CASE REPORT

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Bilateral congenital cholesteatoma presenting as a gradual-onset hearing loss in a young adult

Obostrani kongenitalni holesteatom ispoljen kao postepeni gubitak sluha kod mlade odrasle osobe

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Abstract

Introduction. Congenital cholesteatoma (CC) is a rare pathological condition of the middle ear characterized by keratinizing squamous epithelium located behind an intact tympanic membrane. Bilateral involvement is exceptionally uncommon and may present a significant diagnostic challenge, particularly in the absence of inflammatory symptoms. **Case report.** We present the case of a 19-year-old male with a 4-year history of gradually progressive bilateral hearing loss accompanied by tinnitus and otalgia, without otorrhea or prior otologic disease. Otomicroscopy revealed a pearly white mass visible through intact tympanic membranes bilaterally. Pure-tone audiometry identified severe bilateral conductive hearing loss. Multislice computed tomography of the temporal bones revealed bilateral soft-tissue masses within the middle ear cavities, along with associated ossicular erosion. Bilateral tympanomastoidectomy with ossiculoplasty was performed, and CC occupying the mesotympanum and epitympanum was confirmed intraoperatively. Postoperative follow-up with non-echo-planar diffusion-weighted magnetic resonance imaging, performed 2 years after surgery, showed no evidence of residual or recurrent disease. **Conclusion.** This case emphasizes the importance of considering CC in adolescents and young adults presenting with bilateral severe conductive hearing loss and intact tympanic membranes. Advanced imaging is essential for accurate diagnosis and timely surgical management, even in the absence of classic inflammatory signs.

Keywords:
adult; cholesteatoma, congenital; diagnosis; hearing loss, bilateral; magnetic resonance imaging; otorhinolaryngologic surgical procedures.

Apstrakt

Uvod. Kongenitalni holesteatom (*congenital cholesteatoma* – CC) je retko patološko stanje srednjeg uva, koje odlikuje keratinizujući skvamozni epitel iza očuvane bubne opne. Obostrana zahvaćenost je izuzetno retka i može predstavljati značajan dijagnostički izazov, posebno u odsustvu simptoma zapaljenja. **Prikaz bolesnika.** Prikazujemo slučaj 19-godišnjeg muškarca sa postepenim, progresivnim, obostranim gubitkom sluha u trajanju od 4 godine, praćenog tinitusom i otalgijom, bez otoreje i prethodnih otoloških oboljenja. Otomikroskopskim pregledom viđena je biserno bela masa iza očuvane bubne opne, obostrano. Tonalnom audiometrijom utvrđen je težak obostrani konduktivni gubitak sluha. Multislajsnom kompjuterizovanom tomografijom temporalnih kostiju otkrivene su obostrane mekotkivne mase u šupljinama srednjeg uva, uz eroziju slušnih koščica. Urađena je obostrana timpanomastoidektomija sa osikuloplastikom, pri čemu je intraoperativno potvrđen CC, koji je zahvatao mezotimpanum i epitimpanum. Postoperativnim praćenjem *non-echo*-planarnom magnetnorezonantnom sekvencom difuzionog kretanja, obavljenim 2 godine posle operacije, nisu nađeni dokazi za rezidualnu ili rekurentnu bolest. **Zaključak.** Prikazom se naglašava značaj razmatranja CC kod adolescenata i mladih odraslih osoba koje imaju obostrani, težak konduktivni gubitak sluha i očuvane bubne opne. Napredno snimanje neophodno je za tačnu dijagnozu i blagovremeno hirurško lečenje, čak i u odsustvu klasičnih znakova zapaljenja.

Ključne reči:
odrasle osobe; holesteatom, urođeni; dijagnoza; sluh, gubitak, obostrani; magnetna rezonanca, snimanje; hirurgija, otorinolaringološka, procedure.

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Introduction

Congenital cholesteatoma (CC) is defined as a complex, multilayered, and locally invasive accumulation of stratified, keratinizing squamous epithelium located within the middle ear space behind an intact tympanic membrane. Its reported incidence is approximately 26.44 cases *per* 100,000 live births [1, 2]. Despite extensive investigation, the exact etiology of CC remains controversial, and several theories have been proposed to explain its origin [1]. The most widely accepted hypothesis suggests that CC originates from embryonic epithelial rests that fail to regress during fetal development, particularly at the time of neural groove closure. Abnormal epithelial invagination, persistence of epithelial remnants within the middle ear or antrum, or disturbances in the development of the auditory apparatus may ultimately lead to the formation of keratinized epithelial masses [1, 3].

In the majority of cases, CC is unilateral, whereas bilateral involvement is rare and has been documented only in a limited number of reports [3–7]. Clinically, CC most commonly presents in preschool-aged children with progressive conductive hearing loss (HL), often accompanied by a history of recurrent or persistent acute otitis media. A clear male predominance has been reported, with a male-to-female ratio of approximately 3 : 1. Notably, CC may also be identified in patients without a history of otorrhea, middle ear infection, tympanic membrane perforation, or prior otologic surgery, which may contribute to delayed diagnosis [8].

Otoscopic examination typically reveals a characteristic pearly white mass visible through an intact tympanic membrane, most commonly located in the anterosuperior quadrant [1, 8]. These findings distinguish CC from acquired cholesteatoma, as CC is not associated with tympanic membrane retraction and does not originate from or extend into the external auditory canal. Progressive expansion of the lesion toward the ossicular chain may occur, frequently resulting in conductive HL secondary to ossicular erosion or discontinuity [1]. High-resolution temporal bone computed tomography (CT) commonly demonstrates well-pneumatized mastoid air cells with soft-tissue opacities confined to the middle ear cavity [8].

From a diagnostic standpoint, it is essential to consider and exclude other conditions with overlapping clinical features and variable disease courses, including otitis media with effusion (OME), granulomatous middle ear disease, cholesterol granuloma, and middle ear osteoma [9–13]. Surgical excision remains the treatment of choice for CC; however, complete removal can be technically demanding, particularly in cases with ossicular involvement, where preservation of the ossicular chain poses a significant surgical challenge [8]. Owing to its rarity and often subtle clinical presentation, bilateral CC represents a distinct diagnostic entity of particular clinical interest.

Case report

A 19-year-old male was referred to our clinic with a 4-year history of gradually progressive bilateral HL. The patient additionally reported bilateral tinnitus and progressively worsening otalgia. He denied any episodes of otorrhea. His past medical history was otherwise unremarkable, and he reported no history of childhood head and neck diseases, including recurrent middle ear infections.

Otomicroscopic examination revealed a whitish, pearly appearance visible through intact tympanic membranes bilaterally (Figure 1). Pure-tone audiometry (PTA) demonstrated severe bilateral conductive HL (62.5 dB in the right ear and 55 dB in the left ear). Tympanometric assessment revealed a type B tympanogram on both sides.

Non-contrast multislice CT (MSCT) of the temporal bones demonstrated bilateral soft-tissue, mass-like densities occupying the middle ear cavities, accompanied by erosive changes of the auditory ossicles. In addition, inflammatory material was noted within the mastoid antrum and mastoid air cells, more pronounced on the left side (Figure 2).

Given the severity of HL and the risk of progressive disease-related complications, surgical intervention was indicated. Bilateral tympanomastoidectomy with ossiculoplasty was performed, revealing cholesteatoma occupying the mesotympanum and epitympanum on both sides. Intraoperatively, the tympanic membranes were found to be intact.

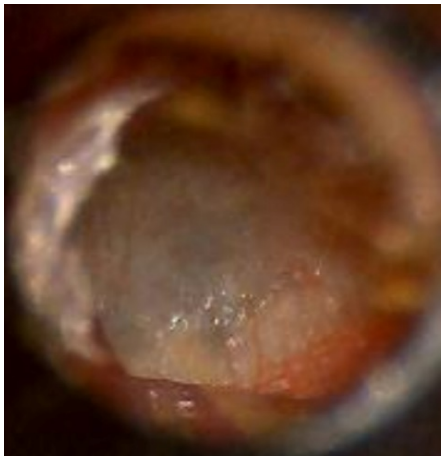

Fig. 1 – Otomicroscopy finding in the left ear pointed out a whitish glow through an intact eardrum.

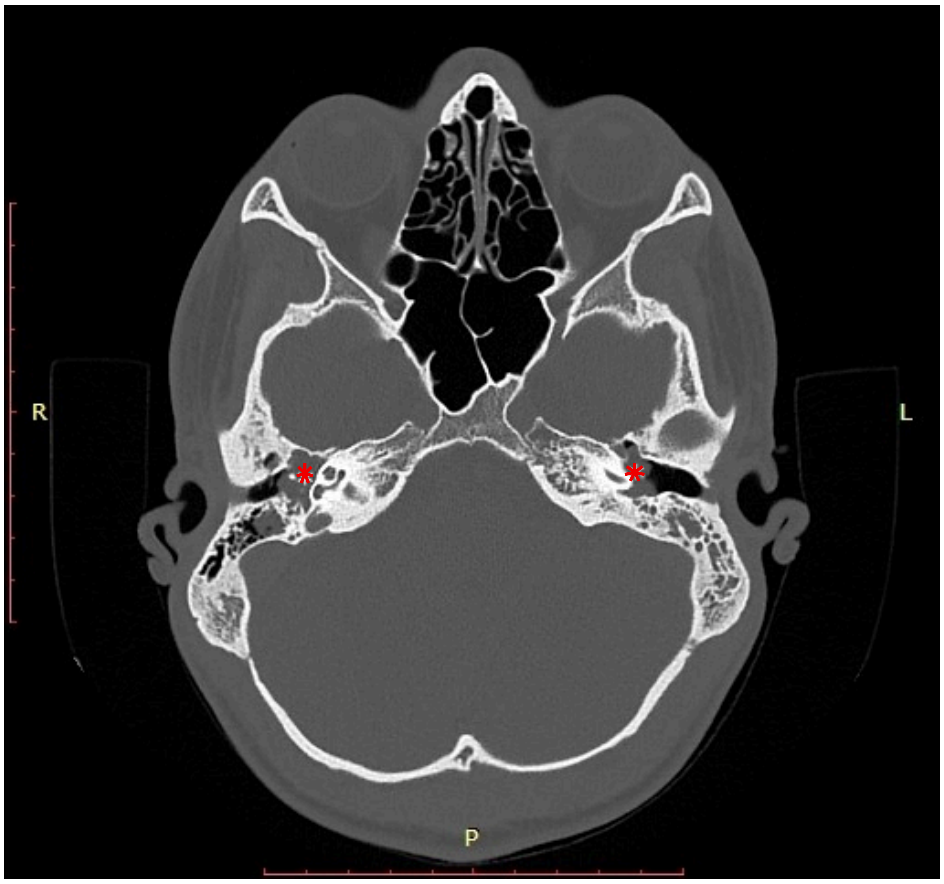

Fig. 2 – Multislice computed tomography of the temporal bones revealing a soft-tissue "mass-like" density in the middle ear cavities (red asterisk) with erosive changes on the auditory ossicles on both sides.

Due to the absence of clinical indicators of recurrence, follow-up imaging was performed 2 years postoperatively. Non-echo-planar diffusion-weighted magnetic resonance imaging (non-EPI DWI MRI) demonstrated no evidence of residual or recurrent disease.

Discussion

The present case illustrates an unusual presentation of bilateral CC in a young adult with intact tympanic membranes and no prior otologic history. The absence of recurrent otitis media, otorrhea, otologic trauma, or previous ear surgery is consistent with the diagnostic criteria for CC and likely contributed to delayed recognition of the disease. Progressive bilateral conductive HL was the dominant clinical manifestation, emphasizing the insidious nature of CC and its potential to remain undiagnosed until advanced stages.

The severity of HL observed in this patient prompted careful consideration of alternative diagnoses. While bilateral middle ear pathology with intact tympanic membranes initially suggested OME, audiometric findings demonstrated severe conductive HL, with a PTA of 62.5 dB in the right ear and 55 dB in the left ear. This is substantially greater than the degree of HL typically associated with OME. HL in OME generally ranges from 18 to 35 dB and is classified as mild conductive HL [10]. This discrepancy necessitated further diagnostic evaluation and ultimately led to exclusion of OME as a likely diagnosis.

Bilateral CC is an exceptionally rare otologic entity, with few documented cases in the literature, thereby limiting the availability of robust, high-level evidence and largely confining current understanding to case reports and small case series. Epidemiologically, bilateral CC predominantly affects the pediatric population, with a mean age at diagnosis of approximately 7.77 years and demonstrates a marked male predominance [3, 8]. Clinically, the condition most frequently presents as an incidental finding during routine otoscopic examination, although conductive HL also remains a common manifestation, underscoring the need for systematic bilateral ear assessment. In contrast to unilateral CC, bilateral cases often exhibit asymmetry, which in turn necessitates individualized and staged surgical management strategies for each ear [3]. While lesions are most commonly situated within the anterosuperior quadrant of the middle ear, more advanced presentations with mastoid extension have also been reported [14–16]. Surgical intervention remains the cornerstone of management, typically involving tympanomastoidectomy utilizing canal wall up or canal wall down techniques, with increasing integration of endoscopic approaches aimed at minimizing morbidity and improving visualization. Importantly, bilateral CC necessitates the critical importance of meticulous long-term surveillance [16]. Furthermore, the potential for sequential or delayed contrala-

teral involvement necessitates vigilant follow-up of both ears, even in cases initially presumed to be unilateral [3]. Within this context, the present case contributes to the limited existing body of evidence by further elucidating the clinical variability, diagnostic challenges, and therapeutic considerations inherent to this rare condition, reinforcing the imperative for early detection and a tailored, bilateral management approach.

Granulomatous disease of the middle ear represents another rare but important differential diagnosis in such presentations [11]. Otologic manifestations may include benign granulomatous lesions of the temporal bone, which can cause bone destruction and diagnostic uncertainty, occasionally presenting with sensorineural HL or facial nerve palsy [17, 18]. In the present case, the absence of sensorineural HL, facial nerve dysfunction, and systemic manifestations reduced the likelihood of this diagnosis, despite the presence of intact tympanic membranes, which could otherwise obscure underlying pathology.

MSCT revealed bilateral soft-tissue, mass-like opacities within the middle ear cavities, accompanied by ossicular erosion, but did not provide definitive confirmation of cholesteatoma. MSCT remains an essential imaging modality in the assessment of middle ear disease due to its excellent spatial resolution and ability to delineate bony anatomy and erosive changes. However, its capacity to characterize soft-tissue lesions is limited without contrast enhancement, often necessitating complementary imaging. In this context, magnetic resonance imaging, particularly diffusion-weighted imaging, plays a critical role in confirming the diagnosis and in postoperative surveillance [19].

Bilateral tympanomastoidectomy with ossiculoplasty was performed, revealing cholesteatoma occupying the mesotympanum and epitympanum on both sides. Intraoperatively, the tympanic membranes were found to be intact, a feature that likely masked the underlying pathology and contributed to delayed diagnosis. Postoperative follow-up is crucial in patients with cholesteatoma due to the risk of residual or recurrent disease. Current guidelines recommend diffusion-weighted MRI at intervals of 1 to 2 years, depending on clinical suspicion and patient-specific factors. In the present case, due to the absence of recurrence of clinical indicators, follow-up imaging was performed 2 years postoperatively. Non-EPI DWI MRI showed no evidence of residual or recurrent disease. This imaging modality is currently favored for postoperative surveillance owing to its high sensitivity and specificity, frequently exceeding 90% for detecting recurrent cholesteatoma [20].

Conclusion

This case highlights the need for a high index of suspicion for congenital cholesteatoma in adolescents and young adults presenting with bilateral severe conductive hearing loss, intact tympanic membranes, and absence of otorrhea. In such clinical scenarios, advanced imaging with multislice computed tomography and/or magnetic resonance imaging is essential for identifying underlying middle ear pathology and for distinguishing congenital cholesteatoma from more common conditions such as otitis media with effusion, thereby facilitating timely surgical management and preventing irreversible hearing loss and disease-related complications.

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