Otodental syndrome: Case report and differential diagnosis with Treacher Collins syndrome



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Abstract

Background Otodental syndrome and Treacher Collins syndrome are rare diseases that have similar clinical features, which can complicate the diagnostic process. These syndromes cause skeletal and dental abnormalities, the differential diagnosis can be based on clinical signs but only the genetic analysis can confirm it. The aim of this case report is to describe and compare clinical signs of these syndromes.

Case report A 7-year-old patient came to our department: he presented abnormal tooth shapes and sizes, delayed teeth replacement and micrognathia. After extra- and intra-oral examination and radiographic exams, a clinical diagnosis of otodental syndrome was made, and a genetic testing was requested to confirm the diagnosis.

Conclusions Dental management of patients with otodental syndrome is challenging due to agenesis, teeth malformation, lack of space for permanent dentition. Proper treatment decision is crucial to obtain the best result for the patient.

KEYWORDS Otodental syndrome; Treacher Collins syndrome; Globodontia.

Introduction

Otodental syndrome is a rare hereditary disease characterised by globodontia associated with sensorineural, high-frequency hearing loss. Transmission is autosomal dominant, related to the microdelection of chromosome 11q13. It occurs with a prevalence of <1:1,000,000. Patients present pathognomonic morphological anomalies of the teeth, mainly on canines and molars, whereas the incisors are normal. The dental abnormalities of otodental syndrome include congenital missing teeth, globodontia and multiple complex odontoma. Globodontia exhibits characteristic features of fusion teeth. The teeth appear with increased size, spherical crowns, poorly defined grooves and defects of enamel and dentin. There are also frequent alterations of the root and root canals that make endodontic treatments not predictable. Malformations affect both the permanent and deciduous dentition, where they are more pronounced [Bloch-Zupan and Goodman, 2006; Cehreli et al., 2014; Colter and Sedano, 2005; Kim et al., 2016; Liu et al., 2017; Su et al., 2019; Winter, 1983].

Treacher Collins syndrome is a congenital disease of craniofacial development with autosomal dominant inheritance, due to the mutation of the TCOF1 gene (5q32-q33.1). Although it occurs with variable phenotypic expressions, it is characterised by hypoplasia of the zygomatic bones (80% of cases), downward obliquity of the eyelid rims (89%) and presence of coloboma of the lower eyelid (69%). It is considered a rare disease, manifesting with a prevalence of 1:50,000 [Martelli-Junior et al., 2009; Trainor et al., 2009; Trainor, 2010].

Case report

A 7-year-old male patient came to our attention. Written informed consent was obtained from the parents of the patient. The anamnesis reveals a pregnancy with ovodonation, fertilisation with paternal sperm and twin birth without complications. At birth, the baby weighed 2,150 g with a length of 45.5 cm. Cow's milk protein allergy is suspected.

The patient's general state of health is good, the mother reports a slight difference in development between the twins. Psychomotor and cognitive development is regular. Primary teeth replacement is delayed and incomplete. The patient has bilateral mild-to-moderate hearing loss.

On the frontal plane the face appears elongated and asymmetrical with a chin deviation to the right. The middle and upper third are balanced, while on the lateral plane there is a reduction of the lower third (micrognathia). Lip competence is present. The head is slightly tilted (Fig. 1, 2, 3). The eyelid rims are tilted downwards and outwards (slanting), the interpupillary distance is normal and the patient shows no signs of iris or pupillary coloboma. The ophthalmologic examination did not show any vision abnormalities.

Deciduous central and lateral incisors in the upper arch are slightly discoloured, there is a delay in dental replacement in the anterior area. Bilateral agenesis of the deciduous canines is present although the permanent canine are touchable under the gums. Distally there are four deciduous molar-like teeth with disproportionate and irregular crowns (Fig. 4, 5, 6). The presence of abundant plaque and dental malposition causes erythematous areas and gingivitis. The lateral frenula have a

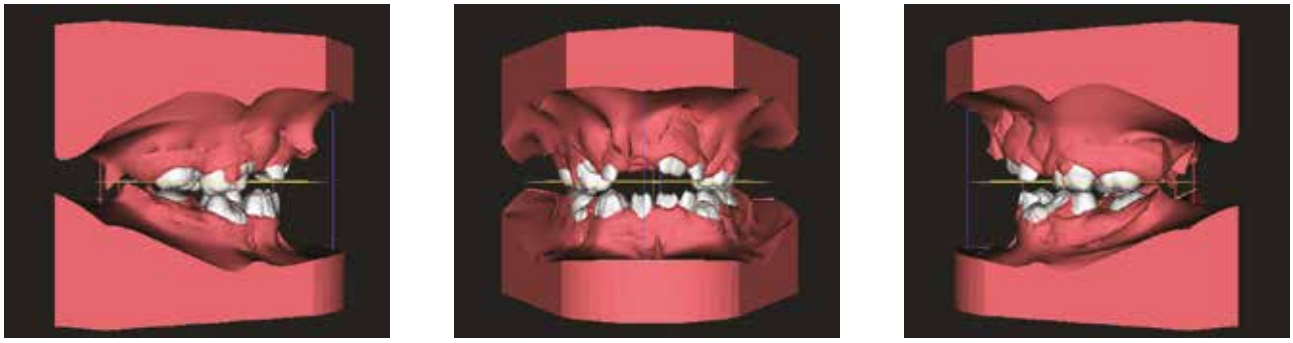
very low insertion, while the central one does not interfere with the incisors. The palate is narrow and ogival. The lower arch appears very similar to the upper one (Fig. 7, 8, 9, 10, 11). Globodontia affects molars and canines, the left canine is semi-included. There are only two incisors. The tongue's mobility appears to be reduced, because of the short frenulum which does not compromise its extrusion, but only its elevation. This anomaly is also found in the twin brother. The central and lateral deciduous incisors are present in the upper arch and the permanent counterparts can be seen on the orthopanoramic x-ray (Fig. 12). Small buds referring to first premolars can be observed, while the buds of the second premolar and of the first molar are not visible. In the upper arch, there are erupted teeth similar to molars with large cubic crowns. The buds of the second and third molars are present. In the lower arch, there is a substantial anterior crowding; the lateral incisors are included



FIG. 1, 2, 3 Patient's frontal and side view.



FIG. 4, 5, 6 Intra-oral photos.



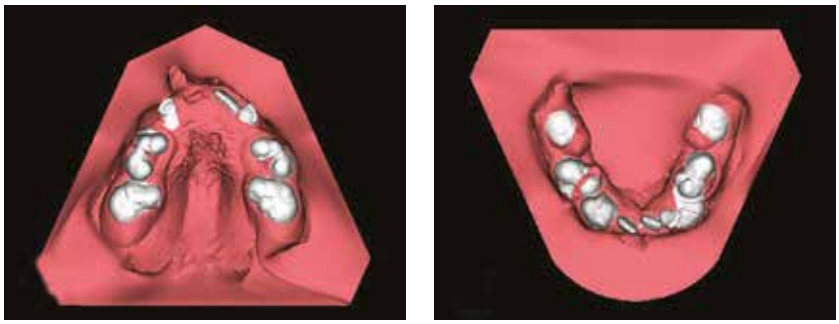


FIG. 7, 8, 9, 10, 11 Digital models of the arches.



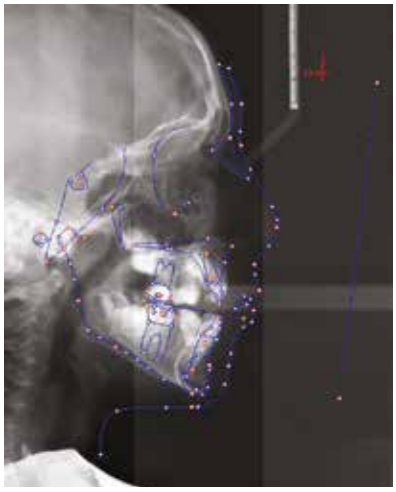


FIG. 12
Orthopantomography.

FIG.13, 14 Lateral teleradiography and cephalometric analysis.

G01 - Classe Scheletrica		A.Fac.Post / A.fac.Ant	57,2 %
SNA	71 °	F M A	40 °
SNB	68,2 °	G04 - Rapporti Dentali	
ANB	2,7 °	S^N-S	87,7 °
Wits	-1,1 mm	Angolo IS - Piano Palatale	105 °
A^N^Pg	2,6 °	B1 - A-Pog	0,2 mm
Convessità (A-NPog)	2,3 mm	IMPA	83,9 °
G02 - Divergenza Scheletrica		Ang. Interincisivo (A1 - B1 angle)	140,1 °
S-N^sna-snp	17,3 °	Overjet (B1-A1 Horiz.)	-0,4 mm
sna-snp^Go-Gn	28,7 °	Overbite (B1-A1 Vert.)	-5,6 mm
S-N^Go-Gn	46 °	G05 - Analisi Estetica	
Altezza Facciale Totale (P^Go-Me)	39,9 °	Labbro Sup (UL int - ULA)	6,6 mm
G03 - Crescita Scheletrica		Labbro Inf (LL int - LLA)	0,5 mm
Metà Sup. Ang. Goniaco (Ar-Goc-N)	52,2 °	Protrusione Labiale (LL-PrPog')	1,8 mm
Metà Inf. Ang. Goniaco (Me-Goc-N)	85,3 °	Pogonion Cutaneo (Pog' - TVL)	-0,5 mm
Angolo Goniaco (Ar-Goc-Me)	137,5 °	Angolo Nasolabiale	132,9 °
Angolo della Sella (N-S-Ar)	120,6 °		
Angolo Articolare (S-Ar-Goc)	150,2 °		
Somma 1-2-3	408,3 °		
Base Cranica Anteriore (S-N)	64,3 mm		
Lun. del Corpo Mandibolare (Goc-Me)	50,8 mm		
Base Cranica Posteriore (S-Ar)	28,6 mm		
Altezza Ramo Mandibolare (Ar-Goc)	35,2 mm		
S-Ar/Ar-Go	0,8 mm		
Altezza Facciale Posteriore	61,7 mm		
Altezza Facciale Anteriore	107,8 mm		

due to lack of adequate space for the eruption. The buds of the premolars and of the first molars are absent bilaterally. The buds of the second and third molars are present distally. Cephalometric analysis shows a skeletal Class I in a hyperdivergent, biretrused patient with growth pattern of clockwise mandibular rotation (Fig. 13, 14).

Discussion

The patient shows signs ascribable both to otodental syndrome and Treacher Collins syndrome. The analysis of the data collected allows to hypothesise a clinical diagnosis but only the genetic analysis can confirm it.
In the otodental syndrome, variable facial dysmorphisms have been described. The patient's poor zygomatic three-dimensionality is also found in his father and, for this reason it would not be appropriate to speak about zygomatic hypoplasia. There is down-slanting of the eyelid rims but absence of ocular coloboma, a typical sign of Treacher Collins syndrome but reported even in otodental syndrome. The patient has multiple agenesis and delayed eruption common in both syndromes but the globodontia of the molars and canines is pathognomonic of otodental syndrome. This clinical condition requires a regular follow-up and prevention program to maintain oral hygiene. A delay in dental eruption has been documented in patients with otodental syndrome, Ankyloglossia was not treated although a possible frenectomy surgery is to be considered in the future.
Both diseases are characterised by hearing disorders. The hearing tests show bilateral peripheral hearing loss, more evident on the left, with stimulations of intensity 80Hz, 90Hz and 95Hz. The data collected show high variation so they cannot be taken into consideration as a diagnostic criteria.
The paediatric visit does not report postural problem or limitation, despite the inclination of the head. Excluding vertebral alterations is important, considering that they are a determining criteria in the diagnosis of Treacher Collins syndrome.

Conclusion

Although not completely exhaustive, the semeiotics and the comparison with the case reports in the literature suggest a case of otodental syndrome. Diagnosis has to be confirmed by a genetic analysis. From the oral point of view we decided to follow the child with periodic controls to evaluate his overall growth, dental replacement and oral hygiene. Proper ophthalmic and hearing follow-up are equally necessary to avoid the onset of complications during growth.

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