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Otological findings in idiopathic hyperphosphatasia

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Abstract
A 17-year-old male patient was admitted because of progressive hearing loss since the age of six. His former blood and radiology investigation had revealed idiopathic hyperphosphatasia. On ENT examination bilateral thickened tympanic membranes with severe mixed-type hearing loss was diagnosed. Computerized tomography (CT) demonstrated expansion of the calvarial bones, including the temporal bones, except for the otic capsule. Middle-ear exploration revealed thickened middle-ear mucosa and a stone hard, immobile bony mass instead of the normal ossicular chain at the posterior superior part of the mesotympanum. No ossicular reconstruction could be attempted and the patient was rehabilitated with a hearing aid.

Key words: Idiopathic hyperphosphatasia; Hearing loss; Middle-ear surgery

Introduction

Idiopathic hyperphosphatasia (IH), also known as hereditary hyperphosphatasia, congenital hyperphosphatasia, chronic idiopathic hyperphosphatasia, and juvenile Paget's disease (Cassinelli *et al.*, 1992) is a rare congenital disorder. Although first described by Backwin and Eiger more than four decades ago in 1956, there have been only occasional case reports in the literature. In 1992, Spindler *et al.* made a review of the literature and found only 22 cases up to 1986. Since then 14 more cases have been reported in the literature.

The clinical picture in IH is well described in the literature. It affects the whole skeleton producing progressive skeletal deformity. The level of the alkaline phosphatase is elevated and this is the reason for the name of the disease. The skull is enlarged with widening of the diploë spaces. There is bowing of the long bones, severe osteopenia and expansion of the medullary cavity.

Our literature review showed that hearing loss in IH was only occasionally mentioned in these case reports. In one of these reports, however, was a detailed description of the hearing loss and its aetiology given. Therefore, in this report the main emphasis is on the otological findings of this rare disorder rather than the characteristic clinical findings which are well documented in the literature.

Case report

A 17-year-old male patient presented to the Department of Paediatrics with difficulty in walking and hearing loss. History revealed consanguinity between his parents. His family mentioned progressive deformities of the extremities and skull before the age of one. His other seven brothers and sisters were healthy and without any similar disease. On physical examination he had macrocephaly, upper and lower limb deformities (Figure 1) and difficulty in walking and sitting. His psychomotor maturation was retarded.

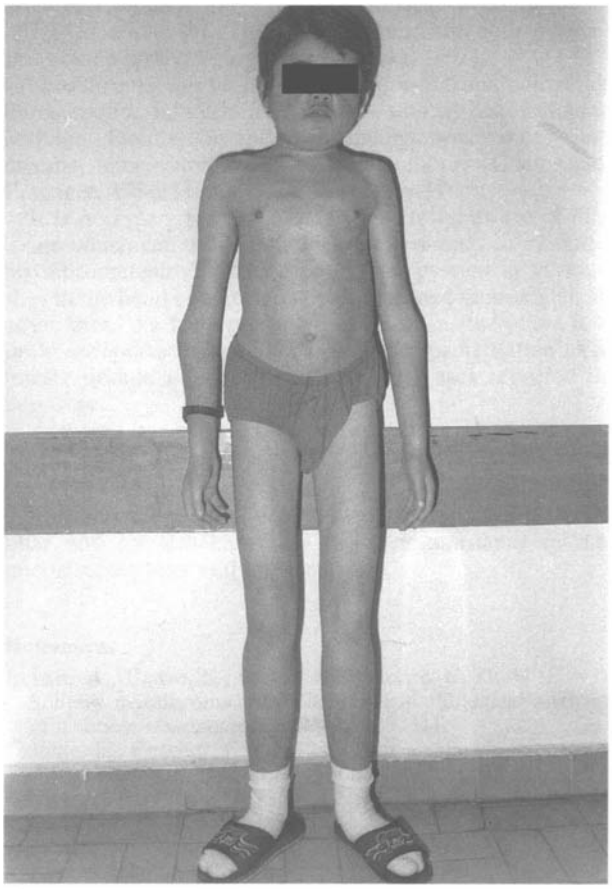

FIG. 1
The patient at the age of 17 years showing macrocephalus and skeletal deformities.

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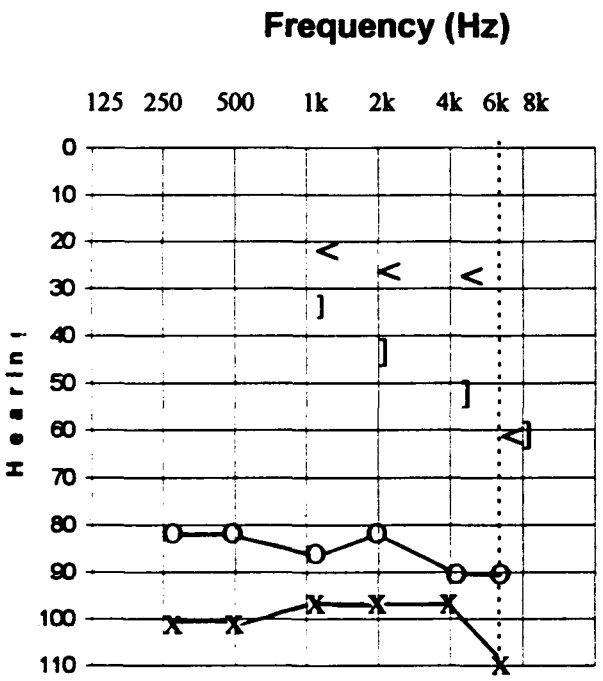

FIG. 2

Audiologic examination demonstrating bilateral profound mixed type hearing loss.

His blood electrolytes were:

		normal values
Calcium:	8.7 mg	9–11 mg
Phosphorus:	3.2 mg	4–6 mg
Parathyroid hormone:	164 pg/ml	10–65 pg/ml
Alkaline phosphatase:	1483 U/l	60–170 U/l

X-rays of his head and extremities demonstrated generalized skeletal involvement. The abnormalities were symmetrical. In the X-rays of long tubular bones the medullary cavities were widened, the cortices were thickened and dense with areas of lucencies. Metaphyseal defects were present. The calvaria was thickened with a cotton-wool appearance.

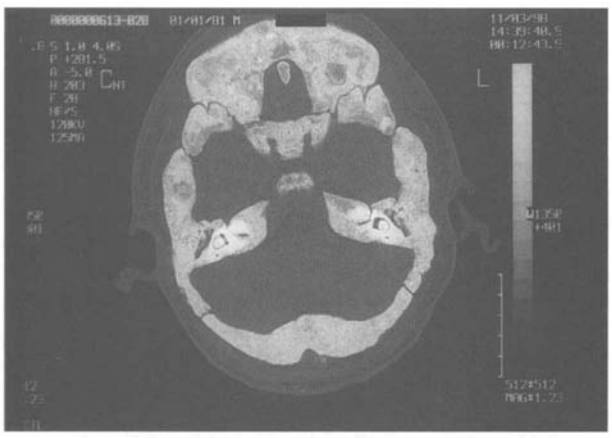

FIG. 3

Computerized tomography of the temporal bone revealed thick and increased diploë on every skull bone with massive sclerosis with normal otic capsule, and narrow internal acoustic canals.

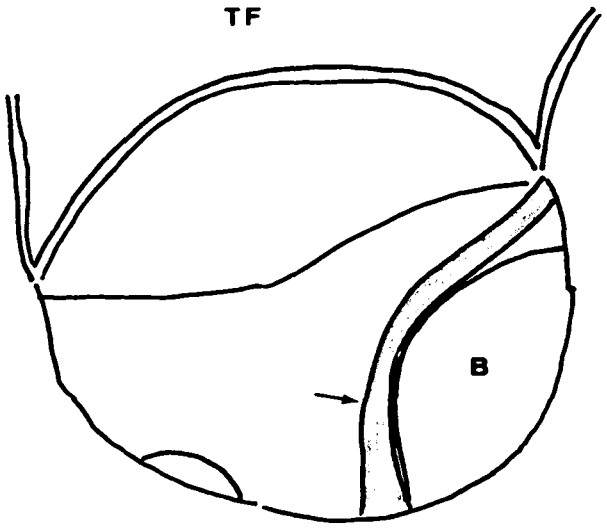

FIG. 4

Schematic representation of middle ear findings: B: stone hard round block of bony tissue, 1 cm in diameter, occupying posterior superior part of the middle ear at the usual location of the ossicles. TF: elevated tympanomeatal flap. arrow: thickened chorda tympani.

With these results the patient was diagnosed as idiopathic hyperphosphatasia and calcium lactate and sodium alendronate (Fosamax®) was started. Because of the progressive hearing loss a complete otolaryngological examination was performed. The history revealed that until the age of six he had had no hearing problem. After that age there was a progressive hearing loss that prevented his communication with other people. He had no discharge, tinnitus or vertigo.

An ENT examination revealed normal ear canals with thickened tympanic membranes on both sides. Audiological examination demonstrated a profound mixed-type hearing loss with -400 mmH_2O pressures on both sides (Figure 2). On paracentesis no fluid was present in either middle ear.

Conventional radiography and CT scan of the temporal bone revealed thick and increased diploë on every skull bone with massive sclerosis (Figure 3). The otic capsule, however demonstrated no pathology. The cochlea and semicircular canals were completely normal. The internal acoustic canals were very narrow on both sides. Middle-ear ossicles could not be identified clearly on CT.

Middle-ear exploration was planned because of the conductive component of the hearing. The left side was chosen because the better hearing ear on the right side was kept as a reserve for rehabilitation with a hearing aid in case further hearing loss occurred during exploration. An endaural incision was used. Bone material was curetted from the external ear canal for histopathological examination. Upon entering the middle ear a round block of bony tissue, 1 cm in diameter, was found occupying the posterior superior part of the middle ear at the usual location of the ossicles (Figure 4). No ossicles could be identified. As this stone-hard block of bone was completely immobile no ossicular reconstruction could be attempted. The middle ear mucosa was very thick and oedematous. This made identification of certain landmarks almost impossible. When the ear was healed it was rehabilitated with a hearing aid. Histopathological examination of the bony material curetted from the external ear canal revealed slight fibrosis of the marrow spaces.

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Discussion

Idiopathic hyperphosphatasia is a very rare disease. Since the original description by Backwin and Eiger in 1956, only 36 cases have been reported. Although the characteristic systemic findings have been well described, our literature survey showed no detailed description of the otological findings in this disease.

Until now different names have been given to this condition. Although the most common of these names is idiopathic hyperphosphatasia this disease most probably results from a congenital inborn error of metabolism. According to Blanco *et al.* (1977) the familial incidence suggests an autosomal recessive genetic defect. But in our case no relative of the patient demonstrated the same kind of disorder. Other authors also had similar cases (Dohler *et al.*, 1986; Spindler *et al.*, 1992).

Our patient had the characteristic findings described by other authors: diffuse and symmetrical skeletal deformity, bowing of the long bones and cranial enlargement. These patients also have limitation of joint movements and short stature. The main feature of the disease appears to be hyperactivity of the osteoblasts and osteoclasts with the failure to replace the immature bone by compact Haversian bone (Blanco *et al.*, 1977). This leads to the subperiosteal deposition of disorganized new bone, bowing and fragility. The intrinsic cause is unknown.

Hearing loss in these patients was noted by some authors previously in their case reports. Spindler *et al.* (1992) reported that their patient had bilateral mixed-type hearing loss. Chosich *et al.* (1991), however, reported that the hearing loss in their patient had been due to the absence of the ossicles since infancy. Apart from this information no other description of the pathology was given.

The bone involvement affects every bone in the skull. In our case the family indicated that until the age of six the patient had normal hearing. The decrease in hearing most probably started at this age. The patient had mixed hearing loss. The conductive component of this hearing loss is due to the stone-hard bony mass in the middle ear. The sensorineural component, however, is most probably due to the progressive narrowing of the internal auditory canal. It is very noteworthy that every bone in the body is affected except the labyrinthine otic capsule. As shown in Figure 3 the otic capsule remained normal in spite of the extensive generalized bone pathology.

We think that the middle-ear mucosal oedema may be due to the Eustachian tube pathology. The bony deformity most probably affected the bony part of the Eustachian tube and prevented normal aeration of the middle ear. It was interesting, however, that there was neither fluid in the middle ear nor retraction of the tympanic membrane, which was abnormally thickened on both sides.

This disease is most commonly confused with Paget's disease (Spindler *et al.*, 1992). Although the clinical picture may look similar, Paget's disease rarely starts before the age of 30, whereas idiopathic hyperphosphatasia usually starts during the first year of life. In hyperphosphatasia bone lesions are generalized and symmetrical in membranous bones, whereas, in Paget's disease it is scattered, focal regional and asymmetric.

Certain medications (aminohydroxypropylidene biphosphonate(Pamidronate) (Cassinelli *et al.*, 1992; Spindler *et al.*, 1992), calcitonin and disodium etidronate (Dohler *et al.*, 1986), and sodium alendronate in our case) are given to these patients with the aim of decreasing the bone turnover. By the age of 17 it is very difficult to expect the normalization of the radiological findings. Only a decrease in the serum alkaline phosphatase level indicates that bone turnover is declining. However, if the diagnosis is made early in childhood and these drugs started much earlier, it may be speculated that ossicular and internal auditory pathology will not be as extensive as in this patient.

Conclusion

As the otological findings in this rare disorder have not been published thoroughly in the literature it is difficult to generalize these otological findings. It would have been very interesting to compare these findings with those of other authors. However, it can be suggested that, in spite of the large air bone gap, surgical exploration to restore ossicular chain may not be effective due to abnormal middle-ear structures.

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