

Auditory Functions In Oto-Palato-Digital Syndrome

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Citation

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Abstract

The oto-palato-digital (OPD) syndrome is a rare X linked disorder characterized by generalized skeletal dysplasia. A case with major features of mild conductive hearing loss, with hypertelorism, cleft soft palate, hand and foot abnormalities is presented. The evaluation of the auditory functions of case was performed by objective and subjective hearing tests with radiological findings of temporal bone. Hearing aids were prescribed for both ears for the mild conductive hearing loss. The patient remained in good health for three years.

INTRODUCTION

The oto-palato-digital (OPD) syndrome is a rare X linked disorder characterized by generalized skeletal dysplasia. Initially described by Taybi in 1962, since then a little over 30 cases have been reported in the literature (1,2). The differences in clinical presentation between candidates have led authors to classify OPD syndrome into two subtypes: type-I and type-II. The principle features of type I are hearing loss, cleft palate, distinctive facial appearance and skeletal dysplasia of the hands and feet, mild mental retardation. Type II is lethal skeletal dysplasia with bowed limbs, overlapping fingers, wavy and short ribs and respiratory difficulties (3). The primary pathogenesis of this disorder is not clarified.

The type of hearing loss with the OPD syndrome may be conductive, sensorineural, or mixed; and is almost always bilateral (2). Conductive hearing loss usually results from an ossicular malformation, which was seen on radiographs of temporal bone and then it may be amenable to surgical intervention. We report a case of OPD syndrome type I, and evaluate the auditory functions by objective and subjective hearing tests with radiological findings of temporal bone.

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CASE REPORT

The male child was a 8-years-old mildly retarded. His height was 132 cm, his weight was 26 kg (below the 10th percentile for age). The occipitofrontal head circumference was 55 cm.

(98th percentile for age). He was born following an uneventful full term pregnancy weighing 3100 g, with a occipitofrontal head circumference of 35 cm and with a height of 53 cm and no antenatal ultrasound had been performed. He was the first child of unrelated parents, 19 years-old mother and a 21 years old father. He walked at 18 months, and spoke at 20 months.

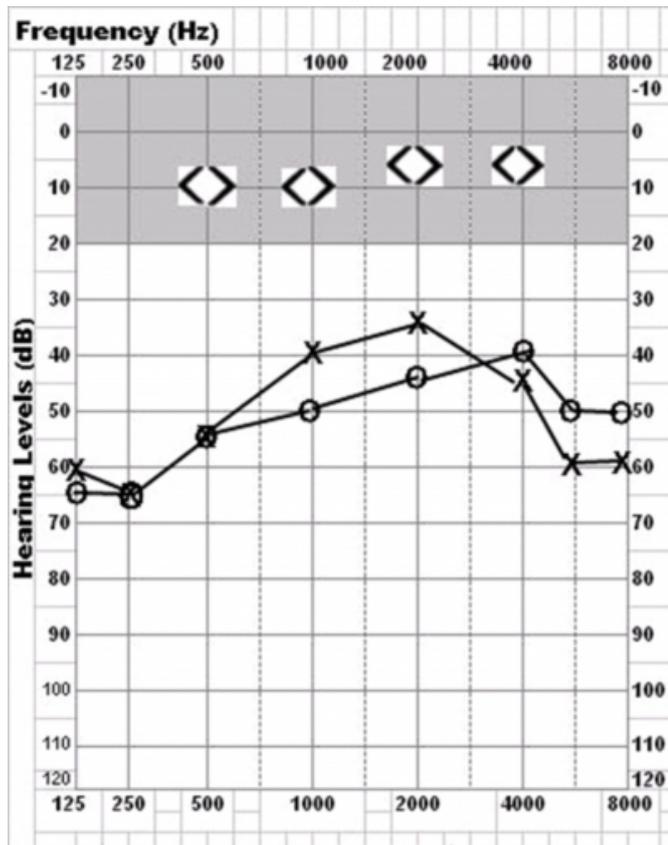
Physical examination showed frontal and occipital prominence with thick frontal bone, prominent supraorbital ridges, hypertelorism with lateral fullness of supraorbital ridges, cleft soft palate and hypodontia. He had slightly low set external ear canals but normally shaped and normal ear drums with mild hearing loss as well. Examination also revealed pectus excavatum, small trunk, short and broad distal phalanges of thumbs.

In audiometric evaluation; immittance audiometry and acoustic reflex measures were performed firstly. Then, pure tone audiometry, otoacoustic emissions (OAE) and auditory brain system responses (ABR) testing were measured serially.

Tympanometric findings showed low compliance, absent stapedius reflex and a characteristic curve of poor motility of the ossicular chain (type As tympanogram). Pure-tone audiometry revealed bilateral mild conductive hearing loss in right and left ear. The air-bone gap value of the right ear was 38 dB and the left ear was 42 dB (Figure1). ABR was performed and an increased interpeak latencies with having 60 dB threshold levels were found.

Figure 1

Figure 1: Mild conductive hearing loss was shown in pure tone audiometry.



Anterior-Posterior (A-P) hand radiography, A-P foot radiography and temporal bone computed tomography (CT) were performed. Poor mastoid pneumatization, soft tissue density in both middle ear, hyperdense bony appearance around ossicles of the middle ear, cochlea and semicircular channels were revealed in temporal bone CT (Figure 2). Prominence at occipital and frontal bone, thick frontal bone, and thick base of skull, lateral fullness of supraorbital ridges, relatively small mandible were found in three-dimensional skull CT (Figure 3). Abnormal tubulation of metacarpals especially 1st on minimal oblique orientation of 2nd metacarpal and fusion w/ coma shaped like trapezoid, cone shaped epiphysis of the thumb, abnormal tubulation of metatarsal bones, oblique orientation of 2nd and 3rd metatarsals, cone shaped epiphysis of distal phalanx of the thumbs were also seen in AP hand and foot radiography (Figure 4).

Figure 2

Figure 2: CT scans through the temporal bone show poor mastoid pneumatization, soft tissue density in both middle ear, hiperdense bony appearance around ossicles, cochlea and semicircular channels.

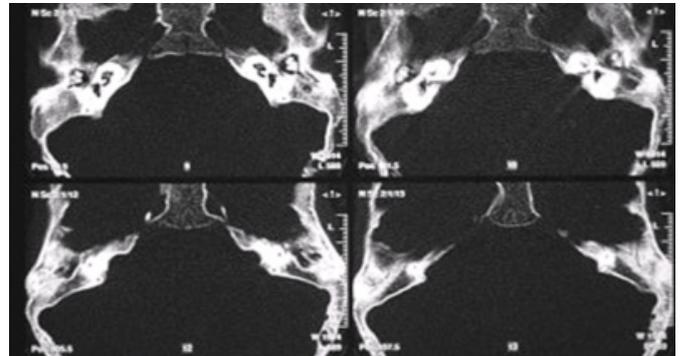


Figure 3

Figure 3: Three-dimensional skull CT findings: Prominence at occipital and frontal bone, thick frontal bone, and thick base of skull.

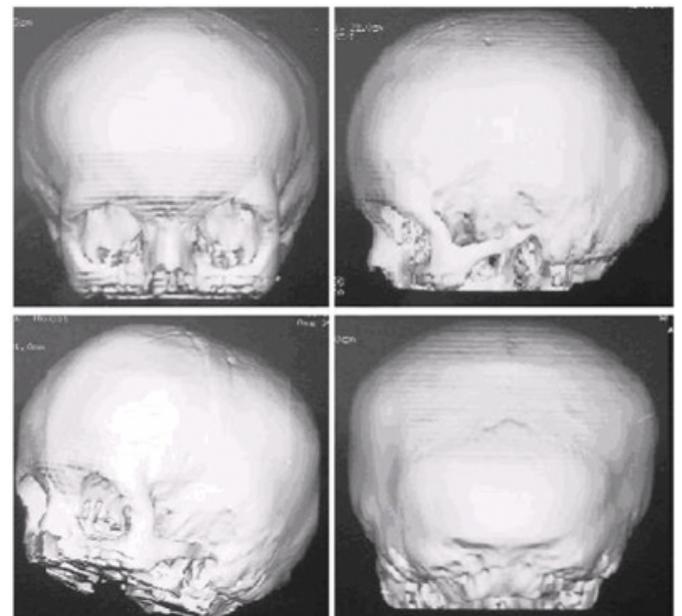


Figure 4

Figure 4: Radiographic findings. Note dysharmonic ossification with hypoplasia of all metacarpals, hypoplastic middle phalanges and broad proximal and distal phalanges. a: AP radiograph of the hand. b: AP radiograph of foot



Assessments of the other organs and bones were made and fortunately bowed limbs, overlapping fingers, wavy and short ribs and respiratory difficulties were not found, and the patient was diagnosed as OPD-I. The surgery procedure of primary veloplasty was performed for the soft palate of the case. The parents rejected exploratory tympanotomy. Hearing aids were prescribed for both ears for the mild conductive hearing loss. The patient remained in good health until now, after three years.

DISCUSSION

The OPD syndrome is a rare genetically inherited X-linked disorder. Duddin et al. noticed the distinctive extremities abnormalities and face appearance, then this disorder was named as OPD syndrome (4). A more lethal variant of the syndrome was described later by Fitch and was termed OPD type II (5). OPD syndrome, frontometaphyseal dysplasia and Menlick Needles syndrome may be a single entity and proposed the name of fronto-otopalatodigital osteodysplasia (6). The principle features of type I are hearing loss, cleft palate, distinctive facial appearance and skeletal dysplasia of the hands and feet, mild mental retardation. The case described in this report shared all the typical clinical, audiologic and radiographic findings of OPD type I.

The histopathological data in the cases indicate that both trabecular and periosteal bone are abnormal. Trabecular bone abnormalities occur as a primary rather than secondary phenomenon in OPD syndrome. Although enchondral ossification is normal, periosteal ossification is defective with islands of cortical bone aplasia and hyperplasia of the periosteum. The basic defect underlying this disorder may involve the extra cellular matrix and its turnover. It is the orderly mineralization of the matrix that is responsible for the strength and resistance to compression of bone (6).

Hearing loss is one of the most characteristic findings in

OPD type I. The type of hearing loss is always bilateral and may be conductive, sensorineural, or mixed; varies in severity. Conductive hearing loss is common in OPD syndrome. Taybi described the conductive hearing loss with the thickened ossicles as showed by X-rays. Abnormal ossicles were found in the middle ear (7). Some authors were tried to improve the hearing by performing stapedectomy. Although good motility of the ossicles was established during the operation, audiological findings after the operation showed no hearing improvement (8). We detected an intermediate conductive hearing loss with type As tympanogram. The ipsi lateral and contra lateral acoustic reflexes were not seen. The hearing threshold levels were also found 60 dB in ABR. Hyperdense bony appearance around ossicles of the middle ear, cochlea and semicircular channels were found in the temporal bone computerized tomography scanning of the case. As the parents rejected exploratory tympanotomy, hearing aids were prescribed for both ears of the present case. Although sensorineural hearing loss was found in some cases (9), there was no reports for explaining the features of sensorineural hearing loss in this disorder. Shi was described findings of Mondini dysplasia for the possible cause of sensorineural hearing loss in cadaver temporal bone (10).

The primary pathogenesis is not clarified, however in the distal Xq chromosomes the mutant gene was confirmed (3). OPD syndrome type I and type II might be due to 2 allelic genes (11). The altered gene has been mapped to Xq28 which involved in bone formation, but it was excluded as a candidate by direct sequencing of cDNA in one case. Also there is a report excluding the Xq26 region for OPD type I syndrome (12). Although the incriminated gene has not been accurately localized, the variability in the clinical expressions of this mutant gene is almost a certainty.

CONCLUSIONS

In conclusion, the OPD syndrome is a rare disorder characterized by generalized skeletal dysplasia and classified into two types. One of the principle features of type I is the abnormality of the middle ear ossicles resulting in hearing loss. Conductive hearing loss is more specific in OPD type I. Temporal bone histopathologic studies and case series are needed to shed light on the cause of the hearing loss as well as gene studies. The physicians who care for the children with OPD type I must be aware of the hearing loss for early rehabilitation.

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