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Clinical Case Reports

Otosclerosis: Presentation, Diagnosis And Management

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Abstract

Summary: Hearing loss profoundly affects quality of life, limiting communication, social interaction, and occupational performance. It is also associated with emotional distress, social isolation, and an increased risk of cognitive decline when left untreated. This case report describes a young male presenting to the ENT OPD with progressive hearing loss in bilateral ear. After exclusion of all associated symptoms and previous history, etiology was sought via otoscopy, revealing a perfectly mobile and normal-looking tympanic membrane. PTA and tympanometry revealed bilateral carhart notch and A-type curve on tympanometry, respectively, making the diagnosis of clinical otosclerosis. Patient underwent exploratory tympanotomy and Stapedotomy with a prosthesis. Follow-up PTA showed closure of air bone gap.

Otosclerosis, though benign, is twice as common in females than male. This case illustrates the classical presentation of otosclerosis in a male patient and highlights the role of keen history taking and high clinical suspicion in a male patient where otosclerosis is often missed as a differential diagnosis of progressive hearing loss, resulting in diagnostic delay, making it a clinical rarity, and hence it is selected as a case report.

Keywords: Otosclerosis, Deafness, Quality Of Life, Tympanic Membrane.

Introduction

Otosclerosis is a localized disorder of bone metabolism affecting the otic capsule, characterized by abnormal resorption and deposition of endochondral bone, most commonly around the stapes footplate. This pathological process leads to stapes fixation and progressive conductive hearing loss, which may develop a mixed or sensorineural component in advanced cases. The condition typically presents in early to middle adulthood and accounts for 5–9% of adult hearing loss in Caucasian populations, with a lower but variable prevalence in Asian cohorts.[1,2] Clinically, otosclerosis manifests as gradually progressive bilateral hearing loss, while tinnitus and vertigo are less common. The two principal forms are fenestral otosclerosis, limited to the stapes footplate and oval window, and cochlear otosclerosis, which involves the otic capsule and may contribute to sensorineural deficits.[3,4]

The present case is reported to highlight the classical presentation of bilateral fenestral otosclerosis in a young adult male, emphasizing the diagnostic role of audiometric and tympanometric assessments, and to demonstrate the effectiveness of stapedotomy with piston prosthesis in restoring hearing thresholds. Such case documentation is of clinical relevance in regions where otosclerosis may be underdiagnosed due to limited access to specialized otological services and radiological investigations, and serves to reinforce surgical intervention as a definitive treatment modality with favorable outcomes.

Case Presentation

A 32-year-old unmarried male shopkeeper presented to the ENT outpatient clinic. He belonged to a lower-middle socioeconomic stratum and was the sole provider for his family. His past medical history was noncontributory, except for an appendectomy performed in 2016. He reported a history of pollen allergy and a smoking history of seven pack-years. There was no history of major systemic illness or trauma. Family history was significant only for maternal hypertension.

History of Presenting Complaint

The patient reported progressive bilateral hearing impairment for the past 10 years. The hearing loss was insidious in onset, gradually progressive, and non-episodic. There were no associated otological or vestibular symptoms, including tinnitus, vertigo, otalgia, otorrhea, or aural fullness. He denied any preceding upper respiratory tract infections, head trauma, or prior ear surgery.

Examination

General physical examination revealed a healthy adult male with stable vital parameters and no stigmata of chronic systemic illness. Local otological examination demonstrated normal auricles and external auditory canals. The tympanic membranes were intact, pearly in appearance (figure 1), and demonstrated a well-visualized cone of light bilaterally. Tuning fork tests revealed a negative Rinne bilaterally, with Weber lateralized centrally, and absolute bone conduction comparable to the examiner, consistent with a conductive hearing deficit. The fistula test was negative, and there was no evidence of facial nerve dysfunction. Comprehensive examination of the nasal cavity, oral cavity, oropharynx, and larynx was unremarkable. Systemic examination, including cardiovascular, respiratory, abdominal, and neurological assessments, was within normal limits.

Investigations

Routine hematological investigations were within reference ranges. Audiometric evaluation demonstrated bilateral conductive hearing loss on pure tone audiometry(Figure 2) as well as a carhart notch seen at 2kHz. Tympanometry revealed findings compatible with stapes fixation(figure 3), while acoustic reflex testing confirmed absent reflexes(figure 4).

Contributions:
KM SC SM AR - Conception, Design
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Differential diagnosis:

Based on the clinical and audiological profile, the differential diagnoses considered included serous otitis media, adhesive otitis media, tympanosclerosis, and congenital stapes fixation.

A working diagnosis of **otosclerosis** was established on the basis of clinical suspicion corroborated by audiometric and tympanometric findings

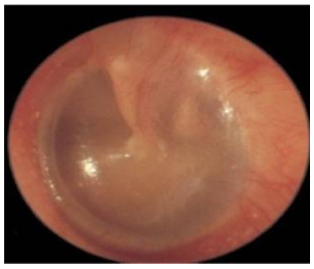

Figure 1: Showing shiny and intact left tympanic membrane with no retraction and cone of light present at 7'o clock position as it appeared on otoscopy

Figure 2: PTA of patient showing bilateral air bone gap of 30dB and presence of carhart notch at 2kHz suggestive of bilateral otosclerosis

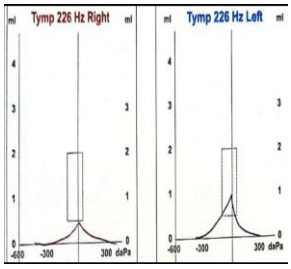

Figure 3: showing low compliance and normal pressure & volume on tympanometry of right ear plotted as As type curve. Left ear tympanometry appears to be normal

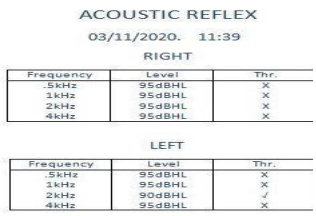

Figure 4: shows absent acoustic reflex on both sides

Management

The patient underwent a right stapedotomy with insertion of a Teflon piston of size 4.5mm prosthesis under general anesthesia. A tympanomeatal flap was elevated, and an exploratory tympanotomy was done to assess middle ear structures. Stapes food plate was found fixed to the oval window while the rest of the ossicles were intact and mobile(ruling out malleolus fracture, congenital stapes fixation). A fenestration was created in the stapes footplate. A piston prosthesis was then placed across the oval window, followed by closure of the operative site(figure 5 A-E). The intraoperative and immediate postoperative courses were uneventful.

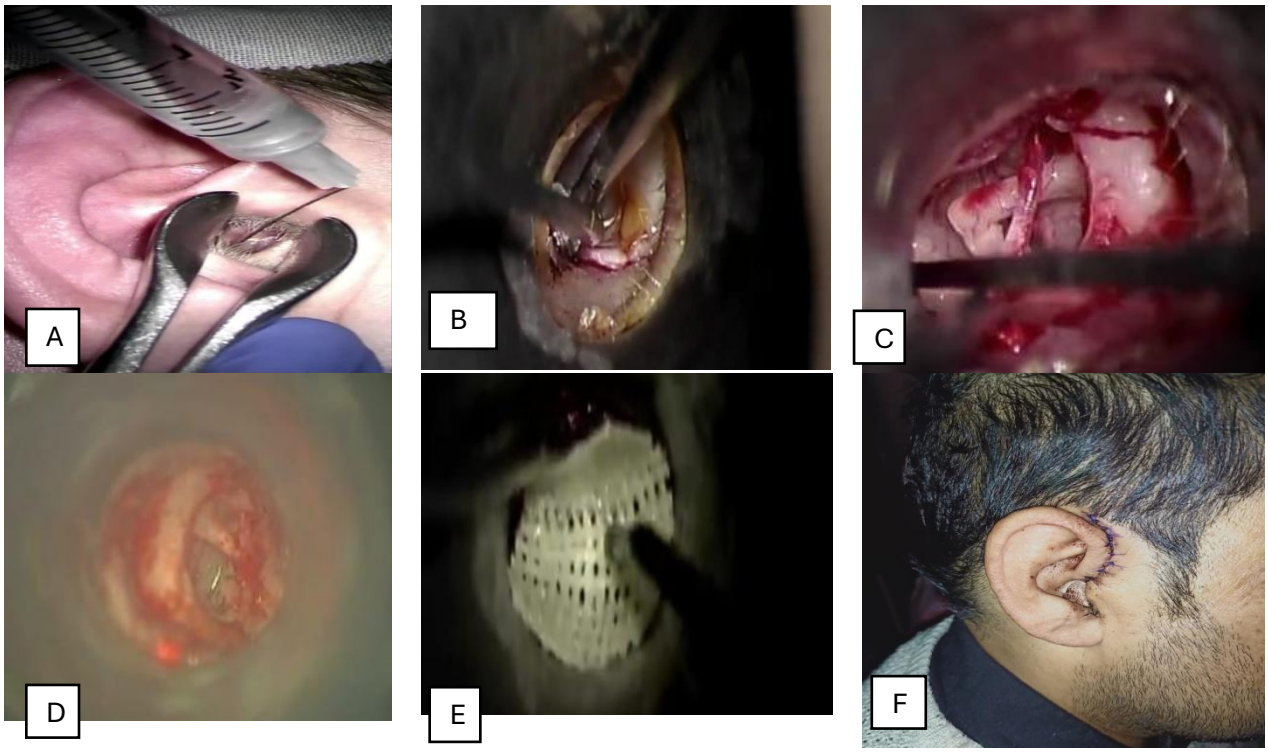

Figure 5: Steps of surgery are as follows:(A) Local anaesthesia given. (B) tympanomeatal flap elevated. (C) shows middle ear structures visualized and per-op diagnosis of otosclerosis confirmed. (D) fenestration made in stapes foot plate piston inserted. (E) shows wound closed. (F) post op scar

Outcome

The patient demonstrated satisfactory recovery with no early postoperative complications. Follow-up pure tone audiometry after 3 months revealed a significant improvement in air conduction thresholds, closure of ABG, and diminished carhart notch, which was consistent with a successful functional outcome (**Figure 6**). He reported subjective improvement in hearing acuity, confirming the effectiveness of stapes surgery in this case.

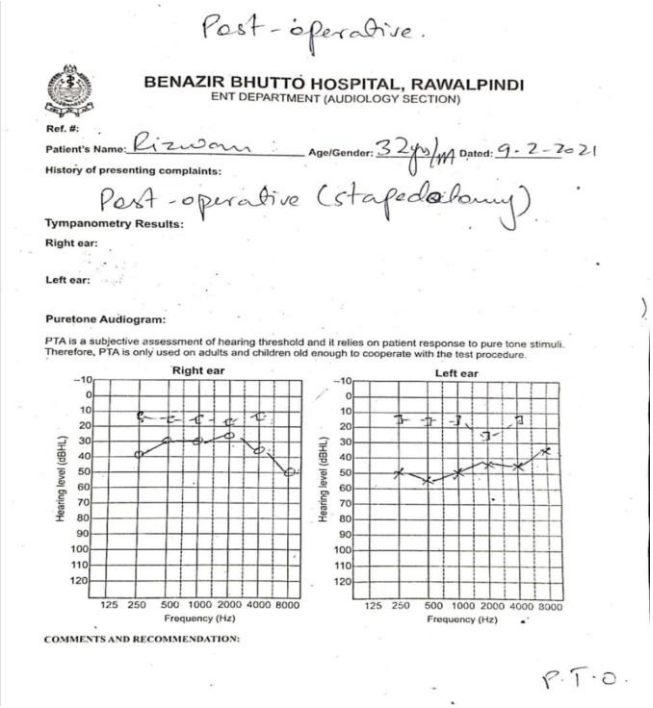

Figure 6: Post-op audiogram shows closure of air-bone gap on operated(right) ear as well as no carhart notch formed

Discussion

Otosclerosis is a progressive osteodystrophic disorder marked by abnormal bone remodeling within the otic capsule and is a leading cause of acquired hearing loss, with histological evidence in approximately 10% of Caucasians.[5] The condition primarily induces conductive hearing loss due to stapes fixation, but sensorineural hearing loss may also occur, potentially resulting from disrupted inner ear fluid homeostasis or dysfunction of the cochlear lateral wall fibrocyte network.[6] Recent synchrotron radiation phase-contrast imaging has revealed that otosclerotic plaques can form shunts and abnormal vascular connections to the labyrinth, leading to hypervascularization, venous overload, and subsequent sensorineural hearing loss.[7] Histologically, otosclerosis is present in 12% of white individuals, but only 0.3–0.4% develop clinical symptoms.[8] Prevalence is lower among Black, Asian, and Native American populations. The average age of onset is around 30 years, and the female-to-male ratio is approximately 1.5–2:1.[8]

To date, only two genome-wide association studies have examined otosclerosis, with the latest identifying 18 significant loci. Targeted resequencing has found 13 variants in five genes—MARK3, LINC01482, AHSG, SUPT3H, and RELN—implicated in bone metabolism, chromatin remodeling, and extracellular matrix regulation, suggesting both protective and pathogenic roles. Variants in TGFβ1 and EYA2 were not significantly associated after correction, though TGFβ1 pathways remain relevant.[9]

Environmental factors, notably viral infections such as measles, are implicated in otosclerosis pathogenesis, possibly by triggering autoimmune responses that disrupt bone remodeling.[10] Hormonal influences, particularly estrogen, have been proposed, given the higher prevalence in females and during periods of hormonal fluctuation such as pregnancy. However, a large case-control study of 1,196 women found no significant association between pregnancy and otosclerosis.[11,12]

Normal bone remodeling occurs at a rate of 10% per year in the skeleton, but the otic capsule remodels at only 0.13% per year.[13] In otosclerosis, this rate increases, resulting in bone deposition that impairs auditory structures and sound transmission. Lesions most commonly occur anterior to the oval window and stapes footplate (80%), but can also affect the round window (30%), pericochlear region (21%), and anterior internal auditory canal (19%).[14]

Clinically, patients experience gradual hearing loss, especially at low frequencies, making it difficult to perceive male voices or vowel sounds. Nearly half report tinnitus, while vertigo is rare (10%) and typically occurs only with vestibular involvement.[8,15] Bilateral disease develops in 80% of cases, though initial presentation is often unilateral.[16]

Otoscopic examination is usually normal, except for the Schwartz sign—reddening of the promontory—seen in about 20% of cases, indicating active osteosclerotic activity, but it is not required for diagnosis.[17] Diagnosis relies on clinical history, examination, and audiometry. Audiometry assesses air and bone conduction, with thresholds above 25 dB considered abnormal. Conductive loss at low frequencies and the Carhart notch at 2,000 Hz are classic findings, though the latter is not a reliable diagnostic marker. Audiometry is also valuable for monitoring disease progression. Early disease causes ossicular stiffening and a small air–bone gap, which widens with stapes fixation. Cochlear involvement in about 10% of patients leads to high-frequency sensorineural loss and a mixed hearing pattern. Tympanometry is typically normal, except in advanced cases with marked ossicular fixation.

High-resolution CT is the first-line investigation for identifying otosclerotic foci, with diagnostic accuracy exceeding 90% in some studies, and can help differentiate other middle ear pathologies. The degree of hearing loss may correlate with the size of otosclerotic lesions, though some

studies report no significant correlation between CT and audiometric findings, suggesting audiometry alone may suffice when CT is unavailable.[18-20]

Medical management aims to slow abnormal bone remodeling. Bisphosphonates and sodium fluoride have shown potential for stabilizing hearing and slowing progression in observational studies and small trials, but large randomized trials are lacking. Systematic reviews indicate that at least six months of sodium fluoride can stabilize hearing thresholds, improve vestibular symptoms, and delay tinnitus progression. Bisphosphonates administered for at least six months have demonstrated improvements in hearing, dizziness, and tinnitus remission, with double-blind studies showing greater stabilization of hearing thresholds compared to placebo.[21]

Surgical advances have refined both traditional and novel techniques. Stapedotomy remains the gold standard, providing significant improvement in air conduction and speech discrimination with low complication rates in experienced hands.[22] Lasers and microdrills have enhanced precision in footplate fenestration, though superiority over conventional methods is still under investigation.[23] Preoperative high-resolution CT is now routinely used for surgical planning. For advanced cases with profound sensorineural loss, cochlear implantation is effective, even with cochlear ossification, and outcomes are improved by technical modifications and electrode selection.[24] Endoscopic approaches offer better visualization and may reduce morbidity, while 3D-printed models are being developed to improve surgical training.[25] Ultimately, the choice between stapes surgery and cochlear implantation is individualized, based on disease extent, hearing profile, and patient factors, with both approaches demonstrating high success rates in recent studies.

Conclusion

This case report emphasizes detailed, careful history taking, examination, and keeping otosclerosis as part of the differentials when there is progressive conductive hearing loss. Audiometry can help diagnose as well as monitor disease progression. Despite the availability of radiological and audiometry facilities, a trained mind with a caring heart is the foundation of patient care.

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