A Case with Neurosensorial Hearing Loss and Anodonty Findings: Is This a Variant of Otodental Syndrome?

Nörosensoryal İşitme Kaybı ve Anodonti Bulguları Olan Bir Olgu: Otodental Sendromun Bir Varyantı mı?

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ABSTRACT Otodental syndrome has been described as a syndrome of sensorineural hearing loss and dental anomalies. In this syndrome, generally autosomal dominant inheritance pattern has been described. In this study, we present an 8-year-old girl with bilateral sensorineural hearing loss and partial anodonty findings in a non-consanguineous family without eye involvement. In literature, this syndrome has been reported only in few cases with high penetrance and variabile expressivity. Therefore, we assume that this case may be accepted as a variant form of otodental syndrome with solely neurosensorial hearing loss, anodonty and deep narrow palate findings without dental dysplasia and eye involvement.

Key Words: Hearing loss, sensorineural; dentin dysplasia; anodontia

ÖZET Otodental sendrom sensorinöral işitme kaybı ve dental anomalilerin eşlik ettiği bir sendrom olarak tanımlanmaktadır. Bu sendromda, genel olarak otozomal dominant kalıtım paterni tanımlanmıştır. Bu çalışmada, aralarında akrabalık bulunmayan bir ailenin iki taraflı sensorinöral işitme kaybı ve parsiyel anodonti bulgularının olduğu, göz tutulumunun olmadığı 8 yaşındaki kız çocukları ile ilgili sunum yapmaktayız. Literatürde, bu sendrom yüksek penetrans ve ekspressivite değişkenliği gösteren, az sayıda olguda rapor edilmiştir. Bu nedenle, dental displazi ve göz tutulumu olmaksızın, sensorinöral işitme kaybı, anodonti ve dar-yüksek damak bulgularının eşlik ettiği bu olgunun, otodental sendromun bir varyantı olarak kabul edilebileceğini düşünmekteyiz.

Anahtar Kelimeler: Sensorinöral işitme kaybı; dental displazi; anodonti

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The otodental syndrome, also called as otodental dysplasia, is characterized by striking dental phenotype known as globodontia, and associated with sensorineural high frequency hearing loss and eye coloboma in some cases.[1,2] Hitherto only a few families have been described with the findings of this syndrome.[1] In most families with this syndrome, dental abnormalities and sensorineural high frequency hearing loss are common whereas eye abnormalities are rare.[1] The syndrome demonstrates variable expressivity and penetrance with autosomal dominant inheritance pattern in familial cases.[1,3]

We assume that the findings of our case may be regarded as a variant form of otodental syndrome.

CASE REPORT

An 8-year-old girl with bilateral sensorineural hearing loss and partial anodonty findings in a non-consanguineous family has been evaluated and informed consent was obtained for the publication of the photographs and other patient data. Second cousin of the case sample had neurosensorial hearing loss with no dentinogenesis problem. In addition to that, another cousin had cleft lip. These two affected cases' and their parents physical examinations were normal and no other abnormalities were detected. There were no other cases with both dental and hearing loss problems in this family (Figure 1). The patient was born at term by cesarean section without any prenatal, natal or postnatal pregnancy complications. The mother was 27 years old at the time of delivery. The patient's birth weight was 2900 gr. Her physical and mental development was within normal limits for her age. On her physical examination, we found partial anodonty and deep-narrow palate with no other morphological abnormalities (Figure 2).

On panoramic mandibular radiograph, primary dentinogenesis was developed normally at the age of 7 (Figure 3). In secondary dentinogenesis, partial absence of canine, premolar and molar teeth, was observed without any other teeth dysmorphology findings. Her last panoramic mandibular radiograph at the age of 8 demonstrated that the

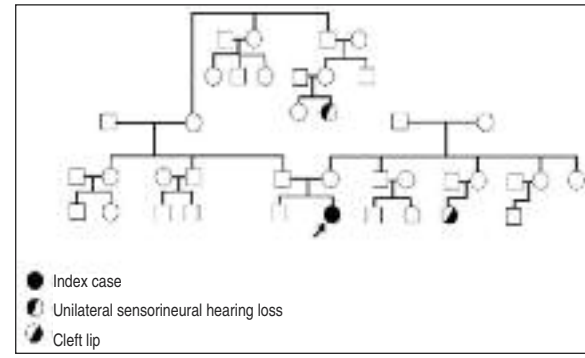

FIGURE 1: Pedigree analysis of the index case.

secondary dentinogenesis of bilateral mandibulary second incisors and first molars were normal (Figure 4). The germinal layer of the bilateral mandibulary canines, and the second molars were present and the primary right incisor was persisting. There were no germinal formation for the first incisors and premolars. The secondary dentinogenesis of bilateral maxillary first incisors and first molars were normal. Germinal formation of the left maxillary second incisor, canine, the first premolar, the second molar and the right maxillary first premolar, the second molar were present. There were no germinal formation for the left maxillary second premolar and the right maxillary second incisor, canine and second premolar.

The patient also had neurosensorial hearing loss (Figure 5). The patient was using hearing aids

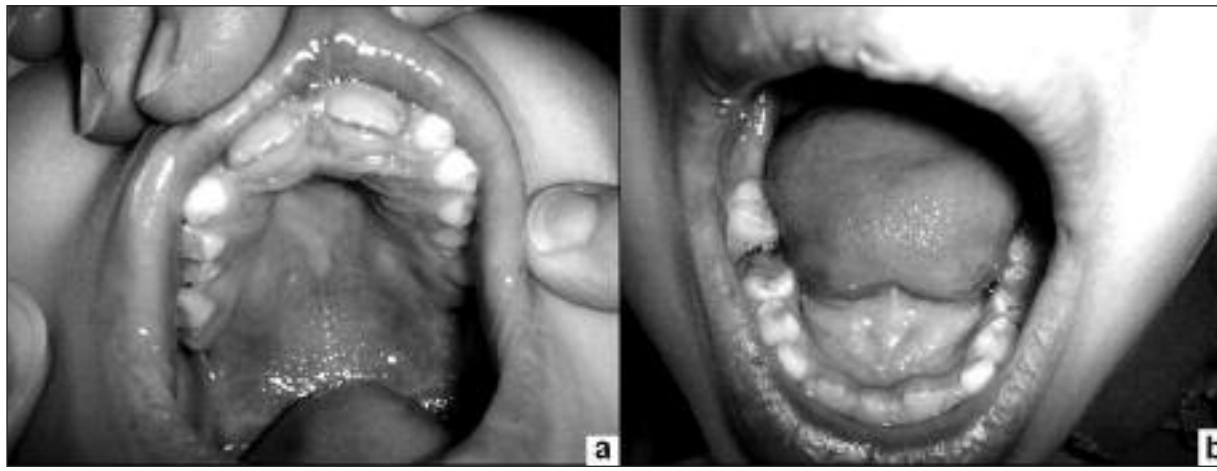

FIGURE 2: a) Deep narrow palate and the maxillary partial anodonty findings in the case.
b) Mandibulary partial anodonty findings in our case.

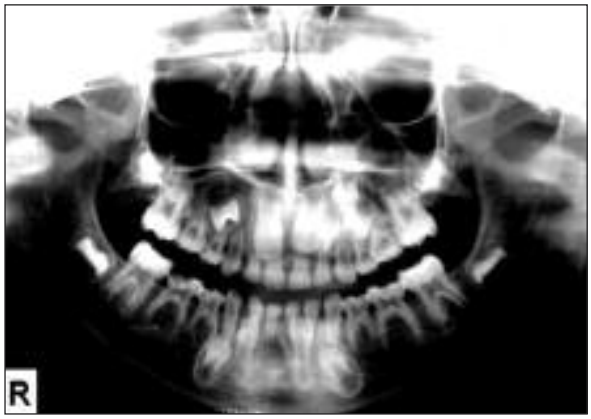

FIGURE 3: Panoramic mandibular radiograph at the age of seven. Development of primary dentinogenesis appears normal. The secondary dentinogenesis shows partial absence of the canine, premolar and molar teeth.

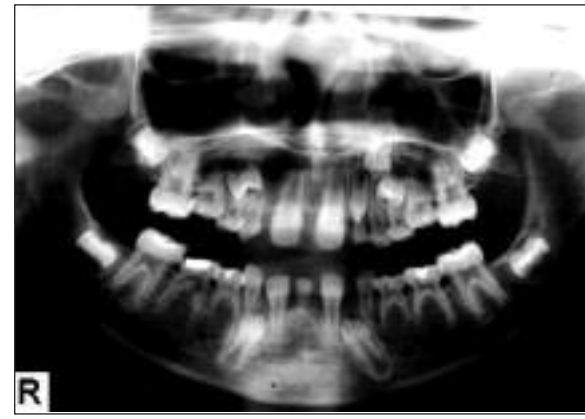

FIGURE 4: Panoramic mandibular radiograph at the age of eight shows persisting primary right incisor. We could not observe the germinal formation for the mandibulary first incisors and premolars. We also found out that there were no germinal formations for the the left maxillary second premolar, right maxillary second incisor, canine and second premolar.

for four years at the time of examination. Consecutive pure tone audiograms revealed sensorineural hearing loss in both ears, which progressed did not significantly during four-year follow-up. Pure tone averages were 47 and 40 dB hearing levels in the right and the left ears respectively (Figure 5). The speech discrimination scores were 88% in the right and 84% in the left ears. On examination, both external auditory canals and ear drums were normal. Both ears showed type A tympanograms and bilateral stapedial reflexes were evoked by intense sound stimuli, greater than 95 dB hearing levels. The patient's articulation was slightly impaired and the patient stated no other possible causes for bilateral sensorineural hearing loss. A magnetic resonance imaging of temporal bone and posterior fossa revealed no abnormalities.

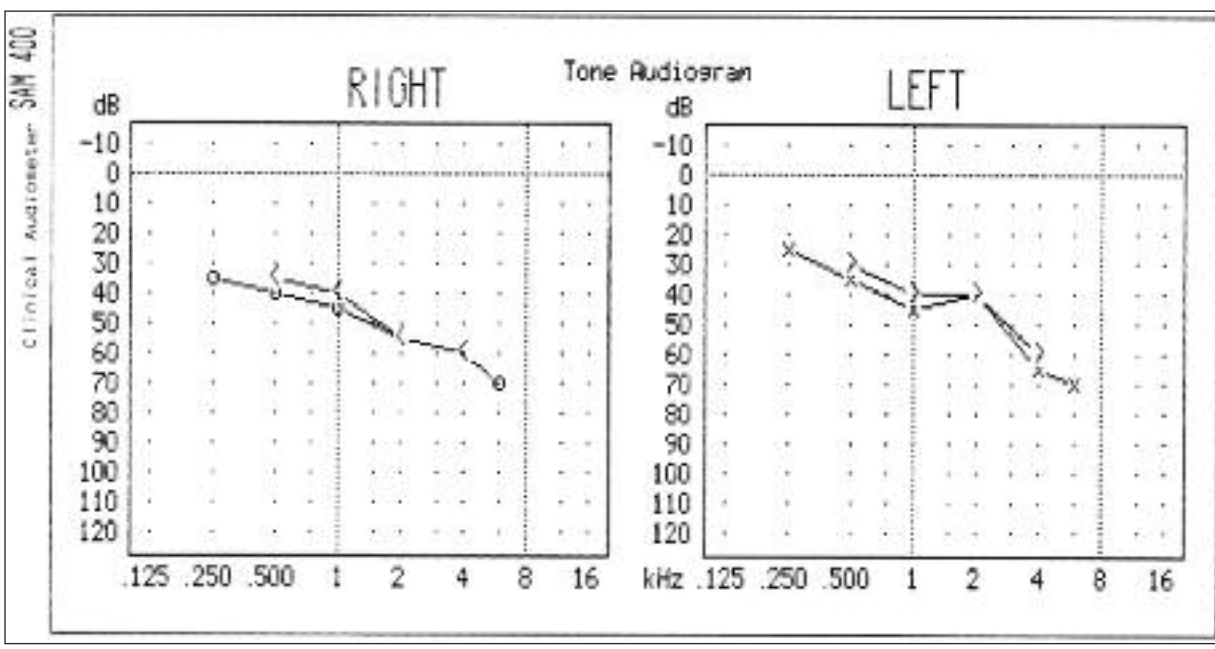

FIGURE 5: Pure tone audiogram revealed a descending-type sensorineural hearing loss. Kindly notice the drops of acuity in high frequencies.

There were no visual or structural abnormalities in ophthalmic examination.

DISCUSSION

Otodental syndrome was first described by Levin and Jorgenson with a high frequency hearing loss varied in onset from early childhood to middle age.[4] According to their findings the maxillary deciduous canines and the deciduous and the permanent molars were large and bulbous, but the incisors were normal. Premolars were congenitally missing in half of affected individuals. On dental radiographs, the deciduous molars frequently appeared to have two separate pulp chambers. Taurodontia and pulp stones were also noted.[4,5] Today, otodental syndrome or otodental dysplasia is characterized by striking dental phenotype known as globodontia, associated with sensorineural high frequency hearing loss and eye coloboma in some cases.[1,5] In our case, no dental dysplasia was observed except missing of incisors, canine premolars and there was no eye involvement. The patient had bilateral neurosensorial hearing loss. A deep and narrow palate was observed in our case, and it was the sole abnormality reported in an otodental syndrome case. According to literature, dental dysplasia seems as a major abnormality observed in otodental syndrome. In our case, the morphology of dental structure seems normal. Therefore so we assume that our case may be a variant of this syndrome with normal tooth structure, but dysplasia of the secondary dentinogenesis.

Bilateral neurosensorial hearing loss is another major abnormality observed in otodental syndrome.[1] Bilateral sensorineural hearing loss of the patient was diagnosed when she was four years old and she has been using hearing aids since then. The hearing loss was moderate, however, marked drops of acuity observed in high frequencies. This finding is constant with the previously reported cases.[3,6] One cousin of our patient also had sensorineural hearing loss without any dental pathologies and another cousin had cleft lip finding without any dental or hearing loss findings. According to our knowledge, clinical synopsis of otodental syndrome does not have in it cleft lip finding. Deep-narrow palate finding in our case is a rare finding in otodental syndrome.[1,3]

In literature, there are lot of syndromes in differential diagnosis of otodental syndrome: hearing loss and/or dental abnormalities like cleidocranial dysostosis, anhidrotic ectodermal dysplasia, osteogenesis imperfecta, and syndromic microphthalmia type 2 (oculofaciocardiodental syndrome). Cleidocranial dysostosis is a rare syndrome that affects craniofacial, dental and skelatal features with or without hearing loss.[7] Anhidrotic ectodermal dysplasia is characterized by hypohidrosis, hypotrichosis and total or partial anodonty.[8] Six types of osteogenesis imperfecta exist. According to type of disease, patients suffer from growth deficiency, skeletal abnormalities like limb deformity, fractures, scoliosis, kyphosis and hyperextensible joints, blue sclera, hearing loss and dental problems.[9] In oculofaciocardiodental syndrome, dental abnormalities accompany ocular, facial and cardiac abnormalities.[10] When we examine all of these syndromes, only the hearing loss and/or dental abnormalities show similarity to our case.

The correct definition of the otodental syndrome is difficult because of its low frequency, variable expressivity and penetrance.[1,3] So, our case may be a variant form of this syndrome with solely neurosensorial hearing loss, anodonty and deep narrow palate, without dental dysplasia and eye involvement findings. They possibly share the similar unknown genetic problem or problems appeared in the development of these cases.

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