Otoscopic Manifestations of Osteogenesis Imperfecta Type I

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A 12-year-old boy with hearing loss, developmental delay, and osteogenesis imperfecta (OI) type I presented with a right traumatic tympanic membrane (TM) perforation, caused by impaction of hearing aid impression material on a recent fitting. The referring otolaryngologist diagnosed the perforation upon removal of the material under anesthesia and referred him for surgical management.

He denied tinnitus, otorrhea, or vertigo, but the patient’s parents and teachers noted worsened hearing and imbalance following the foreign body removal. An audiogram demonstrated bilateral severe to mild hearing loss, which was mixed in at least one ear.

Micro-otoscopy revealed an upsloping left external auditory canal (EAC), with ridges on the floor of the bony canal. The left TM was intact (Figure 1). The right EAC was clear, but also upsloping. The right TM showed a vertically oriented, posterior perforation (Figure 2). Tympanometry demonstrated a large volume on the right and a type A pattern on the left. A temporal bone computed tomography revealed the malleus and incus appeared partly demineralized bilaterally (Figure 3A). The ossicular chain appeared intact on the left, with a poorly defined stapes superstructure on the right (Figure 3B). The patient underwent a right postauricular tympanoplasty with tragal cartilage grafting.

Intraoperative findings included a thickened and fixed stapes footplate, a fractured stapes superstructure (which was removed in anticipation of a second-stage stapedectomy), and an intact and mobile incudomalleal complex. Two weeks later, all ear canal packing was removed, and the right TM was healing well.

Osteogenesis imperfecta type I is transmitted in an autosomal dominant fashion and is due to COL1A1 and COL1A2 gene mutations in type I collagen. Osteogenesis imperfecta type I patients demonstrate abnormalities of the auditory structures, such as the bony walls of the middle ear and ossicles, leading to hearing loss, which has been reported to be as high as 78%. Pathology reports of the temporal bones of patients with OI have shown that there are remnants of cartilage during ossification. Furthermore, there is a paucity of, and delay in, the deposition of bone in the endochondral and periosteal layers of the otic capsule, which has been implicated as the cause of a

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fragile stapedial crura. The fracture of the stapes superstructure in our case is likely the consequence of displaced impression material. The ridged bone pattern we observed in the EAC may reflect the altered endochondral ossification process of OI.

Hearing loss may be conductive or sensorineural in nature. Conductive hearing loss may be due to a thickened stapes footplate, or otosclerotic-like foci fixing the footplate, raising the question of the relationship between OI and otosclerosis. Histopathology of OI patients’ temporal bones has demonstrated islands of membranous bone contributing to weak support at the skull base, explaining the angulation and upsloping ear canals seen in our case.

Hypotheses to explain sensorineural hearing loss include atrophy of hair cells and the stria vascularis, anomalous bone formation of the otic capsule, or susceptibility to skull base injury. Studies suggest that hearing loss may occur at a very early age, and therefore, regular hearing screens in children with OI are necessary in order to provide early intervention.

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Figure 3. A. The CT image showing a mottled and demineralized pattern to the incus and malleus on the left. B. The CT image showing an ill-defined right stapes superstructure. CT indicates computed tomography.