Case 48: Osteogenesis Imperfecta of the Temporal Bone

HISTORY

A 36-year-old African-American woman who had a history of fluctuating bilateral mixed hearing loss that was worse on the left side than on the right complained of worsening of hearing on the right side during follow-up. At physical examination, she was a short obese woman who was wheelchair bound. The scleras were discolored. The tympanic membranes were intact. Audiogram findings demonstrated profound hearing loss on the left side and moderately severe mixed conductive and sensorineural hearing loss on the right side. In the past, she had fluctuations in her hearing with overall progressive deterioration; steroid treatment was unsuccessful. Unenhanced computed tomographic (CT) scans of the petrous temporal bones were obtained with a bone algorithm. Transverse scans of the right side (Fig 1a, 1b) and coronal scans (Fig 1c, 1d) of both sides were obtained.

IMAGING FINDINGS

CT scans were obtained as 1-mm sections in the transverse and coronal planes without administration of intravenous contrast material. The bone of the otic capsule is thickened but has a much lower than normal attenuation (Fig 1). This undermineralized bone pattern has a bilateral symmetric distribution and extensive involvement around the cochleae, vestibules, and semicircular canals and into the distal internal auditory canals, with obliteration of the oval windows.

DISCUSSION

The differential diagnosis of symmetric bilateral otic capsule demineralization, as demonstrated in this case, would include osteogenesis imperfecta (OI), diffuse otosclerosis (otospongiosis), Paget disease, syphilitic involvement of the temporal bone, and bilateral lytic metastases.

OI is one of the inherited connective tissue disorders that results in brittle bones. The molecular defects reside in various mutations that lead to decreased or defective type 1 collagen synthesis (1–3). In greater than 90% of cases, OI results from mutation in one of two genes: COL1A1 gene on chromosome 17 and COL1A2 gene on chromosome 7, which encode the chains of type 1 collagen (1,4,5). The mode of inheritance is variable, and clinical manifestations are heterogeneous. In addition to bone, OI may involve tendons, ligaments, skin, scleras, dentin, fascia, and blood vessels.

The most commonly used classification is that developed by Sillence and colleagues (6). Type 1 OI, which was previously called OI tarda, is the most common form and is a mild-to-moderate disorder inherited as an autosomal dominant trait with varying expression, and it is associated with blue scleras and hearing loss in 50% of patients (3). Type 2 OI, which is lethal in the perinatal period and was previously known as OI congenita, is the most severe form and is inherited as an autosomal recessive trait. Types 3 and 4 OI have clinical manifestations of intermediate severity between that of types 1 and 2 and are often associated with bone deformity, varying degrees of short stature, and hearing loss. Hearing loss commonly occurs in patients with type 3 and occurs in some patients with type 4 OI (3,6).

Loss of hearing is the least constant feature of OI, with an occurrence that varies between 26% and 60% (7). The association of OI, hearing loss, and blue scleras is often referred to as van der Hoeve-de Kleyn syndrome, which was well described in 1918 by these two authors (8). This syndrome usually occurs in the late 2nd or early 3rd decade of life; younger patients have a conductive hearing loss, whereas older patients have a mixed or sensorineural hearing impairment (9).

Findings at histopathologic examination of specimens removed from patients with OI provide some clues to the type of
hearing impairment that can be expected in these patients. Temporal bone specimens removed from patients with OI congenita (ie, type 2) demonstrated markedly delayed and deficient ossification in the three layers of the otic capsule (10). Microfractures, deformities, and dehiscence may involve the otic capsule and middle ear ossicles (10). Conductive hearing loss is expected in these patients and is mainly due to fractures that commonly involve the crus of the stapes or the handle of the malleus. These fractures lead to discontinuity of the ossicular chain or to fixation, by ankylosis, of the head of the malleus to the medial attic wall (10,11). Adult-onset hearing loss with OI congenita, therefore, can be a conductive type and is not always progressive (11). Reports of OI tarda (type 1) are somewhat scant since the availability of specimens depends on surgical procedures or autopsies.

Nevertheless, a few reports of type 1 and 4 OI show extensive bony labyrinth involvement by the otosclerotic or spongiotic foci that can affect all parts of the otic capsule, including the vestibules and semicircular canals (11,12). Marked porosity can also be seen in the ossicles and, at least in one case, bordering the internal auditory meatus at the fundus (12). Not infrequently, otosclerosis can be found concurrently with OI, and hearing loss can be due to otosclerosis, OI, or both (7,11). The type of hearing impairment depends on the extent and site of involvement of the otosclerotic bone dysplasia. Conductive hearing loss often results from stapedial footplate fixation secondary to otosclerosis, and less frequently, it results from stapedial crural fracture (11,13). Additional spongiotic foci with involvement of the cochlear capsule and the cochlear promontory and destruction of cochlear and vestibular endochondral bone will, in turn, contribute to progressive sensorineural hearing loss (11).

Radiologically, OI of the temporal bone can be evaluated with thin-section CT. There is a remarkable proliferation of unmineralized bone involving all or part of the otic capsule (14). This demineralization is similar to that in the cochlear form of otosclerosis (or more appropriately termed, otospongiosis), and in some cases, the two entities cannot be differentiated when there is diffuse otosclerosis (14,15). Bilateral involvement is also common in otosclerosis and occurs in up to 90% of cases, and this bilateral involvement does not help in the differentiation of the two entities.

Overall, the most common type of otosclerosis occurs in the anterior oval window (ante fenestrum), and this type is called fenestral otosclerosis. The other type, called retrofenestral or cochlear otosclerosis, occurs around the cochlea, although it rarely occurs without fenestral involvement. Since OI is a type of systemic bone dysplasia, its involvement of temporal bone tends to be much more extensive and symmetric than that in otosclerosis. Given the severe degree and extent of the tempo-

Figure 1. Unenhanced CT scans of the petrous temporal bone in the right ear of the patient. (a, b) Transverse scans of the right side and (c, d) coronal scans of both sides show that the otic capsules are markedly thickened and undermineralized in a bilateral and symmetric fashion, with dysplastic bones (arrowheads in d) extending to the internal auditory canals (arrows in d).
eral bone findings and clinical features, OI is the most likely diagnosis.

A conventional radiograph of the tibia and fibula (Fig 2) demonstrates multiple healed fractures of the tibia transfixed by an intramedullary rod, as well as an un-united fracture deformity of the middle fibular shaft. The bones are gracile and osteoporotic, features that are characteristic of OI. Review of the patient’s medical records confirmed that she had a clinical diagnosis of type 1 OI, as did her son.

Since OI and otosclerosis seem to share many similar histologic and radiographic features, their relationship is regarded as controversial in the literature. Biochemical data, however, have demonstrated dissimilar protein and certain enzyme concentrations in these two entities (16). Furthermore, histologic analysis shows that although otosclerosis may occur as part of the OI syndrome, it can be distinguished morphologically when the two coexist, because OI involves all three layers of the otic capsule (endosteum, endochondral layer, and perosteum), whereas otosclerosis is limited to the endochondral layer (7,10).

As part of the work-up for this patient’s mixed hearing loss, she underwent a magnetic resonance (MR) imaging examination (Fig 3) 6 years prior to undergoing CT. MR images of the internal auditory canals are shown in Figure 3c and 3d as T1-weighted images obtained before and after administration of gadolinium-based contrast material (gadopentetate dimeglumine, Magnevist; Berlex Laboratories, Wayne, NY). These images demonstrate enhancement of the fundus of the distal internal auditory canals and of the bones around the cochleae, the vestibules, and the semicircular canals bilaterally. No mass lesion is seen. This enhancement pattern of OI of the temporal bone is an interesting finding that, to our knowledge, has not been described in the literature.

Consistent enhancement, but to a varying degree, of lesions in otosclerosis has, however, been observed at MR imaging (17). Such enhancement is thought to be caused by pooling or leaking of gadolinium-based contrast material into the vascular foci of spongiotic bone or, alternatively, caused by the associated inflammation. Given the radiographic and some histopathologic differences between OI and otosclerosis, it is not surprising to see also, in our case of OI, enhancement of the otic capsule and distal internal auditory canal, where there is extensive spongiotic involvement.

Paget disease (osteitis deformans) is a progressive bone disease of unknown cause that primarily involves the axial part of the skeleton with a predilection for the pelvis, the femur, the vertebra, and the skull (usually occurring in that order). As the skull is commonly affected, it is always associated with squamous temporal bone involvement, and the changes are initially lytic and typically begin in the petrous pyramid and progress laterally. The involvement can be diffuse, demonstrating generalized and homogeneous demineralization with a washed-out appearance on a CT scan (15).

The otic capsule is the last region to be affected, as opposed to the finding in this case, in which it was predominantly affected. When affected, the otic capsule may show a diffuse lytic appearance with a smooth resorption or sclerosis in the late phase. Moreover, pagetic involvement of the bony labyrinth is often asymmetric, in contradistinction to the pattern of OI or otosclerosis. The patient’s age (36 years) mitigates against Paget disease, which affects an older population. Paget disease, therefore, is not the most likely diagnosis in this case.

Otosyphilis (luetic involvement of temporal bone) is a rare disease, but its occurrence has risen with the acquired immunodeficiency syndrome, or AIDS, epidemic (18). It can manifest as labyrinthitis, gummatous lesion of the internal auditory canal, and inflammatory resorptive osteitis (18–21). Luetic involvement of the temporal bone manifests as a moth-eaten permeative appearance and can cause secondary involvement of the membranous labyrinth in the late congenital, late latent, and tertiary stages of syphilis. Histologically, this osteitis features round cell infiltration, multinucleated giant cells, and endarteritis, all of which result in a varying degree of bony labyrinth destruction (20). Otosyphilis, therefore, may simulate the demineralized appearance of OI and otosclerosis, but it is typically accompanied by systemic manifestation of syphilitic infection.

Perhaps a more frequently encountered disease than otosyphilis that may simulate the imaging findings in this case would be bilateral lytic osseous metastases. The bilateral symmetric pattern of otic capsule involvement, with sparing of the remaining skull and temporal bone, would, however, be unusual.
On the basis of the history of discolored (blue) scleras, the description of the patient’s appearance (short stature and wheelchair bound), and the symmetric bilateral temporal bone findings, OI is the most likely diagnosis.

References
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