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Case report

MRI-visible pericochlear lesions in osteogenesis imperfecta type I

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Abstract. Osteogenesis imperfecta (OI) is an inherited generalized disorder of type-I collagen synthesis often associated with hearing loss. We present a case of OI type I in which hearing loss led to examination of the temporal bone with MRI. In the osseous otic capsule MRI demonstrated pericochlear lesions with soft tissue signal intensity and contrast enhancement. Changes similar to otosclerosis have been described in the temporal bone of OI patients when applying CT, but reports on MRI findings do not yet exist.

Key words: Osteogenesis imperfecta – MR imaging – Temporal bone – Otosclerosis

Introduction

Osteogenesis imperfecta (OI) is an inherited disorder of type-I collagen synthesis. The current clinical classification [1] divides the disorder into four types. The common feature of all types is abnormal bone fragility. The autosomal dominantly inherited type I manifests with fractures, blue sclerae, osteoporosis, and in a large proportion of patients with hearing loss. Type IV differs from type I in a lower frequency of hearing loss and normal sclerae. Both types are divided into two subgroups, with and without deficient dentition, respectively. Type II, with autosomal-recessive inheritance, represents the most severe type leading to death in the perinatal period due to extreme bone fragility. The autosomal-recessive or dominant type III leads to progressive limb and spine deformity, short stature, kyphoscoliosis, and triangular face.

The reported incidence of hearing loss in OI ranges from 20 to 60% [2, 3], with the highest incidence in type I [4]. In OI type I hearing loss usually starts in the second or third decade and may be conductive, sensorineural, or combined, the combined type being the most frequent one [3, 4].

In this paper we describe MRI findings in the osseous otic capsule associated with hearing loss in a patient with OI type I.

Otosclerosis (OS) is an osteodystrophy of unknown etiology restricted to the human osseous otic capsule. Otosclerosis most frequently affects the anterior portion of the oval window, with possible involvement of the stapes footplate and the cochlea. In OS the earliest changes comprise osteolytic lesions in the enchondral layer of the otic capsule, resulting in disorganized formation of woven bone with marrow spaces containing blood vessels and connective tissue [5]. This active stage of the disease is described by the term otospongiosis. In time these lesions undergo remineralization and reorganization to sclerotic lamellar bone [6]. Histopathology reveals similarities in OI and active OS, OI resembling extensive cases of OS with pericochlear involvement.

The aim of this study was to compare the MRI findings in our OI patient with data of patients with active retrofenestral OS.

Case report

A 59-year-old female patient demonstrated the clinical picture of OI type I with frequent fractures during childhood, osteoporosis, scoliosis, blue sclerae, and occurrence of the disorder in several siblings. She had progressive hearing loss since adolescence resulting in deafness on her right ear 7 years previously. Speech perception had still been possible with a hearing aid on her left ear. Because of acute anacusis on her left ear, MRI was requested to exclude a cerebello-pontine angle tumor.

Magnetic resonance imaging was performed on a 1.0-T scanner. Three-millimeter proton-density-, T2-, and T1-weighted spin-echo images prior to and after contrast administration (0.1 mmol/kg Gd-DTPA) were acquired in the axial and coronal planes.

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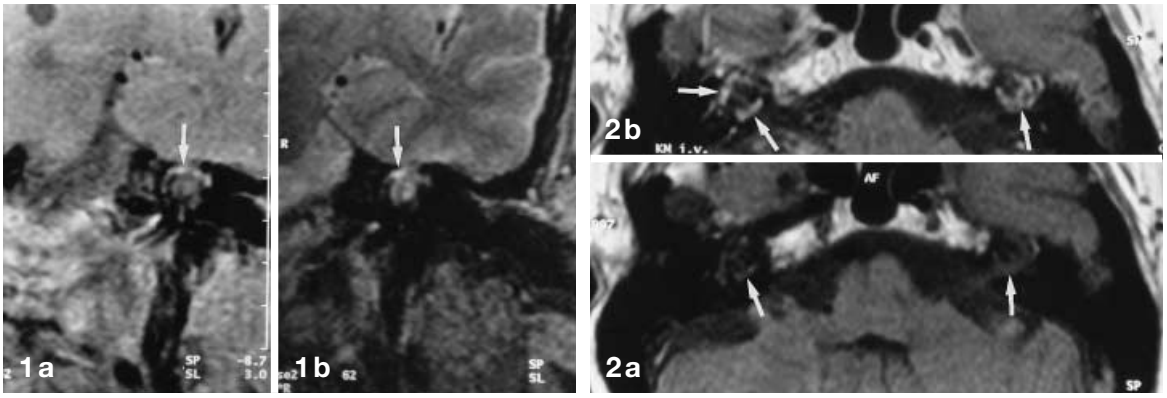

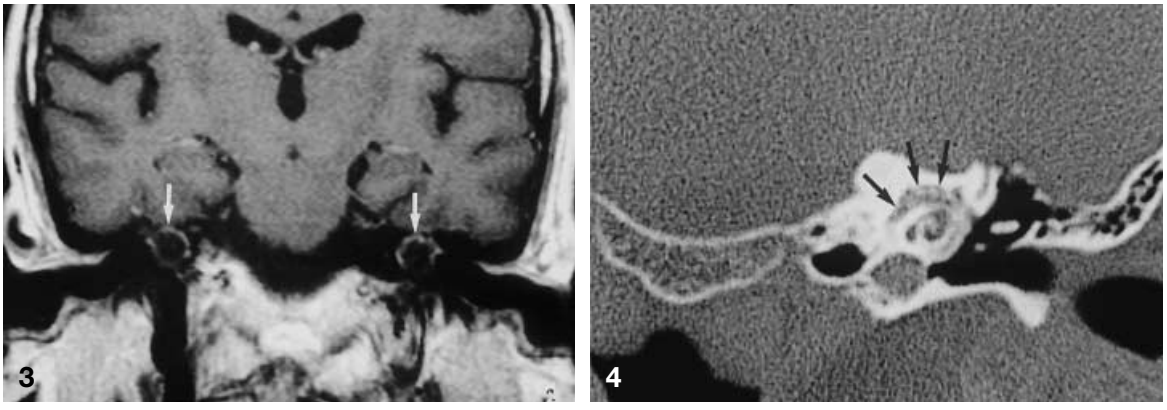

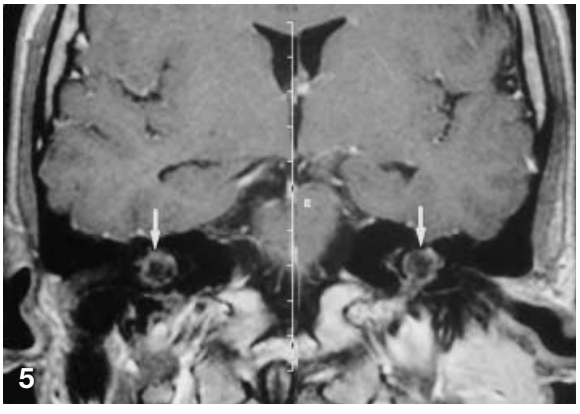

Fig. 1. **a** Coronal proton-density-weighted and **b** T2-weighted images of the left ear in osteogenesis imperfecta (OI). Pericochlear lesions with soft tissue signal intensity are seen *(arrows)*

Fig. 2. Transversal T1-weighted MRI **a** pre-contrast and **b** gadolinium-enhanced in OI. Bilateral bands of soft tissue signal with contrast enhancement in the otic capsule are seen *(arrows)*

Fig. 3. Coronal T1-weighted Gd-enhanced MRI in OI; bilateral ring-like enhancing soft tissue lesions surrounding the cochlea *(arrows)*

Fig. 4. Corresponding coronal CT of the left otic capsule in OI; demineralized pericochlear lesion *(arrows)*

Fig. 5. Coronal T1-weighted Gd-enhanced MRI in otospongiosis. Bilateral pericochlear ring-like enhancing soft tissue lesions *(arrows)* are indistinguishable from findings in OI

Magnetic resonance imaging demonstrated bilateral pericochlear lesions with soft tissue signal and moderate contrast enhancement (Figs. 1, 2). These lesions extended into the oval window region. The highest conspicuity was achieved with T1-weighted gadolinium-enhanced sequences in the coronal plane (Fig. 3). Additionally performed high-resolution CT with 1-mm sections of the inner ear in the axial and coronal planes showed corresponding ring-like lesions of demineralization in the otic capsule (Fig. 4).

The MRI findings in OI were compared with our own published MRI data from 5 patients with otospongiosis [7]. The MRI findings in OI were the same as those in otospongiosis with regard to signal characteristics and location (Fig. 5).

Discussion

Hearing loss is a frequent symptom in OI. Imaging may be indicated in atypical cases, e. g., to exclude a cerebello-pontine angle tumor. Some patients with OI show a benign clinical course, thus remaining undiagnosed. These patients may be referred to cross-sectional imaging because of hearing loss [8].

When applying CT pericochlear lesions of demineralization, comparable to fenestral and retrofenestral manifestations of otospongiosis can be demonstrated [8, 9, 10].

In a previous paper we described the capability of MRI to show soft tissue signal and contrast enhancement in affected areas of the otic capsule and oval window region in the active phase of OS [7]. In the otospongiotic bone these findings correspond to an expansion of marrow spaces containing a fibro-vascular stroma [5].

With regard to lesions in the temporal bone, histopathological similarities between OI and OS were described as early as 1922 by Ruttin [11]. In the non-congenital, autosomal dominantly inherited OI types I and IV, histopathological studies found OS-like lesions in the stapes footplate and osseous cochlear caspsule [3]. The coexistence of OS-like lesions and deficient osteoid formation as a typical reflection of OI has also been described [12].

As a consequence of audiological, histopathological, and radiological similarities, the pathogenetic and etiologic relationships of OI and OS are still under discussion (for review see [3]). In a recent paper collagen gene mutations similar to those in OI were detected in OS patients, a finding that might indicate common genetic factors [13].

Taking into account the histopathological similarities of OI and active OS, it was not very surprising to detect identical changes in MRI; however, until now MRI findings of the inner ear in OI have not been described in the literature.

In previously undiagnosed patients with mild OI, presenting with hearing loss, MRI of the temporal bone may be the clue in establishing the correct diagnosis.

MRI may play an important role in monitoring therapeutic attempts to influence the otological manifestations of OI.

In conclusion, MRI is capable of detecting involvement of the osseous otic capsule in osteogenesis imperfecta.

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