearing aid. The exam performed without the hearing aid detected mixed hearing loss, moderately severe, triton average of 65 dB in both ears. The exam carried out with the adapted hearing aid showed considerable increase in the airways thresholds, triton average of 30 dB in both ears. The free-field test of speech recognition without the hearing aid scored 48%, while the test with the adapted hearing aid scored 100% for a 50 dB intensity.

In the Orofacial Motricity assessment, based on the criteria established by MBGR protocol[13], it was possible to perceive that João features a gothic palate, difficulty in lip sealing, reduction of airflow in the upper airways, asymmetric cheeks and eyes, retrognathic jaw and absence of movements for food placing in the mouth, preparation and chewing.

Swallowing assessment was grounded on the *Functional Oral Intake Scale (*FOIS)[14]. Thus, João was rated Level 2 because he only has liquids and is feeding-tube dependent. In this scale, this level refers to patients making use of an alternative feeding device, and minimum oral food intake[15]. Also regarding swallowing, it is possible to state that the patient features mild to moderate oral dysphagia due to the absence of movements for food placing in the mouth, preparation and chewing, with laryngotracheal protection for liquid and puréed consistencies, without stasis in pharyngeal recesses, according to a dynamic videofluoroscopic swallowing study held in the second semester of 2014.

As for the voice assessment, João, who makes use of a tracheostomy tube, features oral-nasal respiratory mode, hypernasalization associated to pneumophonoarticulatory incoordination. Regarding auditory-perceptual vocal analysis, it was grounded on the parameters established by scales of perceptual vocal evaluation, the *Grade, Roughness, Breathiness, Asteny, Strain, Instability* (GRBASI) Scale[16]. Thus, it was possible to verify that João presents pharyngeal resonance followed by tension in the laryngopharyngeal region, phonatory effort and impaired laryngeal mobility. He also features high *pitch* and lowered *loudness*, maximum phonation time of three seconds to emit the prolonged [a] vowel.

In relation to the acoustic voice analysis, it was used the *VoxMetria* software protocol. However, as João cannot speak, it was not possible to provide a requested sample by the aforementioned software to carry out "voice analysis" option. Thus, the program was used to perform the "vocal quality" analysis, which evidenced that the patient presents fundamental vocal frequency, within children's normal standards, that is, higher than 250 Hz. In relation to *jitter* and *shimmer* values found, they are high, showing that João has irregularities in the variability of the fundamental frequency and short-term wavelength.

The patient's speech evaluation was founded on an interactional and dialogical perspective. From this evaluation, it was possible to perceive that João evidences a significant difficulty in producing speech sounds due to the abnormalities in the musculoskeletal structure of his phonoarticulatory organs, according to what has been formerly mentioned. He produces low-intensity vowel sounds [a, e, E]. When he tries to emit consonant sounds, such as voiceless bilabial [p], voiceless linguadental [t], palate-alveolar [z] and nasal [m], they are atypically distorted in the posterior region of the mouth. Due to this atypical distortion, João produces a similar sound to the voiceless velar [k]. Therefore, when he tries to pronounce words elaborated from articulated sounds in the anterior region of the mouth, as in [pata], for example, he emits [kaka], being the [k] sound frequently produced with a stroke of the glottis.

Thus, the patient pronounces a few words asystematically, such as "mother", "house", and his own name, produced in a distorted way, only understood in the interactive context. He does not elaborate sentences or produces oral accounts. However, he participates effectively in communicative situations, and understands affirmative and interrogative sentences addressed to him as well as personal narrations and stories. In spite of his difficulties of speech, he tries to interact with his listener, making use of facial expressions, hand and body gestures, besides prosodic vocalization of sounds. About these vocalizations, it is deemed to point out that they are systematically reached in a low vocal intensity.

DISCUSSION

There is a reduced number of research studies reporting speech language pathological aspects related to hearing, orofacial motricity, swallowing, voice and speech in the population suffering from TCS[8]. From the search carried out in Cochrane, LILACS, MEDLINE and SciELO databanks using Treacher Collins Syndrome and Speech Language Therapy descriptors, between 2008 and 2014, it enabled to perceive the scarcity of studies focused on speech language clinical procedures for TCS patients, not only in an evaluative point of view, but also in the prolonged follow-up of children and adults' cases. However, in spite of such scarcity, Silva et al.[4] state that children suffering from TCS must be examined early by ear, nose and throat professionals as well as speech language pathologists so that hearing loss and the degree of airways impairment be identified in such children.

In the present study, it was observed that João features mixed bilateral moderately severe hearing loss. This result is compatible with results found in the literature, which report that 80% of patients with TCS present conductive or mixed hearing loss[8,17]. By using the bone-anchored hearing aid, supported by the softband, João's hearing reached normal levels, expanding his possibilities of interaction with other people. This expansion, according to Marsella et al.[18] and Polanski et al.[19] may foster the socialization of patients with TCS, favoring a better interaction with the environment they live in.

As for orofacial motricity and swallowing assessment, the mentioned patient evidences base of the tongue going backwards and epiglottis collapse over the larynx, with reduction of superior airways flow, retrognathic jaw and dysphagia due to the absence of food placing in the mouth, preparation and chewing movements. In this aspect, the screened case is compatible with the study held by Mollinedo Patzi and Quisbert Aquize[20], which refers to respiratory and swallowing problems as recurrent clinical manifestations in patients with TCS. Such problems are caused by micrognathia e glossoptosis, being the latter produced because the tongue is placed further back than normal in the mouth.

In order to correct these manifestations, Mollinedo Patzi and Quisbert Aquize[20] suggest that an evaluation be performed to verify the possibility of correcting the lower third of the face by means of craniofacial surgeries. The current report shows that João has already been submitted to several corrective craniofacial surgeries in order to expand his jawbone gradually, thus improving the functions of the organs involved in breathing, feeding and speech.

Regarding vocal evaluation, it can be pointed out the pharyngeal resonance observed in João's voice. This finding is not in conformity with the majority group identified in a study of Cassab et al.[8], which evidences that 60% of patients with TCS feature hypernasality, 10% feature hyponasality and 30% has normal resonance. As for the respiratory pattern, *pitch*, *loudness* and other aspects of the vocal assessment are compatible with the anatomo-physiological impairment presented by João due to the syndrome described by literature.

Concerning the production of speech sounds, Mollinedo Patzi and Quisbert Aquize's[20] study mentions that problems related to the development of oral language in children with TCSmay be a consequence of hearing loss or difficulties in producing sounds due to structural abnormalities in the phonoarticulatory organs. In this case reported here, it is not possible to relate João's difficulty in speech sound production to his hearing loss because this patient participates in communicative situations not only in family context, but also in other social contexts. He understands sentences and accounts addressed to him. Therefore, his speech difficulty lies specifically on structural anomalies of the organs related to sound articulation.

Patients with TCS are known to have their speech affected by involuntary distortions or compensatory productions due to structural abnormalities[8]. Involuntary distortions are caused by anomalous structure, not by abnormal function, demanding surgery or another structural, physical intervention. In the case presented here, it was already mentioned that João has already undergone several reconstructive craniofacial surgeries, besides making use of mandibular advancement device.

As for the speech development, researchers point out that speech language therapy can be more effective after structural normalization, but it can be recommended before such normalization[8]. However, Colton et al.[21] state that children who need tracheostomy for extended time are particularly challenging for speech language pathologists because speech language development must be objectified without overlooking communicative functional skills.

In the case reported here, in spite of the severe facial deformities, João can establish communicative relationships with family members and other people from his social environment, including the speech language pathology team. His parents invest in his total development and social inclusion, making sure that he participates in all family activities. Thus, the reported case points out the role that a family may perform in social participation, and consequently, in the speech development of children with TCS. In spite of presenting craniofacial anomalies, João appears to be integrated with his environment.

In this sense, besides being concerned with the orofacial aspects involved in breathing, chewing, swallowing and production of speech sounds, the speech

language pathologist should not disregard that patients with TCS can make use of the language, by means of enunciation situations which lead these patients to interact in the social milieu they live in. Despite his difficulty in speaking, João demonstrates that he can communicate with others by means of semiotic expressions, such as gestures, facial expressions and vocalizations, which must be considered in order to contribute to his social interaction.

FINAL CONSIDERATIONS

The reported case states relevant speech language changes in hearing, orofacial motricity, swallowing, voice and speech. It is a case which evidences the need of broad speech language therapy to evaluate and refer to therapeutical procedures. Besides, it points to the ultimate need of an interdisciplinary follow-up for the patient with TCS, which implies not only the speech language pathology, but also the otorhinolaryngological and orthodontic areas; including, the two last ones must collaborate with aspects related to the surgical interventions deemed necessary[22].

Another point to be highlighted in the present study is the patient's willingness to participate in interactions with people around him in spite of being unable to produce speech sounds. It is meaningful that despite the major structural impairment related to his craniofacial anatomy, he is able to interact with his family and other people around him, including the professionals who care for him. It is also worth mentioning that this ability was reported in the semi-structured interview with his parents and confirmed in the speech assessment, showing the extent speech language therapy may invest in the interactional process in TCS cases.

Finally, it is deemed to consider the limitations of this study in terms of the difficulty in generalizing its results. Facing the rarity and complexity of TCS, and also the scarcity of specific literature, it is absolutely necessary to carry out further studies capable of contributing to knowledge production on TCS, underpinning assessment and therapeutic planning of future cases.

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