

Otodental syndrome: three familial case reports

Lourdes Santos-Pinto, DDS, PhD Maria del Pilar Oviedo, DDS, MS Ary Santos-Pinto, DDS, PhD
Helda I. Iost, DDS, PhD N. Sue Seale, DDS, MSD Anil K. Reddy, DDS, MPH

Otodental syndrome, also named otodentodysplasia,¹ otodental dysplasia,²⁻⁴ and globodontia,^{5,6} has been reported as a type of ectodermal dysplasia, with an autosomal dominant inheritance of variable expressivity.¹ It was first reported by Toledo et al.⁷ as a multiple dental anomaly in three brothers in whom the authors observed abnormal crown morphology with globe-shaped molars and short roots. The clinical and radiographic aspects were not sufficient to allow differentiation between primary and permanent teeth. The histologic findings showed that the overall structure of enamel, dentin, cementum, and pulp appeared to be normal.

In 1972, Levin and Jorgenson¹ reported a new syndrome that they named "familial otodentodysplasia", characterized by posterior teeth with abnormal morphology in association with high-frequency hearing loss. Absent or small premolars were also reported. Radiographic analysis of the abnormal teeth showed large pulp chambers, some of which were duplicated, and the root length was short compared with crown height.

Witkop et al.⁶ reported "globodontia" in otodental syndrome and observed that the patients had a long facies, anteverted nostrils, a long philtrum, and a full-cheek appearance. The primary canines and molars were larger than normal and presented with globe-shaped crowns having anomalous grooves extending from the labial and lingual onto the incisal or occlusal surface of the crowns. Localized yellow-white spots were present on the enamel, particularly on the labial surfaces of the primary canines.

Chen et al.² observed the presence of extra incisors in the maxillary anterior region and conical supernumerary microdontic teeth on the palatal side of primary maxillary molars.

Conical maxillary lateral incisors were reported by Stewart and Kinirons⁵ in patients with otodental syndrome. However that case report was considered as doubtful by Gorlin et al.⁸

Griffin⁹ reported a case diagnosed as a possible variant of otodental syndrome. However, the clinical finding showed only fusion of a maxillary second permanent molar with a supplemental premolar. The posterior teeth did not show globular deformities and

the hearing impairment was not only for high-frequency sound.

This case report describes the occurrence of familial otodental syndrome in three generations of a family, including the manifestations of the condition in both the primary and permanent dentitions of three affected patients.

Case report

The transmission of this disorder through generations is depicted in Fig 1.

Case 1

Patient II-13 (Fig 1) was examined for the first time, along with her brothers, when she was 6 years old by Toledo et al.⁷ and all were reported as having multiple dental anomalies with size and shape alterations in posterior teeth. Recently, at age 30, she demonstrated a normal physical appearance but her lower anterior facial height was excessive with an abnormal lip posture.

Oral examination of the maxillary arch revealed three erupted incisors, which were normal in size and shape. It was impossible to say if the canines were primary or permanent. The two premolars on the right side and one on the left side were conical, and the first and second molar crowns were remarkable for their rounded or globular shape.

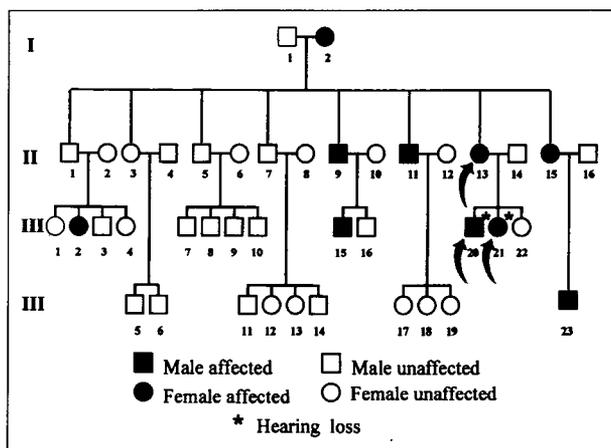


Fig 1. Distribution of the individuals in the family with otodental syndrome.

In the mandibular arch, incisors and canines had the same shape. Only one conical premolar was present on each side, the left first permanent molar had been extracted, and the remaining molars presented with many developmental grooves radiating from the center pit on to all surfaces, dividing each crown into lobules of different sizes.

The patient demonstrated a posterior bilateral crossbite and a maxillary midline shift to the right due to the missing lateral incisor. The mandibular arch was "U" shaped and the maxillary arch was "V" shaped and constricted, so that the distance between the lingual surfaces of the premolar measured 2.5 cm, resulting in a deep palate.

Dental radiographs showed an impacted maxillary right lateral incisor and a periapical radiolucency over the mandibular left first permanent molar. The roots of affected teeth were short compared to crown size and taurodontism was evident.

Case 2

Patient II-13 (case 1) had three children, two of whom were affected with the syndrome. The oldest son, III-20 (case 2) was examined clinically at 3 years of age and presented with normal physical development and a symmetrical face. Oral examination revealed delayed eruption of about 1 year, with only primary central and lateral incisors and first molars erupted (Figs 2, 3). The incisors were normal size and shape, and the molars were bulbous with occlusal fissures that were greatly reduced in depth. In the maxillary first molars, developmental grooves radiated from the occlusal pit onto the buccal, lingual, and proximal surfaces to the cervical area and divided the crown into lobules of different sizes. A yellow spot was present on the buccal surface of the maxillary left first molar. The mandibular first molars appeared to be fused with a supernumerary tooth on the distal surface.

One year later, the patient was re-examined (age 4) and gingival hyperplasia was a common clinical finding around erupting teeth (Fig 4). This tissue appeared as a pedunculated mass on the buccal of

the maxillary molar and was not biopsied.

At age 9, re-examination of patient III-20 showed that only the permanent incisors had replaced their primary antecedents (Figs 5, 6). The mandibular first permanent molars had erupted and the primary canines were bulbous with labial and lingual cusp-like protrusions separated by shallow grooves. Dental plaque accumulations were found on the primary molars. Dental radiographs showed roots of the teeth to be short compared to their crowns (Fig 7). Molars were taurodontic, and a vertical septum was noted in the pulp chamber of each mandibular second primary molar and first permanent molar; thus, the pulp chamber appeared bisected. No evidence of permanent maxillary canines, left first and second premolars, right second premolar, and mandibular right second premolar were found. There was a horizontally positioned tooth or an anomalous premolar crown on the maxillary right side. At 9 years of age, the patient began to lose his hearing.

At age 14, the patient showed a long face. When in habitual lip posture, a wide gap between the upper and lower lip was present. Lip closure increased contraction of the orbicularis oris and mentalis muscles (Fig 8). He had a malocclusion with an anterior open bite and maxillary lateral incisors lingual to the central incisors (Fig 9).

Case 3

The oldest daughter of II-13 (case 1), patient III-21 (case 3), was examined when she was 10 years old. She had lost her hearing in childhood and had a history of frequent ear abscesses. A long face, full-cheek appearance, and strong mentalis activity were present. She showed the same tooth characteristics as her brother, III-20 (case 2).

In the radiographic analysis no evidence of maxillary premolars and mandibular left second premolar was found. The mandibular left lateral incisor was horizontal.

Three generations of this family had been examined since 1970 but some could not be followed afterward

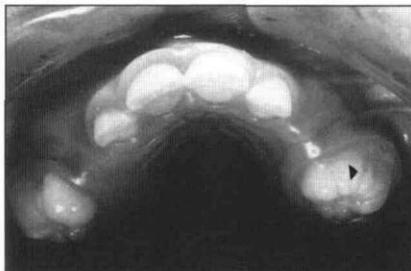


Fig 2. Patient III-20, at age 3, with delayed eruption, anomalous first primary molars, and normal incisors. Note the yellow spot on the buccal surface of the right first molar at the arrow.

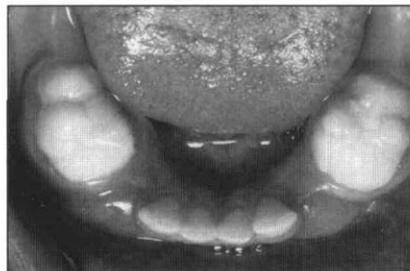


Fig 3. Lower arch of patient in Fig. 2, with delayed eruption, large and globe-shaped first primary molars, and normal incisors.

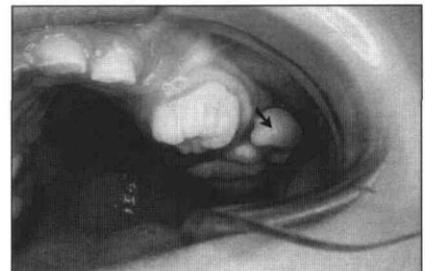
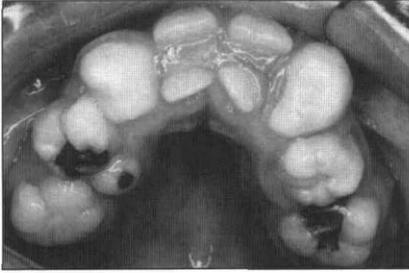
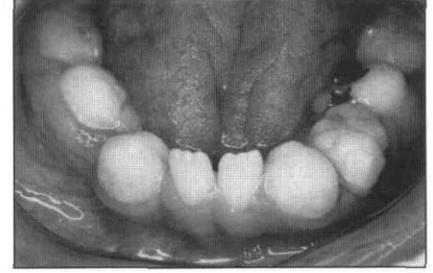


Fig 4. Gingival hyperplasia (arrow) during tooth eruption of patient III-20, at 4 years of age.



◀ Fig 5. Patient III-20, at age 9, with delayed eruption and anomalous primary canines and molars. The incisors were normal but the laterals were lingually positioned.



▶ Fig 6. Lower arch of patient in Fig. 5, with large and globe-shaped first permanent molars and primary molars and canines. The central incisors were normal and the lateral incisors were absent.

because they moved to another city. The grandmother (I-2) reported to be an affected person, but we could not confirm that information because all her teeth had been extracted.

Discussion

Previous reports considered otodontal syndrome to have an autosomal dominant inheritance.^{1-4,6} This syndrome is characterized by abnormal crown morphology of the posterior teeth and canines and sensorineural hearing loss.

We were able to examine three generations of patients transmitting this syndrome and to follow the mother (II-13) and her son (III-20) and daughter (III-21). Chromosomal analysis of these patients has not been done, but the pattern of inheritance and the features demonstrated were consistent with the syndrome as it is reported in the literature.^{1-4,6,8} All patients presented deformed maxillary arches, large and bulbous canines, globe-shaped molars, delayed eruption of teeth of about 1 year, and multiple missing primary and/or permanent teeth.

The mother (II-13) did not have hearing loss, her son (III-20) showed partial hearing loss in adolescence, and her daughter (III-21) lost her hearing in early childhood following frequent ear abscess. However, the mother could still demonstrate hearing loss, because its onset varies from early childhood to middle age as reported in Chen et al.² Bilateral symmetric hearing loss

was reported by Cook et al.¹⁰ in a patient after age 40.

Levin et al.³ suggested that this hearing loss may be due to a genetic defect in neuroectoderm; however, it could have some nongenetic cause. Cook et al.¹⁰ reported that the sensorineural hearing loss could be caused by a local lesion in the cochlea with an unknown mechanism. Witkop et al.⁶ reported a persistent purulent ear infection that had resulted in perforation of the drum.

Abnormal morphology of the crowns of selected groups of teeth is the most consistent anatomical finding. The molars are globe shaped with absent or shallow fissures and cusps and the canines are bulbous in both dentitions.

We found similar abnormalities in primary and permanent teeth, as also reported by Levin et al.³ However, Chen et al.² reported that the deformities of primary teeth were more severe than for permanent teeth.

Small or absent premolars may also be associated with otodontal syndrome.^{2-4,6} Our patients showed no consistent pattern of missing teeth. Patient III-20 did not have permanent maxillary canines, first and second premolars, or a mandibular right second premolar. In patient III-21, primordia of the maxillary first and second premolars and the mandibular left second premolar could not be identified.

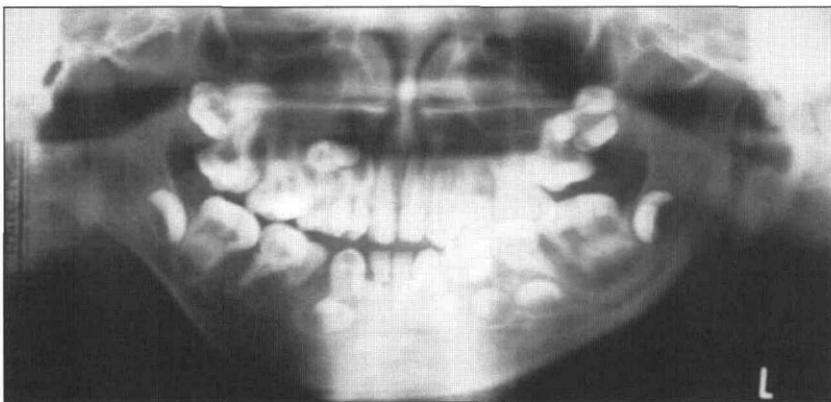


Fig 7. Panoramic film of patient in Fig. 5. Note the absence of permanent maxillary canines, left first and second premolar, right second premolar, and mandibular right first and second premolars.

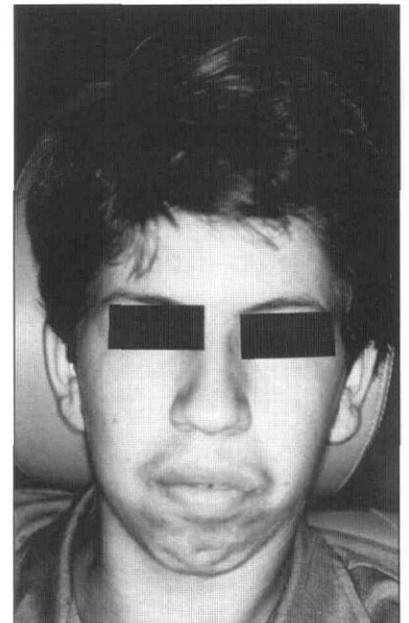
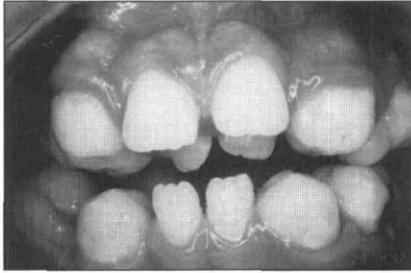
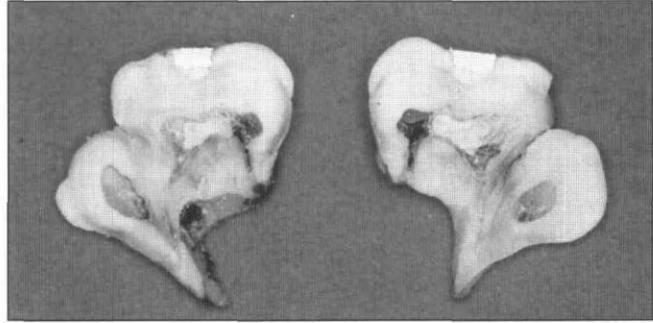


Fig 8. Clinical view of patient III-20, at age 14, with long face and strong mentalis activity.



◀ Fig 9. Occlusion of patient in Fig. 8, at age 14. Note malpositioned lateral incisors and open bite.

▶ Fig 10. Exfoliated primary mandibular first molar, sectioned to show a separated pulp chamber.



The combination of normal primary or permanent incisors with malformed posterior teeth makes it very difficult to identify a single factor responsible for the alterations observed.

Radiographic analysis of the affected teeth showed large and apparently divided pulp chambers and taurodontism. The taurodontic appearance is a consequence of the massive size of the crown and large appearance of the pulp chamber rather than enlargement of the pulp chambers extending into the roots. The same characteristic was observed by other authors.^{2,6} Exfoliated teeth were sectioned and some primary molars demonstrated two separated pulp chambers, giving the impression that either fusion or gemination with a supernumerary tooth had occurred (Fig 10).

The patients followed in this report did not demonstrate other anomalies reported to be associated with this syndrome: congenital coloboma of the eye, complex odontoma, and numerous microdontic teeth.¹¹ Our patients had yellow-white areas on the labial surface of the first primary molar. According to Witkop et al.,⁶ the enamel in this area contains voids similar to those in hypomaturation defects of enamel. The eruption of the primary and permanent teeth was delayed.

The association of sensorineural hearing loss and dental anomalies can also be found in other syndromes. Lee et al.¹² found sensorineural hearing impairment associated with permanent anterior missing teeth in two patients. Gorlin et al.⁸ described association between bilateral sensorineural hearing loss and multiple anterior *dens invaginatus* combined with fusion that resulted in unusual crown shapes.

The authors thank Dr. Tereza Gonzales for helpful suggestions in the discussion.

Drs. Santos-Pinto are professors in the department of pediatric dentistry and orthodontics in Araraquara College of Dentistry,

UNESP, Brazil and visiting professors in the pediatric dentistry and orthodontic departments at Baylor College of Dentistry, Dallas, Texas. Dr. Oviedo is a graduate student in pediatric dentistry graduate program at Araraquara College of Dentistry and Dr. Iost is professor in the department of pediatric dentistry at Araraquara College of Dentistry. Dr. Seale is professor and chairman and Dr. Reddy is assistant professor in the pediatric dentistry department at Baylor College of Dentistry, Dallas, Texas.

References

1. Levin LS, Jorgenson RJ: Familial otodontodysplasia: a "new" syndrome. *Am J Hum Genet* 24:61, 1972.
2. Chen RJ, Chen HS, Lin LM, Lin CC, Jorgenson RJ: "Otodontal" dysplasia. *Oral Surg Oral Med Oral Pathol* 66:353-38, 1988.
3. Levin LS, Jorgenson RJ, Cook RA: Otodontal dysplasia: a "new" ectodermal dysplasia. *Clin Genet* 8:136-44, 1975.
4. Jorgenson RJ, Marsh SJ, Farrington FH: Otodontal dysplasia. *Birth Defects Orig Artic Ser* 11:115-19, 1975.
5. Stewart DJ, Kinirons MJ: Globodontia. A rarely reported dental anomaly. *Br Dent J* 152:287-88, 1982.
6. Witkop CJ Jr, Gundlack KK, Streed WJ, Sauk JJ Jr: Globodontia in the otodontal syndrome. *Oral Surg Oral Med Oral Pathol* 41:472-83, 1976.
7. Toledo OA, Rocca RA, Vono RMG, Bausells HII: Anomalia dental múltipla em três irmãos. Caso clínico. *Rev Fac Farm Odont Araraquara* 5:207-214, 1971.
8. Gorlin RJ, Cohen MM Jr, Levin LS: *Syndromes of the Head and Neck*, 3th Ed. New York: Oxford University Press, pp 859-77, 1990.
9. Griffin GJ: Case report: a possible variant of otodontal syndrome. *J Paediatr Dent* 1:27-33, 1985.
10. Cook RA, Cox JR, Jorgenson RJ: Otodontal dysplasia: A five year study. *Ear Hear* 2:90-94, 1981.
11. Winter GB: The association of ocular defects with the otodontal syndrome. *J Int Assoc Dent Child* 14:83-87, 1983.
12. Lee ML, Levin LS, Kopstein E: Autosomal recessive sensorineural hearing impairment, dizziness, and hypodontia. *Arch Otolaryngol* 104:292-93, 1978.