Otodental syndrome: a case presentation in a 6-year old child

ABSTRACT

Background Otodental syndrome is a rare condition characterised by globodontia, and sensorineural high frequency hearing loss. To date, only 20 cases of otodental syndrome have been reported.

Case report A 6-year-old girl presented with a chief complaint of delay in the eruption of primary canines. Following clinical, radiographic and audiologic evaluations, the patient was diagnosed with otodental syndrome.

Conclusion Globodontia is a diagnostic feature of the otodental syndrome, which often provides the path to discovery of the associated hearing loss. Missing teeth, arch-size discrepancies, chewing problems and teething disturbances are the other major complications.

Keywords Globe-shaped teeth; Globodontia; Hearing loss; Otodental syndrome.

Introduction

Otodental syndrome is a rare anomaly, inherited on an autosomal dominant basis [Hennekam et al., 2010]. The condition has also been reported under various names, including familial otodentodysplasia [Toledo et al., 1971] and otodental dysplasia. [Levin et al., 1975; Chen et al., 1988]. Otodental syndrome is characterised by globodontia [Witkop et al., 1976] and sensorineural high frequency hearing loss [Hennekam et al., 2010]. Globodontia is a striking dental phenotype, characterised by abnormal bulbous enlargement of tooth crown with almost no discernable cusps, which is both pathognomonic and diagnostic of the otodental syndrome [Hennekam et al., 2010]. Globodontia occurs in both primary and permanent dentition, affecting canine and posterior teeth [Witkop et al., 1976]. Primary and permanent incisors are not affected and display normal shape and size.

After Winter [1983] reported an associated ocular trait, the condition was later named oculo-oto-dental syndrome (OOD) by Vierra et al. [2002]. However, that particular family which exhibited ocular coloboma is considered as a different condition, since no other case in the literature has presented the same ocular trait [Hennekam et al., 2010]. Gregory-Evans et al. [2007] evaluated 3 families with globodontia, one being the original case presented by Winter [1983]. Following molecular tests, the authors suggested that FGF3 haploinsufficiency is likely the cause of otodental syndrome and that FADD haploinsufficiency accounts for the associated ocular coloboma [Gregory-Evans et al., 2007].

Other reported dental findings of otodental syndrome include localised enamel defects (hypoplasia) on canines, missing or microodontic premolars, odontomas and conically-shaped supernumerary teeth [Hennekam et al., 2010; Bloch-Zupan et al., 2006]. Delayed eruption is a frequent finding in both primary and permanent dentitions [Bloch-Zupan et al., 2006].

In otodental syndrome, bilateral sensorineural hearing deficiency to about 65dB is found at all frequencies but is more pronounced at about 1000 Hz. [Hennekam et al., 2010]. The age of onset of the hearing loss ranges from early childhood to middle age [Cook et al., 1981; Jorgenson et al., 1975; Colter et al., 2005]. Regardless of the age of onset, hearing loss is progressive but it usually plateaus by the fourth decade [Cook et al., 1981].

The present case report describes the clinical features of otodental syndrome in a 6-year-old girl.

Case report

A 6-year-old girl was referred to the Paediatric Dentistry Department with a chief complaint of delay in the eruption of her primary canines. According to the hospital records, the child had experienced an emergency intervention at the age of 4 due to a piece of cake lodged in her throat. The cake was uneventfully removed with a nasogastric tube, but thereafter the child consistently reported difficulty in chewing.

Intraoral examination showed that the maxillary and
mandibular incisors were normal in size, shape and color (Fig. 1). However, the crowns of the canines and molars were large and bulbous in shape, indicating globodontia (Fig. 1, 2, 3). All teeth were caries-free, but small, yellow hypoplastic lesions were present on the labial enamel of the primary canines. On radiographic examination, the primary molars had a striking radiographic appearance of double pulp chambers (Fig. 2). The premolar buds could not be detected in either jaw. The patient was scheduled for consultation at the Children’s hospital, where detailed physical examination and hearing tests were later carried out. According to the ENT examination, she had bilateral hearing loss up to 85 dB. Genetic investigation of family members was rejected by the father, who also had hearing loss. However, based on physical examination, the department of genetics did not report any facial dysmorphism.

Following topical fluoride varnish application and hygiene instructions, the patient was scheduled for semiannual control visits. After 3 years, a panoramic radiograph (Fig. 5) revealed that the buds of the permanent first premolars and second molars had appeared in both jaws. Since the posterior teeth were not in proper occlusion, the patient still refrained from chewing fruits like cherry or soft cakes, which she reportedly aspirated on a frequent basis. The patient is still attending regular checkup visits.

Discussion and conclusion

Tooth, ear and eye development are all under strict genetic control as revealed by gene mutations that are associated with arrested tooth development [Thesleff, 2000], deafness [Petit, 2006], and coloboma [Gregory-Evans et al., 2004]. Otodental syndrome (MIM 166750) is an autosomal dominant condition of unknown cause. Colter and Sedano [1981] suggested that 6 genes which are associated with determining location of tooth germ development and tooth morphology could be involved in the syndrome [Sarkar and Sharpe, 2000; Sedano et al., 2001; Neubuser et al., 1997]. These genes include bone morphogenetic protein 4 (BMP4), muscle segment homeo box 1 (MSXI), fibroblast growth factor 8 (FGF8), homeo box gene-branchial arch neural crest 1 (BARX1), and distal-less homeo box 1 and 2 (DLX1 and DLX2) [Colter and Sedano, 1981]. In a British family described by Winter et al. [1983], the locus for Oculo-Oto-Dental syndrome (OOD) was mapped to 20q13.1 within a 12-cM critical chromosomal region [Vieira et al., 2002]. However, the otodental syndrome is found to be mapped in 11q13 [Gregory-Evans et al., 2007]. The association of sensorineural hearing loss and dental anomalies can also be found in conditions such as missing teeth [Lee et al., 1978] and multiple anterior dens invaginatus combined with fusion [Gorlin.
et al., 2001]. Sensorineural hearing loss has also been associated with oculo-facio-cardio-dental syndrome [Aalfs et al., 1995]. Kantaputra and Gorlin [1992] described double dens invaginatus, multituberculated mandibular incisors, canines, and first premolars in a female patient with developmental delay and congenital progressive sensorineural hearing loss. Dentinogenesis imperfecta has also been associated with progressive sensorineural high frequency hearing loss [Xiao et al., 2001; Cauwels et al., 2005].

Witkop et al. [1976] reported facial characteristics of the otodental syndrome including long facies, anteverted nostrils, a long philtrum and a full cheek appearance. The present case did not represent any facial dysmorphism. A comparison of the clinical features of the present case with those of previously reported findings also fail to show coloboma of the eye, odontomas, microodontic or conic-shaped teeth and intrapulpal calcifications. However, there were yellow hypoplastic lesions on the labial surfaces of primary canines, as demonstrated by Vieira et al. [2002].

In odontal syndrome, both the enamel hypoplasia and the deep fissures on bulbar molars are susceptible to dental caries. In an affected patient, Mesaros et al. [1996] reported a permanent tooth which needed endodontic treatment from what might have initially been occlusal pit and fissure caries. The authors emphasised the challenges of endodontic treatment and propensity of globodontic teeth to develop endodontic-periodontal lesions due to their aberrant coronal and pulpal morphology. Thus, preventive procedures (i.e., topical fluorides, sealants) should be carried out on a regular basis in patients with otodental syndrome.

References


FIG. 5 Panoramic radiograph of the patient at 3 years follow-up.