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Progressive Conductive Hearing Loss in Osteogenesis Imperfecta (Lobstein Disease): A Case Report

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ABSTRACT

Background: Osteogenesis imperfecta (OI), also known as Lobstein disease, is a rare inherited connective tissue disorder characterized by bone fragility and various extra-skeletal manifestations. Hearing loss is a frequent but often underestimated complication that may significantly impair quality of life. Case presentation: We report the case of a 34-year-old woman with clinically established osteogenesis imperfecta type I who developed progressive bilateral conductive hearing loss over a three-year period. Audiological evaluation revealed symmetrical conductive impairment with preserved speech discrimination. High-resolution temporal bone computed tomography (CT) demonstrated diffuse otic capsule demineralization, bilateral stapes footplate thickening, and atypical crown-shaped hypodense lesions surrounding the cochlea. Conclusion: Early recognition of hearing loss in osteogenesis imperfecta, combined with detailed imaging and multidisciplinary management, is essential to optimize functional outcomes.

KEYWORDS

osteogenesis imperfecta, lobstein disease, conductive hearing loss, temporal bone CT, case report.

1 Introduction

Osteogenesis imperfecta (OI) is a heterogeneous inherited connective tissue disorder caused by defective synthesis of type I collagen, most commonly associated with mutations in the *COL1A1* and *COL1A2* genes (Kuurila et al., 2003). Its estimated prevalence ranges from 1 in 15,000 to 1 in 20,000 live births (Macêdo et al., 2024).

Beyond skeletal fragility, recurrent fractures, and blue sclerae, hearing loss represents one of the most frequent extra-skeletal manifestations of OI (Waissbluth et al., 2020). Hearing impairment affects approximately 33% to 70% of patients and typically develops during early adulthood, initially presenting as conductive hearing loss before progressing to mixed or sensorineural patterns (Ugarteburu et al., 2022; Macêdo et al., 2024).

Because of overlapping clinical and radiological features, OI-related conductive hearing loss may be misdiagnosed as otosclerosis, which has important implications for management and surgical risk (Ugarteburu et al., 2022). This case report highlights the diagnostic challenges, radiological characteristics, and therapeutic considerations of hearing loss in osteogenesis imperfecta.

2 Case Presentation

A 34-year-old woman with a longstanding diagnosis of osteogenesis imperfecta type I was referred to our otolaryngology department for progressive bilateral hearing loss evolving over three years. The hearing impairment was insidious in onset and gradually worsening, without associated tinnitus, vertigo, otalgia, or otorrhea.

Her medical history was notable for multiple low-energy fractures during childhood and adolescence, consistent with mild OI. Several first-degree relatives were also affected. The patient was under regular orthopedic follow-up and had received standard medical management including calcium and vitamin D supplementation. She reported no history of chronic otitis media, otologic surgery, or acoustic trauma.

General examination revealed blue sclerae. Otoscopic examination showed normal, intact tympanic membranes bilaterally, without signs of middle ear pathology (Figure 1).

Pure-tone audiometry demonstrated bilateral conductive hearing loss, with an air–bone gap of approximately 30 dB in the right ear and 20 dB in the left ear (Figure 2). Speech discrimination scores were preserved. Tympanometry showed normal middle ear compliance, and stapedial reflexes were present bilaterally.

High-resolution temporal bone CT demonstrated bilateral perichochlear "crown-shaped" hypodensities and thickened stapes footplates (Figure. 3). The hypodense foci were described between the oval window and the vestibule, spreading in a crown-like distribution around the cochlea. Because footplate thickening and fenestral involvement may also be seen in otosclerosis, otosclerosis

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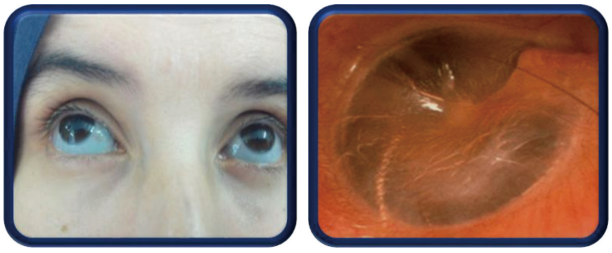


Figure 1 Clinical features showing blue sclerae and normal bilateral otoscopic examination with intact tympanic membranes.

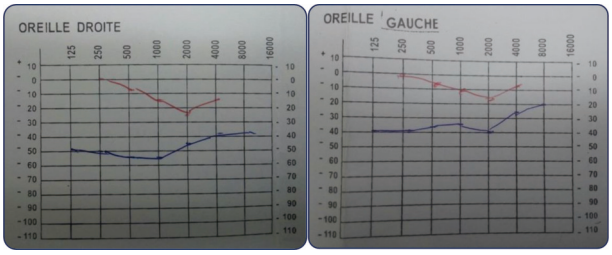


Figure 2 Pure-tone audiometry demonstrating bilateral conductive hearing loss, with an air–bone gap of approximately 30 dB in the right ear and 20 dB in the left ear.

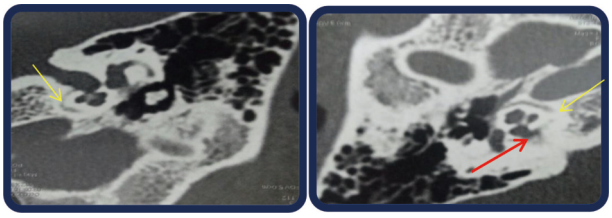


Figure 3 Axial and coronal high-resolution temporal bone CT images showing diffuse otic capsule demineralization with crown-shaped hypodense foci surrounding the cochlea and bilateral thickening of the stapes footplates (arrows).

was considered in the differential diagnosis; however, the systemic phenotype (OI features and family history) and the diffuse peri-cochlear pattern favored OI-related temporal bone involvement.

Molecular testing for COL1A1/COL1A2 was not performed at the time of evaluation. The diagnosis of osteogenesis imperfecta type I was therefore established on clinical grounds, based on the association of recurrent low-energy fractures, blue sclerae, positive family history, and characteristic temporal bone CT findings. Although genetic confirmation may refine prognostic stratification and familial counseling, the absence of molecular testing does not preclude a reliable clinical diagnosis in typical cases, particularly in resource-limited settings. This limitation was discussed with the patient, and genetic counseling was proposed.

3 Discussion

Hearing loss in osteogenesis imperfecta results from abnormal type I collagen synthesis and defective bone remodeling affecting the ossicular chain and otic capsule (Claeys et al., 2023). The association of bone fragility, blue sclerae, and hearing impairment—known as Van der Hoeve syndrome—typically manifests during the second to fourth decades of life (Kuurila et al., 2003).

Otosclerosis represents the principal differential diagnosis in adults presenting with conductive hearing loss and stapes footplate involvement (Ugarteburu et al., 2022). However, otosclerosis is characterized by focal otospongiotic lesions, most commonly involving the fissula ante fenestram, whereas OI-related otological involvement produces diffuse and often bilateral demineralization of the otic capsule (Kuurila et al., 2003).

In contrast, osteogenesis imperfecta produces diffuse and often bilateral demineralization of the otic capsule, reflecting generalized abnormal bone remodeling rather than localized otic pathology. In the present case, the bilateral peri-cochlear "crown-shaped" hypodensities extending beyond the fenestral region strongly favored OI-related temporal bone involvement over otosclerosis (Claeys et al., 2023).

Paget's disease of bone represents another differential diagnosis; however, it typically affects older individuals and demonstrates marked bone enlargement, cortical thickening, and a characteristic mosaic pattern on imaging. The absence of these radiological features, combined with the presence of classical OI stigmata and family history, made Paget's disease unlikely in this patient (Claeys et al., 2023).

High-resolution CT is the imaging modality of choice for evaluating hearing loss in osteogenesis imperfecta (Ugarteburu et al., 2022). Reported temporal bone findings include ossicular chain fragility, stapes fixation, cochlear cavitation, and diffuse otic capsule hypodensity (Kuurila et al., 2003). The crown-shaped peri-cochlear lesions observed in our patient are uncommon and may expand the radiological spectrum of OI-related temporal bone involvement.

Magnetic resonance imaging was not performed in this case because there was no evidence of sensorineural hearing loss or retrocochlear symptoms. Nevertheless, MRI may be useful during long-term follow-up to assess potential cochlear or neural involvement (Waissbluth et al., 2020).

Tympanometry revealed preserved stapedial reflexes despite conductive hearing loss. This finding, although atypical for classical otosclerosis, has been reported in osteogenesis imperfecta and may be explained by partial stapes fixation, ligamentous laxity, or early ossicular chain involvement rather than complete footplate ankylosis. Sufficient ossicular mobility may persist to allow reflex activation, particularly in early or intermediate stages of the disease, and may therefore represent a useful diagnostic clue favoring OI over otosclerosis (Waissbluth et al., 2020).

Management of hearing loss in osteogenesis imperfecta must be individualized (Ugarteburu et al., 2022). Hearing aids are considered first-line therapy for mild to moderate conductive hearing loss (Waissbluth et al., 2020). Stapes surgery may provide hearing improvement but is associated with higher complication rates than in otosclerosis, including footplate fracture, perilymphatic gusher, and prosthesis instability due to bone fragility (Ugarteburu et al., 2022).

In the present case, surgical intervention was deferred because hearing thresholds were relatively stable, amplification provided satisfactory benefit, and surgical risks were considered significant. Implantable hearing devices, including cochlear implants, represent effective options in advanced cases when appropriately selected, despite technical challenges related to bone fragility (Macêdo et al., 2024).

Management of hearing loss in osteogenesis imperfecta must be individualized (Ugarteburu et al., 2022). In the present case, surgical intervention was deliberately deferred due to relatively stable hearing thresholds, preserved speech discrimination, satisfactory benefit from hearing aids, and the increased risk of intraoperative

Table 1 Comparison between Osteogenesis Imperfecta–related hearing loss and Otosclerosis

Feature	Osteogenesis Imperfecta	Otosclerosis
Age at onset	Childhood to early adulthood	Young to middle adulthood
Laterality	Often bilateral	Often bilateral, sometimes asymmetric
Audiometric pattern	Conductive → mixed / SNHL	Predominantly conductive
Stapedial reflexes	**May be preserved**	Typically absent
CT findings	**Diffuse otic capsule demineralization, peri-cochlear hypodensity**	Focal fenestral lesions
Bone quality	Fragile, abnormal collagen	Localized otic bone remodeling
Surgical risk	**High**	Lower
Management	Hearing aids, cautious surgery	Stapedotomy/stapedectomy

complications in OI, including footplate fracture, perilymphatic gusher, and prosthesis instability.

Long-term audiological follow-up was planned to monitor progression toward mixed or sensorineural hearing loss. In advanced cases, implantable hearing solutions, including bone-anchored devices or cochlear implantation, may be considered, despite technical challenges related to bone fragility (Macêdo et al., 2024).

4 Conclusion

Hearing loss is a frequent, progressive, and clinically significant manifestation of osteogenesis imperfecta. Accurate differentiation from otosclerosis is essential, as it directly influences management strategies and surgical risk assessment. Regular audiological monitoring, detailed radiological evaluation, and individualized multidisciplinary management remain the cornerstones of optimal care.

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Data Availability

The data supporting the findings of this study are available from the corresponding author upon reasonable request, in accordance with patient confidentiality and ethical regulations.

Ethical approval

No ethical approval was required for this case report as per institutional guidelines. All measures were taken to ensure patient confidentiality.

Informed consent

The patient provided informed, written consent for publication of this case report and all accompanying images and data.

Conflicts of interest

The authors declare no conflicts of interest.

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