Otodental Syndrome: A Case Report

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Abstract

The purpose of this article is to describe the clinical features of otodental syndrome. A 9-year-old boy presented with dental abnormalities that have been described for otodental syndrome. The characteristic findings included large bulbous crowns in canine and molar teeth of both dentitions, deep vertical enamel fissures separating the cusps of affected molars, and hypoplastic yellow areas on the labial surfaces of the canines. Radiographs revealed the abnormal molars to possibly be the product of fusion of multiple tooth buds. The pulp chambers appeared to be duplicated, and possibly a supernumerary tooth or complex odontoma is present. (Pediatr Dent 2005;27:482-485)

KEYWORDS: BULBOUS CROWNS, GLOBODONTIA, OTODENTAL DYSPLASIA, SENSORINEURAL HEARING LOSS, HYPOPLASIA

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Otodental syndrome is characterized by abnormalities of dental crown morphology as well as other dental findings and sensorineural hearing loss. The dental findings present in the otodental syndrome were first described in a mother and her son by Dénes and Csiba[1] of Hungary in 1969. In 1971, Toledo et al[2] found similar abnormal tooth morphology in two brothers and a sister. The alteration in tooth morphology described in both articles was multiple globular shaped teeth.

Levin and Jorgenson[3] in 1972 described, in depth, 28 members of an Italian family who presented with abnormal tooth morphology and sensorineural hearing loss typical of familial otodentodysplasia, as it was termed in this study. This syndrome was later named otodental dysplasia in 1974 by Levin and Jorgenson.[4] Then, in 1976, Witkop et al[5] named the characteristically abnormal tooth morphology as globodontia and also proposed the name otodental syndrome. In 2001, Sedano et al[6] proposed some possible genetic mechanism that might help in understanding the original defect in patients with this syndrome.

Otodental syndrome has been established in several articles to be of autosomal dominant inheritance with variable penetrance and expressivity.[6-10] The sensorineural hearing loss associated with this syndrome is typically presented as a loss of frequencies above 1,000 Hz.[6] This hearing loss can start as early as 2 or 3 years of age for some, while in others it may not manifest until after puberty. Due to the variable expressivity of the syndrome, not all patients present with hearing loss.

Several dental findings are associated with this syndrome. Maxillary and mandibular primary and permanent incisors are of normal shape and size. The crowns of canines and molars, however, are characteristically large and bulbous in both dentitions. Primary and permanent canines present hypoplastic yellow areas, particularly on the labial surfaces. The cusps of the affected molars are separated by deep vertical enamel fissures. The abnormal molars could be the result of fusion of multiple molar and premolar tooth buds. The pulp chambers of the affected molars appear to be duplicated. Other dental findings in this syndrome include absent or microdontic premolars, conical supernumerary teeth, and odontomas.

This purpose of this case report was to describe the clinical findings in a 9-year-old male patient with otodental syndrome.

Case report

The patient was a 9-year-old Caucasian male referred by a local pediatric dentist to the Children's Dental Clinic at the University of California Los Angeles, because of the presence of abnormal looking teeth. He was asymptomatic, with a history of an extraction of his maxillary left primary second molar secondary to an ectopic eruption of his maxillary left permanent first molar. The patient's medical history was unremarkable. He appeared well developed and well nourished (Figure 1). His hearing was not tested by an otologist because the parents preferred to postpone the

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procedure, but the patient responded to the normal range of the human voice. The patient's mother and grandmother were not aware of any other family members with similar oral or auditory findings.

The patient's maxillary and mandibular incisors appeared unaffected. The canine and molar crowns were large and bulbous or spherical in appearance in both the primary and permanent dentitions (Figures 2 and 3). Deep vertical enamel fissures separated the cusps of the affected primary and permanent molars, resulting in 6 to 8 cusps on each tooth. Hypoplastic yellow areas were present in the enamel on the labial surfaces of the primary canines, which also demonstrated both carious and nonassociated carious lesions (Figures 4 and 5).

Radiographs suggested that the abnormal molars could be the result of fusion of multiple molar and premolar tooth buds, and the pulp chambers appeared to be duplicated (Figure 6). The panoramic radiograph suggested the possibility of a supernumerary tooth or a complex odontoma in the maxillary right posterior quadrant (Figure 7). The development of the succedaeous teeth suggested that the premolars were of normal shape, but slightly smaller in size, whereas the permanent canines appeared to be slightly enlarged. Although the permanent teeth appeared to be developing normally, the radiographs revealed that this patient is congenitally missing a maxillary left premolar and possibly his third molars.

As this was his first presentation to the author's clinic at 9 years of age, the development of his dentition and eruption pattern prior to this was unknown. Several studies have noted the eruption of affected primary canines and molars is often delayed until after 2 years of age.[4,5,8,9] Eruption of the permanent posterior teeth may also be delayed,[4,9] as it

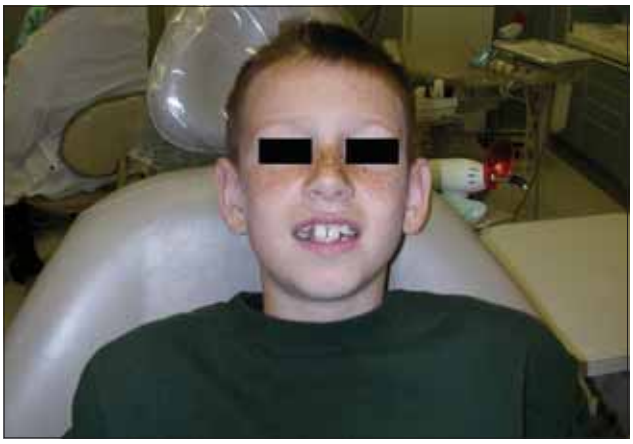
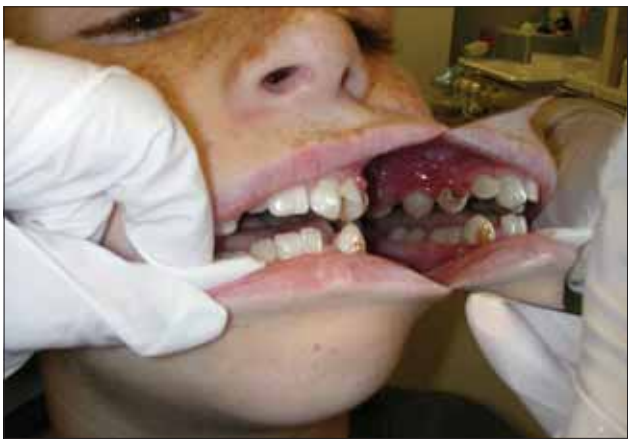


Figure 1.



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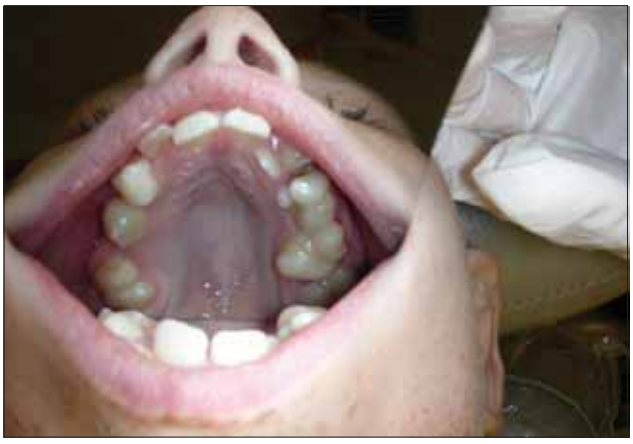

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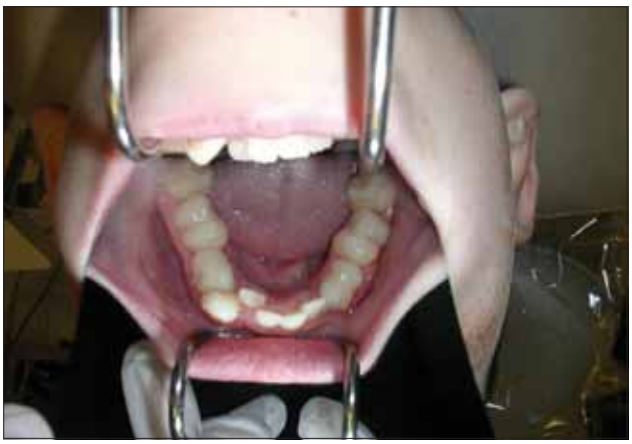

Figure 4.

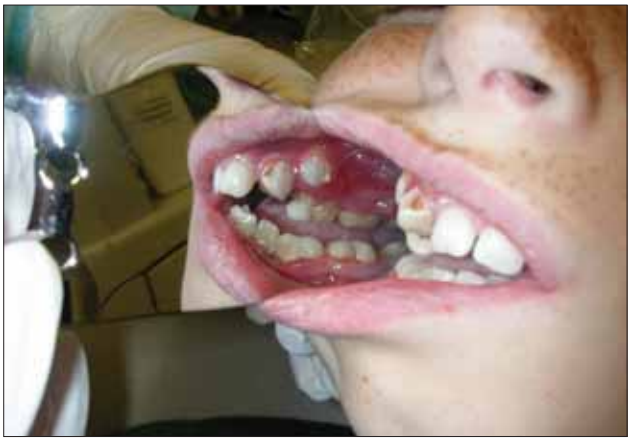

Figure 5.

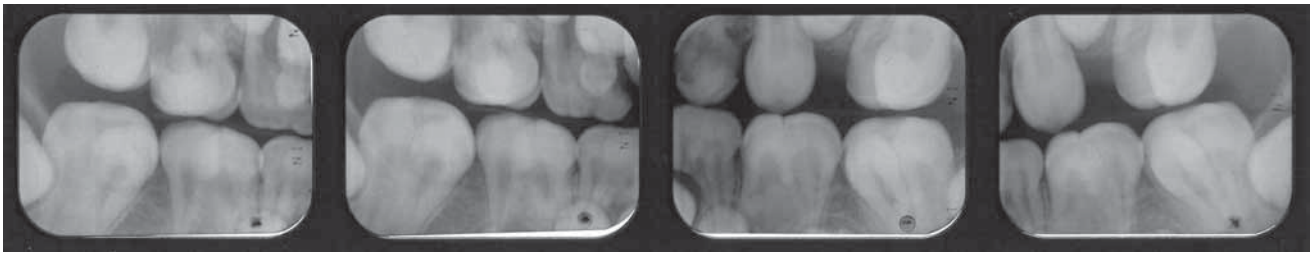

Figure 6.

appeared to be in this case. The patient presented with severe crowding in the maxillary and mandibular arches, particularly in the anterior areas. The anterior crowding could be due to the presence of the globular shaped posterior teeth, whereas the posterior crowding that was present was due to space loss. His occlusion also presented as an Angle class III relationship.

A comprehensive treatment plan was developed to include restorative therapy to treat the carious lesions, as well as a preventive program. A referral was also given to this patient for a hearing evaluation. As of the writing of this paper, however, both have yet to be completed.

Discussion

Otodental syndrome is characterized by dental abnormalities and sensorineural high-frequency hearing loss. It is inherited as autosomal dominant with variable penetrance and expressivity. Variable expressivity means that some family members only exhibit dental anomalies or hearing loss.[6] The most important dental abnormality is the globed-shaped or spherical appearance of the primary and permanent posterior teeth. Sensorineural high-frequency hearing loss is the second major abnormality of this syndrome. The onset of hearing loss usually occurs early in life, in the first or second decade. It is characterized by an abnormally raised hearing threshold, usually greater than 25 dB in one or both ears, in the high frequency region (3,000 to 6,000 Hz).[10]

The precise cause of otodental syndrome is unknown. There are 6 genes that could possibly be involved. These genes are associated with determining location of tooth germ development and tooth morphology.[6,14-20] These genes include bone morphogenetic protein 4 (BMP4), muscle segment homeo box 1 (MSX1), fibroblast growth factor 8 (FGF8), homeo box gene-branchial arch neural crest 1 (BARX1), distal-less homeo box 1 and 2 (DLX1 and DLX2).[6,14-20]

BMP4 and MSX1 are active in the incisor region and are assumed to be functioning within normal parameters, since patients with otodental syndrome have normal incisors. Therefore, the cause most possibly lies in either mutations or abnormal signaling responses by FGF8, BARX1, DLX1, and DLX2, which exert their influence in the molar region. It has also been suggested that, with audiometric testing, the site of the ear lesion is the cochlea.[13] Interestingly, MSX1 and BMP4 participate in the embryogenesis of the ear.[6] BMP4 also has a role in the development of hair cells and sensorineural cells of the inner ear.[6,21-24] Although the cause of this syndrome is unknown and beyond the scope of this report, it will be exciting to continue to investigate, and understand more about this very rare syndrome.

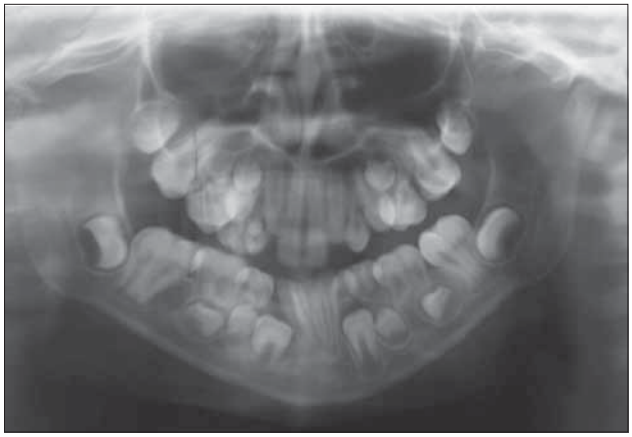

Figure 7.

The patient's dental history presented in this case reveals some possible dental disorders or complications with a case of otodental syndrome. The patient presented with several carious lesions needing to be restored. Oral hygiene, as seen in the figures, is fair at best. Therefore, a preventive program is mandatory. Most of the carious lesions can be restored, as with a normal tooth. Endodontic therapy, however, could be predicted to be quite complex, due to the duplicated pulp canals in the affected posterior teeth. Mesaros and Basden[12] presented a case in which the complexity of endodontic treatment in a tooth with globodontia was demonstrated, and indicated the propensity of these teeth to develop endodontic-periodontic lesions, possibly due to their aberrant coronal and pulpal morphology.

In conclusion, the dental abnormalities associated with otodental syndrome have been presented in this young boy. The characteristic finding of large bulbous crowns of the canine and molar teeth in both dentitions is the most prominent feature of this autosomal dominant syndrome with variable expressivity. Sensorineural hearing loss is a variable finding associated with otodental syndrome.

Acknowledgements

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References

1. Dénes J, Csiba A. An unusual case of hereditary developmental anomalies of the cuspids and molars. Fogorv Sz 1969;62:208-212.
2. Toledo OA de, Bausells HI, Vono RM, Rocca RA. Anomalia dental múltipla em três irmãos–Caso Clínico. [Multiple dental anomaly in three brothers. Case report.] Rev Fac Farm Odontol Araraquara 1971;5:207-214.
3. Levin LS, Jorgenson RJ. Familial otodentodysplasia: A "new" syndrome. American Society of Human Genetics, Annual Meeting, October 11-14, Philadelphia, Pa [abstract] 1972;24:61a.
4. Levin LS, Jorgenson RJ. Otodental dysplasia: A previously undescribed syndrome. Birth Defects Orig Artic Ser 1974;10:310-312.
5. Witkop CJ Jr, Gundlach KK, Streed WJ, Sauk JJ Jr. Globodontia in the otodental syndrome. Oral Surg Oral Med Oral Pathol 1976;41:472-483.
6. Sedano HO, Moreira A, Alvez R, Moleri A. Otodental syndrome: A case report and genetic considerations. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2001;92:312-317.
7. Stewart DJ, Kinirons MJ. Globodontia: A rarely reported dental anomaly. Br Dent J 1982;152:287-288.
8. Levin LS, Jorgenson RJ, Cook RA. Otodental dysplasia: A "new" ectodermal dysplasia. Clin Genet 1975;8:136-144.
9. Chen RJ, Chen HS, Lin LM, Lin CC, Jorgenson R. "Otodental" dysplasia. Oral Surg 1988;66:353-358.
10. Van Doorne L, Wackens G, De Maeseneer M, Deron P. Otodental syndrome: A case report. Int J Oral Maxillofac Surg 1998;27:121-124.
11. Santos-Pinto L, del Pilar Oviedo M, Santos-Pinto A, Lost HI, Seale NS, Reddy AK. Otodental syndrome: Three familial case reports. Pediatr Dent 1998;20: 208-211.
12. Mesaros AJ Jr, Basden JW. Otodental syndrome. Gen Dent 1996;44:427-429.
13. Cook RA, Cox JR, Jorgenson RJ. Otodental dysplasia: A five-year study. Ear Hear 1981;2:90-94.
14. Sarkar L, Sharpe PT. Inhibition of Wnt signaling by exogenous Mfrzb 1 protein affects molar tooth size. J Dent Res 2000;79:920-925.
15. Zhang Y, Zhang Z, Zhao X, et al. A new function of BMP4: Dual role for BMP4 in regulation of sonic hedgehog expression in the mouse tooth germ. Development 2000;127:1431-1443.
16. Bei M, Maas R. FGFs and BMP4 induce both Msx-1-independent and Msx-1-dependent signaling pathways in early tooth development. Development 1998;125:4325-4333.
17. Dassule HR, McMahon AP. Analysis of epithelial-mesenchymal interactions in the initial morphogenesis of the mammalian tooth. Dev Biol 1998;202:215-227.
18. Tucker AS, Matthews KL, Sharpe PT. Transformation of tooth type induced by inhibition of BMP signaling. Science 1998;282:1136-1138.
19. Tucker AS, Al Khamis A, Sharpe PT. Interactions between Bmp-4 and Msx-1 act to restrict gene expression to odontogenic mesenchyme. Dev Dyn 1998; 212:533-539.
20. Neubuser A, Peters H, Balling R, Martin GR. Antagonistic interaction between FGF and BMP signaling pathways: A mechanism for positioning the sites of tooth formation. Cell 1997;90:247-255.
21. Gerlach LM, Hutson MR, Germiller JA, Nguyen-Luu D, Victor JC, Barald KF. Addition of the BMP4 antagonist, noggin, disrupts avian inner ear development. Development 2000;127:45-54.
22. Chang W, Nunes FD, De Jesus-Escobar JM, Harland R, Wu DK. Ectopic noggin blocks sensory and nonsensory organ morphogenesis in the chicken inner ear. Dev Biol 1999;216:369-381.
23. Morsli H, Choo D, Ryan A, Johnson R, Wu DK. Development of the mouse inner ear and origin of its sensory organs. J Neurosci 1998;18:3327-3335.
24. Takemura T, Sakagami M, Takebayashi K, et al. Localization of bone morphogenetic protein-4 messenger RNA in developing mouse cochlea. Hear Res 1996; 95:26-32.