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When Bone Forms Beyond Bone: An Otologic Diagnostic Dilemma in a 28-Year-Old Woman with Fibrodysplasia Ossificans Progressiva (Stone Man Syndrome)

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ABSTRACT

Objective: To describe the audiologic findings and diagnostic challenges of hearing loss in a patient with fibrodysplasia ossificans progressiva (FOP).

Methods:

Design: Case Report
Setting: Tertiary Private University Training Hospital
Patient: One

Results: A 28-year-old Filipino woman with a known diagnosis of FOP presented with a 12-year duration of bilateral hearing loss. Otoscopic examination demonstrated patent external auditory canals and intact, non-hyperemic tympanic membranes bilaterally. Pure-tone audiometry revealed moderate sensorineural hearing loss in the right ear and severe mixed hearing loss in the left ear. Tympanometry showed a Type As tracing on the right and a Type B tracing on the left. High-resolution computed tomography of the temporal bones showed no ossicular chain fixation, or middle ear mass, failing to demonstrate a structural correlate for the degree of hearing loss observed. The use of hearing aids for amplification provided benefit.

Conclusion: This case highlights the complexity of evaluating hearing loss in FOP, where audiometric and immittance findings may not correlate with otoscopic or radiologic features. Recognition of this variability is essential to avoid unnecessary invasive surgical interventions and to emphasize conservative management, including hearing rehabilitation. FOP-related hearing loss likely represents a multifactorial process involving ossicular stiffness, altered middle ear compliance, and potential cochlear biomechanical dysfunction. High-resolution imaging may underestimate middle ear pathology, necessitating reliance on comprehensive audiologic assessment. Surgical intervention should be approached with extreme caution due to the risk of heterotopic ossification flare-ups.

Keywords: *fibrodysplasia ossificans progressiva; stoneman syndrome; hearing loss*

Fibrodysplasia ossificans progressiva (FOP) is a rare genetic disorder characterized by progressive heterotopic ossification of soft tissues, resulting in cumulative functional impairment.[1] The estimated worldwide prevalence is approximately one in two million individuals, with limited reports from Southeast Asia, including the Philippines.[2] Given its rarity, the full spectrum of clinical manifestations including otologic involvement may be underrecognized.

Hearing loss has been described in individuals with FOP and is commonly attributed to conductive mechanisms related to middle ear or ossicular chain involvement.[2,3] However, audiologic presentations are variable, and formal hearing assessment is essential for accurate characterization of auditory function.

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We present the case of a 28-year-old woman with a known diagnosis of FOP who developed progressive bilateral hearing loss and describe the audiologic findings and diagnostic challenges of hearing loss in her case.

CASE REPORT

A 28-year-old Filipino woman with a known diagnosis of fibrodysplasia ossificans progressiva (FOP) presented with progressive bilateral hearing loss over the past 12 years. She denied otalgia, otorrhea, vertigo, tinnitus, recurrent ear infections, previous otologic surgery, or significant noise exposure.

The diagnosis of FOP had been established during childhood based on progressive heterotopic ossification and characteristic skeletal deformities. *(Figure 1)* She reported marked limitation of cervical spine mobility but had not undergone prior otologic evaluation or intervention.

On physical examination, skeletal deformities and restricted range of motion consistent with FOP were noted. Otoscopic examination revealed bilaterally patent external auditory canals. The tympanic membranes were intact, non-hyperemic, and demonstrated preserved cone of light bilaterally. *(Figure 2)* There was no evidence of effusion, retraction or retrotympanic mass.

Pure-tone audiometry demonstrated moderate sensorineural hearing loss in the right ear and severe mixed hearing loss in the left ear. A 2000 Hz dip in the left ear bone conduction threshold was observed. *(Figure 3)* Speech discrimination scores were 88% in the right ear and 74% in the left ear.

Tympanometry showed a Type As tympanogram in the right ear, consistent with reduced middle ear compliance, and a Type B tympanogram in the left ear. Despite the flat tracing on the left, no clinical signs of middle ear effusion were present on otoscopy.

High-resolution computed tomography of the temporal bones revealed a high-riding left internal jugular bulb. The ossicular chain appeared intact, with no evidence of ossicular discontinuity, fixation, middle ear mass, or radiologically apparent heterotopic ossification. *(Figure 4)*

Given the absence of surgically correctable pathology and the risks associated with operative intervention in FOP, the patient was managed conservatively with bilateral hearing aids, resulting in functional auditory improvement.

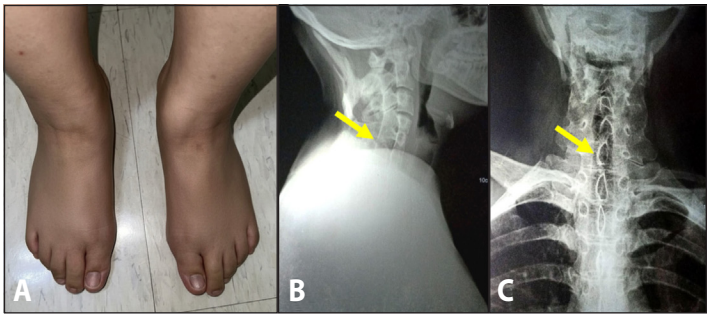


Figure 1. Other findings for FOP in this patient: **A.** congenital malformation of the great toes, short and malformed; **B.** cervical lateral x-ray showing straightening of cervical curvature (arrow); and **C.** cervical A-P view showing fusion of the bodies and posterior elements of C4-C6 (arrow)

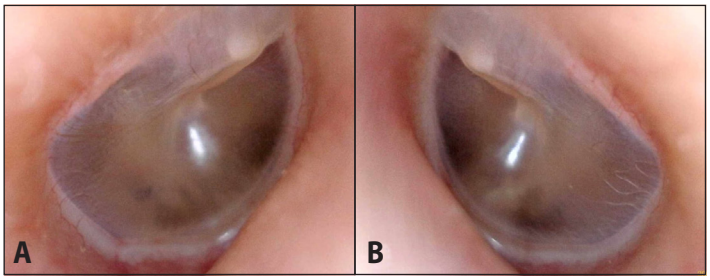


Figure 2. Otoscopy: **A.** right ear; and **B.** left ear, showing a patent external auditory canal and an intact tympanic membrane for both ears

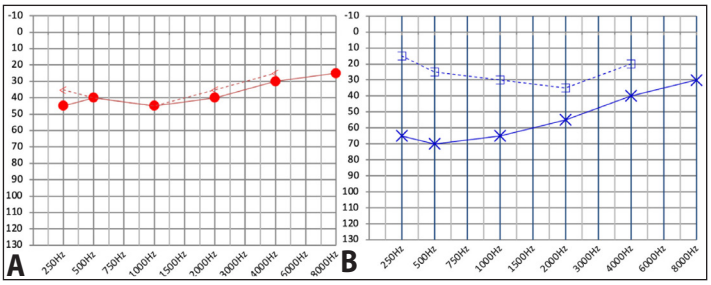


Figure 3. Pure tone audiometry demonstrating a moderate sensorineural hearing loss for the **A.** right ear and a severe mixed hearing loss for the **B.** left ear

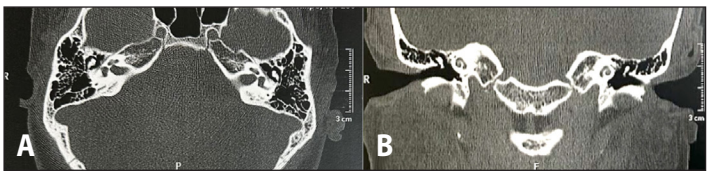


Figure 4. Temporal bone CT scans: **A.** axial (left), and **B.** coronal (right) views, demonstrating a clear middle ear cavity and mastoid antrum with normal ossicles, cochleas, vestibules and semicircular canals

DISCUSSION

FOP is a rare autosomal dominant disorder characterized by progressive heterotopic ossification of connective tissues.[1,2] Although musculoskeletal involvement predominates, otologic manifestations are increasingly recognized. Hearing loss in FOP may be conductive, sensorineural, or mixed, reflecting pathology at multiple levels of the auditory system.[3,4]

This case demonstrates asymmetric and discordant audiometric and immittance findings. The right ear showed moderate sensorineural hearing loss with a Type As tympanogram, suggesting reduced middle ear compliance. The left ear exhibited severe mixed hearing loss with a Type B tympanogram despite normal otoscopy and no radiologic evidence of effusion or ossicular discontinuity.

Conductive and mixed hearing loss in FOP are most commonly attributed to ossicular chain fixation.[3] Heterotopic ossification may involve the annular ligament, stapedial tendon, or ossicular joints, leading to progressive mechanical stiffening.[3] The Type As tracing may indicate early ossicular rigidity, whereas the flat Type B tympanogram

without effusion suggests markedly reduced tympanic membrane–ossicular mobility.

The 2000 Hz dip in the left ear bone conduction threshold is consistent with a Carhart notch, classically associated with stapes fixation. This pseudo-sensorineural depression results from altered ossicular resonance rather than true cochlear dysfunction and further supports mechanical impairment at the level of the stapes.[3] However, high-resolution computed tomography failed to demonstrate ossicular ankylosis or visible heterotopic bone formation, emphasizing that functional fixation may precede radiographically apparent ossification.[1,5]

The sensorineural component raises consideration of inner ear involvement. Although the otic capsule is typically resistant to remodeling, dysregulated bone morphogenetic protein signaling in FOP may alter otic capsule homeostasis.[1,2,6] Subtle changes in otic capsule stiffness or cochlear micromechanics may contribute to cochlear dysfunction without overt radiologic abnormalities.[4] The reduced speech discrimination score and asymmetric presentation suggest a multifactorial process involving both ossicular fixation and possible otic capsule remodeling. Audiometric variability in FOP has been previously reported.[3,7]

An incidental high-riding left internal jugular bulb was identified but without dehiscence or impingement, making its contribution uncertain.

Overall, this case highlights the diagnostic complexity of hearing loss in FOP. Discordance among audiometric, immittance, otoscopic, and radiologic findings suggests that both middle ear mechanics and inner ear biomechanics may be affected. Imaging may underestimate early or microscopic ossicular involvement.[1,5] Given the risk of trauma-induced heterotopic ossification, surgical exploration should be approached cautiously.[1,2] In the absence of clear surgically correctable pathology, conservative management with amplification and longitudinal monitoring remains the preferred strategy.

This case underscores the importance of multidisciplinary surveillance in patients with FOP. While musculoskeletal morbidity often dominates clinical attention, auditory dysfunction may significantly impair quality of life and functional independence.[8] Routine audiologic screening in FOP patients may therefore be warranted, even in the absence of overt otologic symptoms. Early identification of hearing impairment allows timely rehabilitation with amplification devices, which may substantially improve communication outcomes while avoiding invasive procedures.

Due to limited financial access to genetic testing, chromosomal analysis was not performed. As a result, molecular confirmation of the suspected FOP was not obtained, representing a limitation of this case. Diagnosis was based solely on clinical and radiologic findings. Conversely, this report contributes to the limited body of literature describing discordant audiometric and radiologic findings. The absence of radiographically demonstrable ossicular fixation despite functional evidence of middle ear stiffness suggests that current imaging modalities may lack sensitivity in detecting early or microscopic ossicular or ligamentous involvement. This highlights a potential area for future research. Greater understanding of the pathophysiologic mechanisms underlying auditory involvement may help refine diagnostic criteria, guide surveillance strategies, and augment the development of therapeutic interventions.

This case illustrates the diagnostic complexity of hearing loss in fibrodysplasia ossificans progressiva. The patient demonstrated asymmetric audiometric findings, including a mixed hearing loss with a Type B tympanogram in the absence of middle ear effusion and no radiologic evidence of ossicular fixation. The presence of a Carhart notch suggested stapes fixation, while the sensorineural component raised the possibility of otic capsule remodeling or cochlear biomechanical dysfunction.

Our findings highlight the potential discordance between audiologic, immittance, and radiologic evaluations in FOP. Imaging may underestimate early or microscopic ossicular involvement, and auditory dysfunction may be multifactorial. Given the risk of trauma-induced heterotopic ossification, surgical intervention should be approached with caution. Conservative management with amplification and longitudinal monitoring remains the preferred strategy when no clear surgically correctable pathology is identified.

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