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Otodental Syndrome

M. Paglia[1], M. Severino[2], R. Gatto[3], G. Giani[4], S. Caruso[5]

[1] Department of Paediatric Dentistry, Istituto Stomatologico Italiano, Milano, Italy
[2] Researcher Department of Medicine and Surgery University of Perugia, Perugia, Italy
[3] Ordinary Professor in Paediatric Dentistry, Department of Life, Health and Enviromental Science, Paediatric Dentistry, University of L'Aquila, L'Aquila, Italy
[4] Private Practice in Busto Arsizio, Varese, Italy
[5] Department of Life, Health and Enviromental Science, Paediatric Dentistry, University of L'Aquila, L'Aquila Italy

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Abstract

Otodental syndrome is a rare autosomal dominant condition characterised by a dental phenotype known as globodontia often associated with high-frequency hearing loss. Globodontia occurs both in the decidous and permanent dentition and affects canine and molar teeth.

KEYWORDS Otodental syndrome, globodontia, sensorineural hearing loss

Introduction

Otodental syndrome (OMIM 166750) is a rare autosomal dominant condition characterised by grossly enlarged globe-shaped canine and molar teeth (globodontia) associated with sensorineural high-frequency hearing loss [Su et al, 2019; Witkpo et al1976].

Globondontia has characteristic features of tooth fusion: abnormal bulbous enlargment of the crown, which appears spherical and with poorly defined grooves. Molars display taurodontism (inverted crown/body ratio), there are also frequent alteration of roots and pulp canal, that makes the endodontic treatment not predictable [Agnès Bloch-Zupan et al., 2006].

Besides globodontia, other dental abnormalities of otodental syndrome are congenital missing teeth, multiple complex odontoma and hypoplasia (enamel defect). The latter is frequently found on the buccal side of the canines. Hystopathological findings showed in the area of hypoplastic enamel: slightly reduced enamel thinckness, irregular incremental lines and rod sheath area cointaining voids, defects very similar to those observed in enamel hypomaturation [Agnès Bloch-Zupan et al., 2006].

Odontoma occurs mainly in children and young adults, especially during the second decades of life. Complex odontoma tends to occur in the posterior part of the jaws and consist in disorganised masses of hard and soft dental tissues with no morphological resemblance to normal teeth [Ji-mei Su et al., 2019]. Many authors reported dismorphic facial features including long face, full cheeck, anteverted nostrils, long philtrum and a cleft lip in some patients with otodental syndrome [Witkpo et al., 1976; Santos Pinto L et al., 1998].

Bilateral sensorineural hearing deficiency to about 65 dB is found at all frequencies but is more pronounced at about 1000 Hz in these patients. The age of onset of hearing loss ranges from early childhood to middle age, and the hearing loss is progressive and bilateral [Cook et al., 1975]. Mutations in GJB2, GJB3, SLC26A4 and mtDNA genes are reported as a prominent cause of congenital sensorineural hearing loss [Qing et al., 2015; Fang, et al., 2015].

Transmission is autosomal dominant, related to microdeletion of chromosome 11q13. The prevalence of otodental syndrome is < 1:1,000,000 [Kim et al., 2016].

Otodental syndrome show variable expressivity: dental abnormalities and hearing loss usually coexist. However some patients develop only sensorineural hearing loss, while others show dental abnormalities without hering loss [Manpreet et al., 2010, Sedano et al., 2001]. Genetic counselling is important; while the inheritance is autosomal dominant there is a variable expression. and genetic or enviromental factors may influence disease severity and could explain the different synthomps in the affected patient [Vieira et al., 2002].

Otodental syndrome has been described under various name: otodental dysplasia [Chen et al., 1988; Levin et al., 1974], familial otodentodysplasia [Levin et al., 1972], globodontia [Gundlach et al., 1977; Witkpo et al., 1976]; in some families, an associated ocular phenotype was recognised and therefore named as Oculo-Oto-Dental Syndrome (OOD) [Vieira et al., 2002].

Otodental syndrome has been described in at least 12 families: the first case was described in a Hungarian mother and her son [Denes and Csibain, 1969].

Differential diagnosis

In the case of otodental syndrome the dental phenotype is per se diagnostic, but the association of sensorineural hearing loss, dental anomalies and variable facial dysmorphism can be found in other syndromes:

- Autosomal recessive sensorineural hearing impairment, dizzines and hypodontia [Lee K Et al., 1995]
- Bilateral sensorineural hearing loss and multiple anterior dens invaginatus [Gorlin RJ et al., 2001]
- Kantaputra and Gorlin [1992] described the molarisation and premolarisation of anterior teeth in double dens invaginatus in an female who had developmental delay and congenital progressive sensorineural hearing loss. In addition, multituberculated mandibular incisors, canines and first premolar were observed.

Clinical description-case report

A 7-year-old boy come to our attention. The medical history reveals pregnancy with ovodonation, fertilisation with paternal sperm and twin birth without complications. The patients general health was good: psychomotor and cognitive development were normal, the mother reported a slight difference in devolopment between the twins, his twin brother did not exhibited signs of dental abnormalities or hearing loss [Paglia M et al., 2022]. The patient came for follow-up in our

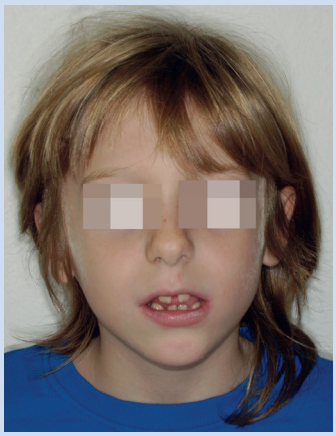


FIG. 1

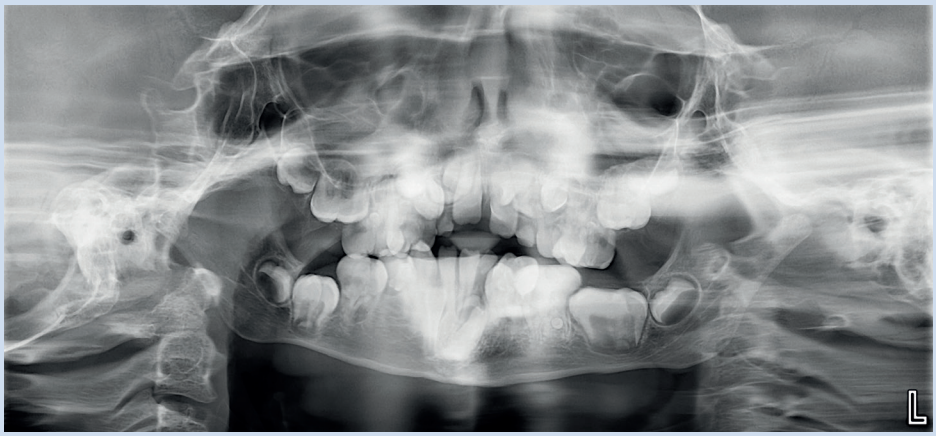


FIG. 2

clinic every four months, he is followed for oral hygiene and undergoes regular dental checkups for decay due to the abnormal anatomy of his teeth, enamel hypoplasia and poor oral hygiene.

Sensorineaural high-frequancy hearing loss

The patient has bilateral mild to moderate hearing loss. The performed hearing test showed bilateral peripheral hearing loss, more evident on the left, with stimulations of intensity 80Hz, 90Hz and 95 Hz..

Facial examinations: eye phenotype

On the frontal plane the face appears elongated and asymmetrical with chin deviation to the rigth. 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).

There is a down-slanting of the eyelid rims, the interpupillar distance is normal and the patients shows no signs of iris or pupillary coloboma. The ophthalmologic examination did not show any vision abnormalities.

Oral and dental anomalies

The mother referred that primary teeth dentition was delayed and the decidous teeth exfoliated later than average. The panoramic x-ray shows the dental status at the time of his first visit (Fig. 2).

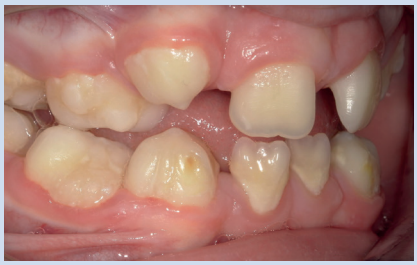
New photos were taken at his latest follow-up to follow the eruption, when the patient was 9 years old (Figs. 3, 4, 5,6, 7).

In the arch are present the upper and lower central incisors - which are are slightly discoloured - and permanent-like canines, teeth n.13, 33 and 43: the canine draft of tooth 23 can be palpated under the gum.

The premolars areas were occupied by macrodontic teeth: deciduous molars-like teeth with disproportionate and irregular large cubic crowns, globodontic teeth with almost not discernable cups or grooves (Figs. 8 and 9).

The presence of abundant plaque and dental malposition causes erythematous areas and gingivitis, the enalmen is hypoplastic and with yellow-brown spots; the deep fissures on bulbous molars are susceptible to dental caries.

The upper and lower lateral incisors are not present. The palate is narrow and ogival. The lower arch is very similar to the upper one.



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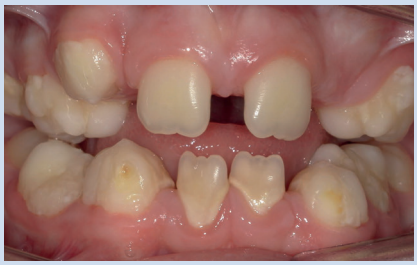

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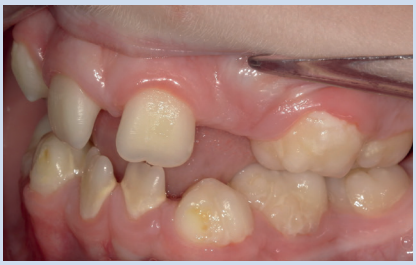

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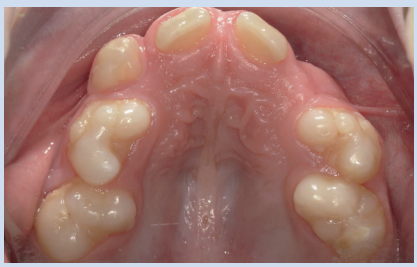

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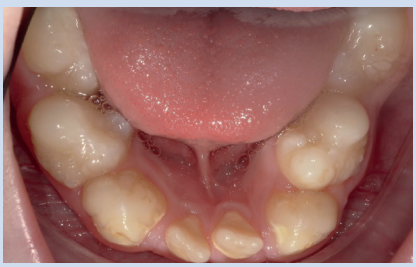

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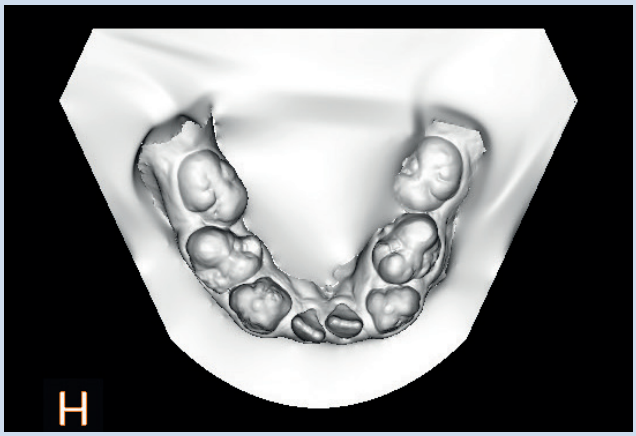
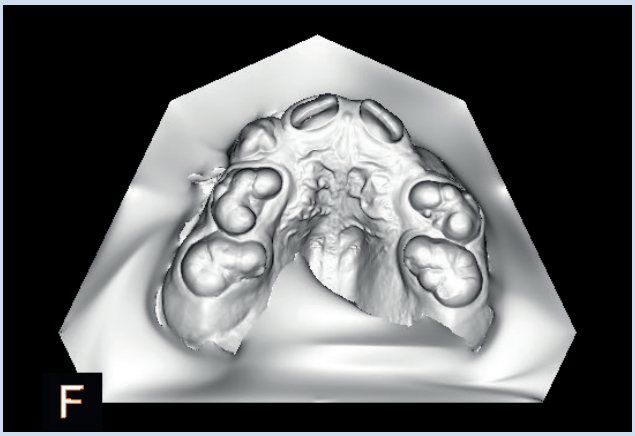


FIG. 8



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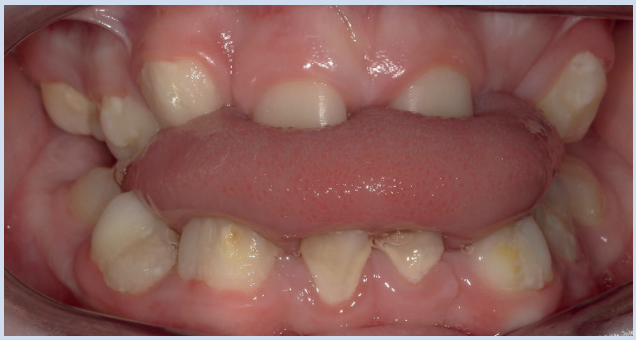
FIG. 10

Tongue's mobility appears to be reduced because of the short frenulum, which does affects its extrusion, but only its elevation; there is atypical swallowing, an anomaly is also found in the twin brother (Figs. 7 and 10).

Conclusion

Globondontia is the diagnostic feature of otodental patient, but in this syndrome are often present other dental problems as enamel hypoplasia, odontoma and delayed eruption.

Is important to study this syndrome to make dentist more familiar with the diagnosis and the managment of this rare patient. The shape that characterized the teeth and the area of enamel hypoplasia make this teeth susceptible to decay: oral hygiene and preventive procedures (topical fluoride and sealants) are really important. If needed endodontic treatment could be really challeging and complex due to the abnormal morphology of the teeth. Some authors reported the propensity of globodontic teeth to develop endodontic -periodontic lesions. (Mesaros et al)

Dental managment is complex, interdisciplinary and must include regular follow up.

Hearing check is mandatory, as well as eye examinations.

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