CASE REPORT

Congenital Cholesteatoma Presenting as Isolated Conductive Hearing Loss: A Diagnostic Dilemma

Upasana Saikia[1], Zareen[2], Manu CB[3], Jeffry[4], Suvamoy Chakraborty[5]

Received on: 23.08.2026; **Accepted on:** 27.08.2026; **Published on:** 18.09.2026

ABSTRACT

Congenital cholesteatoma is a rare pediatric condition characterized by the presence of keratinizing squamous epithelium within the middle ear behind an intact tympanic membrane, without a history of otorrhea, tympanic membrane perforation, or previous ear surgery. It is a rare disease with an incidence of 3 to 6 per 100,000 persons; congenital cholesteatoma accounts for 4% to 24% of the cholesteatomas in children and 2% to 5% of all cholesteatomas. The mean age at presentation is 5 to 7 years.[1] It is more commonly seen in children and may present with conductive hearing loss or may be detected incidentally during otoscopic examination. The exact pathogenesis remains uncertain, although persistence of embryonic epithelial remnants within the developing middle ear has been proposed as an important mechanism. We present a case of congenital cholesteatoma in a 13-year-old female with a history of decreased hearing for 2 years.Examination revealed an intact tympanic membrane, while exploratory tympanotomy revealingcholesteatoma sac in the middle ear. Modified Radical Mastoidectomy was subsequently performed. The case highlights the importance of clinical examination, radiological evaluation, and surgicalassesment in the diagnosis and management of congenital cholesteatoma. Although initially limited in extent, the lesion can progressively involve the ossicular chain and surrounding temporal bone structures if left untreated.

Keywords: Congenital cholesteatoma; pediatric cholesteatoma; conductive hearing loss; temporal bone.

CASE PRESENTATION

A 13-year-old female presented with decreased hearing in the left ear for 2 years. The hearing impairment was insidious in onset and gradually progressive in nature, with no associated history of ear discharge or any ear pain.Otoscopic examination revealed intact tympanic membranes bilaterally. Pure tone audiometry showed normal hearing in the right ear (10 dB) and moderate conductive hearing loss in the left ear (67 dB), with gross Eustachian tube dysfunction. High-resolution computed tomography (HRCT) of the left temporal bone revealed soft tissue density involving the epitympanum and mesotympanum, extending into the aditus and antrum, with erosion of the incus and stapes, blunting of the scutum, and sclerosis of the left mastoid air cells. The patient was started on intravenous antibiotics preoperatively and was planned for left exploratory tympanotomy. Intraoperatively, cholesteatoma was identified within the mastoid antrum extending up to the sinodural angle, with ossification of the aditus completely. The malleus and stapes footplate were present, while the incus was eroded and the stapes suprastructure was absent. Subsequently, left type IV tympanoplasty with canal wall-down mastoidectomy and conchomeatoplasty was performed. The patient was discharged on oral antibiotics following surgery. Follow up of the patient at 2 months is unremarkable.

Otoendoscopic image showing an intact left tympanic membrane with well defined anatomical landmarks and normal cone of light. There is no evidence of any perforation, discharge, granulation tissue or retraction.

Junior Resident[1], Associate Professor[2,3], Senior Resident[4], Senior Professor & Head[5]

Department of ENT North Eastern Indira Gandhi Regional Institute of Health & Medical Sciences (NEIGRIHMS), Shillong, India

Corresponding Author: Upasana Saikia

Junior Resident,
Department of ENT NEIGRIHMS, Shillong, India.

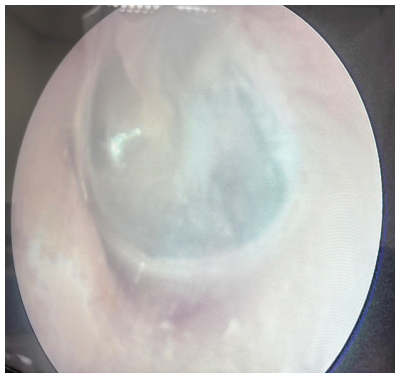
Email ID: upasanasaikia1996@gmail.com

Source of funding: None

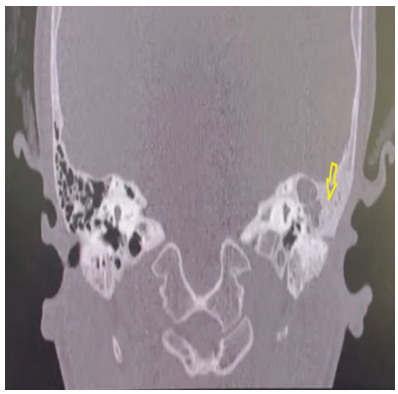
Conflict of interest: None declared

Access this article online	
Website: **https:**//ajohns.net/	Quick Response Code
DOI: 10.66243/Krishers-AJOHNS-037	

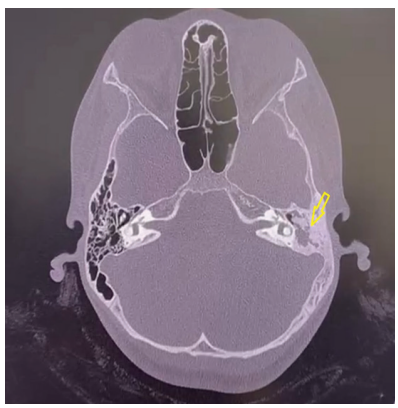
How to cite this article: Saikia U, Zareen, CB Manu, Jeffry, Chakraborty S. Congenital Cholesteatoma Presenting as Isolated Conductive Hearing Loss: A Diagnostic Dilemma. J Otolaryngol Head Neck Surg. 2026; 2(2):18-20



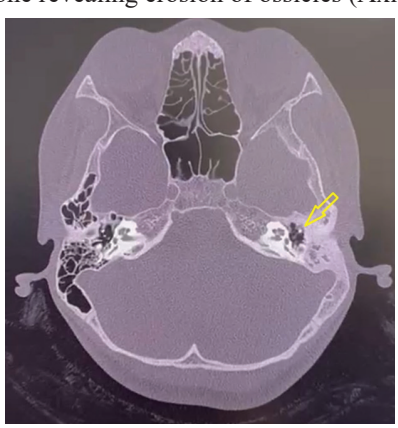
High Resolution Computed Tomography (HRCT) Of left Temporal bone revealing cortical defect (Coronal cut)



High Resolution Computed Tomography (HRCT) Of left Temporal bone revealing cortical defect (Axial cut)



High Resolution Computed Tomography (HRCT) Of left Temporal bone revealing erosion of ossicles (Axial cut)



DISCUSSION

Congenital cholesteatoma (CC) is classically defined as a white, keratinizing mass located behind an intact tympanic membrane in the absence of a previous history of otitis media or otologic surgery.[2] Although relatively uncommon, accounting for approximately 2%–5% of all cholesteatomas, its true incidence may be underestimated because of its often insidious and clinically occult presentation. Conductive hearing loss is the most common presenting symptom.[1] Congenital cholesteatomas may originate from various regions of the temporal bone, with the middle ear being the most frequent site, while lesions may also arise from the petrous apex, external auditory canal, mastoid, and other temporal bone structures. Among these, congenital cholesteatoma isolated to the mastoid is exceptionally rare and may remain undetected for a prolonged period owing to its concealed location and nonspecific clinical manifestations.[3] The term "primary mastoid cholesteatoma" refers to a congenital-type lesion occurring in the mastoid in the presence of an intact tympanic membrane and without a history of otorrhea, infection, or previous otologic surgery, irrespective of the extent of intracranial extension.[3] We presentd an unusual case of a 13-year-old female with a 2-year history of left-sided ear discharge and progressive hearing loss, in whom the tympanic membrane remained intact on clinical examination and the cholesteatoma was identified only during surgery. There was no pearly white mass typically described in cases of congenital cholesteatoma. This case highlights the diagnostic challenges associated with occult congenital cholesteatoma and emphasizes the importance of maintaining a high index of suspicion despite an apparently intact tympanic membrane. This will also help us in adequate patient counselling preoperatively.

CONCLUSION

Congenital cholesteatoma in children is an uncommon but potentially destructive condition that may progress silently despite an apparently intact tympanic membrane. Early clinical recognition, appropriate imaging, and timely surgical intervention are essential to limit disease progression and preserve hearing. A high index of suspicion, particularly in cases with an intact tympanic membrane and disproportionate hearing loss, can facilitate diagnosis of occult lesions. Multidisciplinary collaboration among otologists, radiologists, and pediatric care teams further contributes to accurate diagnosis, appropriate surgical planning, and prevention of long-term complications.

DISCLOSURE

This case was managed in the Department of ENT, NEIGRIHMS, Shillong, and is presented to emphasize the uncommon yet clinically significant entity of congenital cholesteatoma in children. No external funding was obtained, and there are no conflicts of interest to declare.

REFERENCES

1. Liao PS, Lan WC, Wang CY, Lin CD, Aoh Y. Adult-onset congenital cholesteatoma in the hypotympanum initially presenting as Bell's palsy: A case report. Medicine (Baltimore). 2024 Mar 22;103(12):e37511. doi:10.1097/MD.0000000000037511
2. Xie L, Zeng L. Congenital Middle Ear Cholesteatoma: A Report of 3 Cases and a Literature Review. Ear Nose Throat J. 2026 Feb;105(2). doi:10.1177/01455613241283799
3. Misale P, Lepcha A. Congenital Cholesteatoma in Adults-Interesting Presentations and Management. Indian J Otolaryngol Head Neck Surg. 2018 Dec;70(4):578–82. doi:10.1007/s12070-018-1362-z
4. Gamoneda DE, Patel RA, Yanez-Siller JC, Rivera AL. Presenting a Case of A Congenital Cholesteatoma of the Mastoid in Adult.
5. Swain SK. Congenital cholesteatoma: a review. Int J Adv Med. 2022 Sep 23;9(10):1072. doi:10.18203/2349-3933.ijam20222409
6. Kalmanson O, Francom C, Darr O, Hamilton S. Pediatric cholesteatoma associated with congenital aural atresia and stenosis. Am J Otolaryngol. 2023 Nov;44(6):103986. doi:10.1016/j.amjoto.2023.103986
7. Hasan B, Hamdan Z, Al Baker R. Early diagnosis and complete excision of congenital cholesteatoma in a 4-year-old boy: A case report. Int J Surg Case Rep. 2025 Sep;134(C). doi:10.1016/j.ijscr.2025.111820