International Journal of Advances in Medicine
Swain SK. Int J Adv Med. 2022 Oct;9(10):1072-1077
http://www.ijmedicine.com
pISSN 2349-3925 | eISSN 2349-3933
DOI: https://dx.doi.org/10.18203/2349-3933.ijam20222409

Review Article

Congenital cholesteatoma: a review

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Received: 25 July 2022
Accepted: 30 August 2022

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ABSTRACT

Congenital cholesteatoma (CC) is a whitish mass lesion found in the middle ear cleft behind the intact tympanic membrane in early life. It is a relatively uncommon disease and shows its growth and extension over a period of time. CC is evidenced with no prior history of tympanic membrane perforation, otorrhea, previous otologic procedures, normal pars tensa, and pars flaccida. The etiopathogenesis of CC is still controversial; however, the epithelial cell rest theory is the most commonly accepted one. The most common sites of CC are the anterior-superior and posterior-superior quadrants of the tympanic cavity. The most common clinical presentation of CC is conductive hearing loss. Early detection and intervention are needed to avoid the complications of CC. The treatment of choice in CC is still surgical. The frequency of recurrence seems to be lower in patients with CC than in acquired cholesteatoma. Rapid progression of the CC may occur in older children and the recurrence has been associated with advanced progression of the cholesteatoma. There are very few reports for CC available in the medical literature This objective of this review article is to discuss the prevalence, etiopathology, clinical manifestations, diagnosis, and treatment.

Keywords: CC, Middle ear cleft, Conductive hearing loss, Bony erosion

INTRODUCTION

The cholesteatoma is a non-neoplastic lesion characterized by a three-dimensional sac-like structure lined with keratinized squamous epithelium placed over fibrous stroma and contains keratin debris in the center and seen in temporal bone with its characteristic bone eroding properties.[1] It can be classified as congenital or acquired. CC refers to an epidermal cyst originating from congenital remnants of keratinizing squamous epithelium in the temporal bone. The term congenital has been used rather conventionally as the etiopathogenesis of the CC remains unclear and the hypotheses range from origin during the fetal period to an acquired condition in the infant.[2] The strict criteria for CC are the presence of a white pearl behind the intact tympanic membrane with no history of perforation, otorrhea, or otologic surgery.CC may be associated with the history of otitis media with effusion, acute otitis media, brief otorrhea, or myringotomy.[3] Cholesteatoma is a destructive lesion of the temporal bone that gradually expands and results in complications by eroding the adjacent bony structures.[4] The bony destruction CC can cause ossicular chain and otic capsule damage and leads to hearing impairment, vestibular dysfunction, facial nerve paralysis, and intracranial complications. Surgery is the only treatment of choice in the case of CC.

METHODS FOR LITERATURE SEARCH

Research articles regarding CC were searched via multiple approaches. We started by searching the Scopus, Pub Med, Medline, and Google Scholar databases online. A search strategy using PRISMA (Preferred reporting items for systematic reviews and meta-analysis) guidelines was developed. This search strategy recognized the abstracts of published articles, while other research articles were discovered manually from the citations. Randomized controlled studies, observational studies, comparative studies, case series, and case reports were evaluated for

eligibility. There were a total number of articles 64 (18 case reports; 16 cases series; 30 original articles) (Figure 1). This article reviews the epidemiology, etiopathology, clinical profile, investigations, and current treatment options of CC. This review article presents a baseline from which further prospective trials can be designed and which may help as a spur for further research in this rare clinical condition for which very few studies are performed.

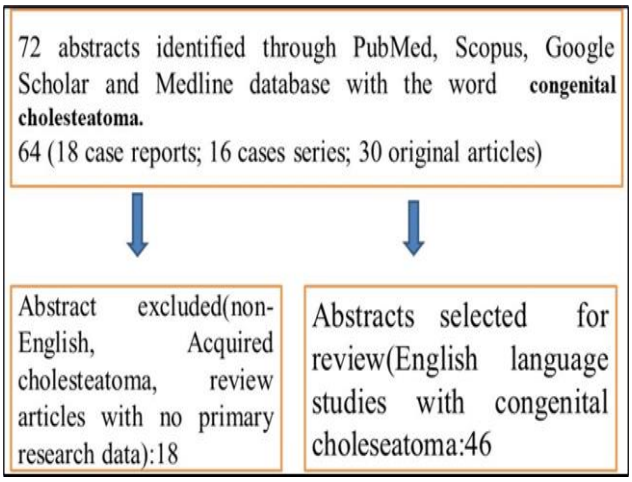

Figure 1: Method of literature search.

EPIDEMIOLOGY

Cholesteatoma is a term first used by Muller in 1838.[5] The first documented case of cholesteatoma-like mass was reported by Du Verneey in 1683.[5] He described a whitish mass between the cerebrum and cerebellum, the first published description of CC in 1885 by Luchae.[6] In 1953, CC of the middle ear was first described by House.[7] CC is an uncommon entity, accounting for 4% of pediatric cholesteatoma.[8] CC has historically been considered a rare disorder. However, a review of the literature shows an incidence of CC ranges from 4 to 24%, and these values are often underestimated.[5] Derlacki and Clemis first described the CC as pearly white mass medial to an intact tympanic membrane in 1965 with a modification done in 1986 by Levenson et al.[9] CC is more commonly affecting the boys (64%).[10] Levenson proposed that prior incidence of otitis media are not grounds for exclusion of the CC.[11] The collection of a large group of patients with CC is difficult to get in a short period. Most series of CC include 50/ so patients or when concerning more than 100 patients, data collected over 20 years or from multiple centers.[8]

ETIOPATHOGENESIS

The origin of the CC is a subject of controversy but appears to be independent of eustachian tube dysfunction, as in acquired cholesteatoma.[12] There are several hypotheses have been presented for the etiopathogenesis of CC. The etiopathogenesis of CC is often difficult to determine as the incidence of CC is a low and direct assessment of whole middle ear cleft in young children and asymptomatic infants are not possible. There are sometimes questions raised about whether this lesion is truly congenital or acquired. It was thought that ectodermal tissue from the external auditory canal may migrate into the middle ear cleft due to failure of the inhibitory function of the tympanic ring.[13] The epidermoid formation hypothesis is based on the findings of the congenital cell rest of the squamous epithelium in the middle ear of the fetus and has been accepted as the most accepted pathogenesis hypothesis.[14] However, the invasion of the squamous epithelium has been seen after-acquired inflammatory injury, and cholesteatoma in early childhood time has been described as an acquired inclusion of keratinized squamous epithelium.[15] Bone erosion is an important feature of acquired cholesteatoma and appears due to the release of osteolytic enzymes from the associated inflammatory cells and granulation tissues rather than squamous epithelium. However, inflammation and recurrent infections are not major features of CC, bone erosion is expected to occur relatively late in the course of the disease and the symptoms are minimal. Based on the operative findings, the cholesteatoma is classified morphologically into open type and closed type. In the open type, the cholesteatoma does not form an epithelial pearl or cyst, but developed as flat keratinizing epithelium whereas the closed type presents with a cystic form. CC is often found in the anterosuperior part of the middle ear in western countries.[16] It is commonly seen at a posterosuperior location in Japan.[17] This difference may be partly because the majority of cholesteatoma is found in the posterosuperior portion of the middle ear cavity in Japanese patients. In western countries, the children often present with a white mass visible through the anterosuperior quadrant of the tympanic membrane, and the majority of the cholesteatoma found in Japan is already at the advanced stage. However, the study in western countries shows that cholesteatoma is also common in the posterosuperior portion of the middle ear cavity.[18] So there is no consensus has yet been finalized about the origin of CC. There is minimal inflammation and well-developed mastoid air cells in CC unlike the findings in the case of acquired cholesteatoma. Most CC was documented to originate in the anterosuperior quadrant and grow into the posterosuperior quadrant, eroding the ossicles and finally invading the mastoid bone.[19] The pathogenesis may represent the natural history of CC as shown by serial illustrations of disease extent in the study done on 34 patients.[20] However, in that study, the authors did not report the ages of the patients at operation and did not completely exclude the possibility of extension in opposite direction i.e., from posterosuperior to anterosuperior as 29 out of 34 patients had extensive lesions in several middle ear cleft sites. Another study reported that anterosuperior quadrant of the middle ear cleft is the most likely origin of the CC.[21] There is also evidence that the CC originated from posterior quadrants of middle ear cleft.[22]

CLINICAL PRESENTATIONS

CC is commonly found in children the age of 3 to 5 years, but the diagnosis of this lesion is seen in the age of 1 to 2

years due to progress in technology such as imaging and otoendoscopy.[23] CC is more common in boys than girls by a ratio of 3:1.[23] The classical presentations of CC include normal pars tensa and pars flaccida with the white mass behind the intact tympanic membrane.[24] There is no previous history of otorrhea, surgical procedures, or perforation of the tympanic membrane. The clinical presentations are often incidental. Patients with CC can present with hearing impairment, blocking sensation in the ear.[25] Most patients with CC complain of hearing loss, some are unaware of the condition till they are informed about abnormal tympanic membrane or hearing deficit.[26] There may be scanty discharge but it is uncommon due to the sac in the middle ear bulging out from the medial to lateral and eroding/rupturing the tympanic membrane. Patients may present with facial nerve palsy of different grades.[27,28] Patients of CC may present with profound hearing loss with vestibular symptoms such as vertigo. Vertigo may occur due to erosion of the labyrinthine bone. Derlacki's criteria for CC include definite CC where a white pearl behind an intact tympanic membrane or probable CC where the patient presents with a voluminous retrotympanic mass with the bulged tympanic membrane.[29] CC involving petrous bone often presents with facial nerve paralysis, profound sensorineural hearing loss, and vestibular symptoms.

DIAGNOSIS

The diagnosis of CC is made on clinical findings and radiological findings. Otoscopy or otoendoscopy is useful to examine the tympanic membrane and shows a whitish mass behind the intact tympanic membrane in case of CC (Figure 2).

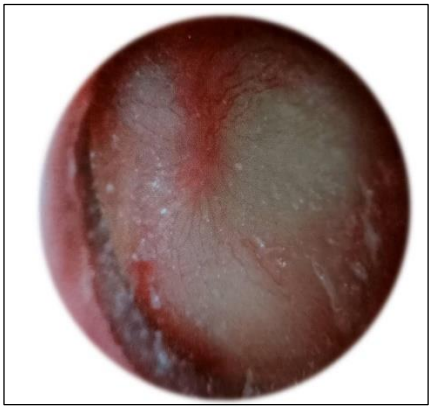

Figure 2: Otoendoscopic of white mass behind intact tympanic membrane.

In children with persistent asymmetric or unilateral conductive hearing loss, particularly speech reception threshold (SRT) over 40 decibels, CC should be suspected.[8] The mean age of diagnosis is 4 to 5 years.[8] In doubtful cases, myringotomy can be done to confirm the diagnosis of CC. High-resolution computed tomography (HRCT) of the temporal bone can show the extent of the involvement, ossicular status, and surrounding bone involvement by CC. Magnetic resonance imaging (MRI) findings in the case of CC are helpful to differentiate it from other pathologies of the temporal bone. In MRI of CC, T1: Hypo/Iso intense; T2: Hyperintense; T1 with contrast: nonenhancement; non-echoplanar DW1: Diffusion restriction present which appears as a bright signal. The surgical findings in CC are normal pars tensa and pars flaccida, well-pneumatized mastoid, lateralized chorda tympani nerve, and lateralized handle of malleus. In CC, middle ear mucosa is usually normal and the tympanic plexus is usually clearly seen over the promontory and the first ossicle to get eroded is stapes in most cases of CC. The CC is classified into different types based on the extent of the lesion in imaging and further confirmed in intra-operative findings. There are five types of the CC of the temporal bone.[30] Type 1 CC: Small intact cholesteatoma sac is present in the middle ear and not involving the ossicles and mastoid; type 2:Choeseatoma is present in the whole middle ear but without affecting mastoid and ossicles, type 2a-Without facial nerve paralysis, and type 2b-With facial nerve paralysis; type 3: Choesetaoma sac of CC extend into the mastoid (3a-without facial nerve paralysis and 3b-with facial nerve paralysis); type 4:CC arising from the supralabyrinthine region and extends into the middle ear and internal auditory meatus (4a-without facial nerve paralysis; 4b-with facial nerve paralysis); type 5: CC arising from the petrous apex with profound sensorineural hearing loss (5a-without facial nerve paralysis and 5b-with facial nerve paralysis). In case of CC arising from petrous bone, HRCT temporal bone shows soft tissue involving petrous bone along with the involvement of internal auditory meatus, cochlea, and all semicircular canals. Non-echoplanar DW1 MRI shows restriction to diffusion confirming the diagnosis of CC in petrous bone.

TREATMENT

Patients with CC need early surgical intervention, otherwise, the late stage of this disease results in a high recurrence rate. If the CC is removed surgically, the lining mucosa will rapidly regenerate afterward, unlike the case following removal of acquired cholesteatoma.[31] So, the external auditory canal can be preserved by many surgeons for CC due to the rare chance of recurrence unless there is major damage to the mucosa like extensive removal because of severe changes in the middle ear cavity.[32] Canal wall down tympanoplasty is often performed for CC. However, canal wall-up tympanoplasty is performed currently for CC.

The staged surgery is preferred when the cholesteatoma infiltrates the air cells or spreads into the anterior part of the middle ear cavity or the mastoid antrum.[33] Canal wall down tympanoplasty is sometimes done along with mastoid obliteration as a one or two-stage procedure. When the staged surgery is planned, the duration of six months to one year after the first surgery is taken before performing the second look surgery. The frequency of

recurrence is usually lower for CC than the patients with acquired cholesteatoma.[34]

One study showed the recurrence rate after canal wall-up tympanoplasty was approximately 3.9% and this recurrence was due to the exposed bone because of unable to preserve the mucosa from the tympanic isthmus to the mastoid antrum.[35] The residual CC after surgery develops in the anterosuperior part of the middle ear cavity or around the ossicles, particularly around the stapes where surgical access is likely to be minimal. The recurrence from the residual cholesteatoma is likely in some cases of open type cholesteatoma due to the border between the epithelium of the cholesteatoma and the normal mucosa is not clear, so the staged tympanoplasty or second look surgery is required in such patients.[36]

Residual cholesteatoma is seen at the sites where removal is difficult during the initial surgery, such as oval window, round window, and sinus tympani, or where the cholesteatoma infiltrated the small cells like tympanic recess and the mastoid antrum. The endoscopic approach is helpful for complete removal of the cholesteatoma in doubtful cases and even after second-look surgery as the cholesteatoma is seen at the pyramidal apex or extended into the cochlea from behind the apex or extended into the cochlea behind the mastoid segment of the facial nerve. As such situations may be encountered; caution must be done to be exercised in treatment of CC.

The frequency of recurrence is lower in patients with CC than in acquired cholesteatoma. One study on the recurrence rate after canal wall-up tympanoplasty was approximately 3.9%. In these cases of recurrence, bone was exposed as mucosa could not be preserved from the tympanic isthmus to the mastoid antrum.[35] One study reported that a residual cholesteatoma was seen postoperatively in approximately 50% of the patients with CC.[37] In that study, to explain the residual cholesteatoma, the authors suggested that CC commonly develops in the anterosuperior portion of the middle ear or around the ossicles, especially around the stapes where surgical accessibility is likely to be limited.[37]

The chance of recurrence from residual cholesteatoma is likely in a few cases of open type cholesteatoma because the border between the epithelium of the cholesteatoma and the normal mucosa is not clear, so staged tympanoplasty or second look surgery is required in such cases.[38]

The residual cholesteatoma is often seen at the sites where the removal of cholesteatoma is difficult during the initial surgery, such as the round window, oval window, and the sinus tympani, or where the cholesteatoma has infiltrated the areas such as epitympanic recess and the mastoid antrum. Endoscopy is very useful for performing surgery to remove the cholesteatoma completely.CC is completely removed from the middle ear cleft by considering the stages of the disease (Table 1).[39,40]

Table 1: Description of congenital cholesteatoma in different stages.

Stages	Description
Stage I	One quadrant involved. No ossicles affected or no mastoid extension
Stage II	Multiple quadrants involved. No ossicles affected or no mastoid extension
Stage III	Ossicles involved. Erosions of ossicles and no mastoid extension
Stage IV	Mastoid bone involved regardless findings elsewhere

CONCLUSION

CC is a rare disorder and seems to be on the rise. Heightened awareness about CC with its early diagnosis is imperative. Early treatment reduces the extent of the disease and decrease the risk of recidivism and complications of CC. Suspicions should be raised if a whitish mass is found medial to the intact tympanic membrane. The treatment of CC is surgical. Preoperative CT scan is helpful to know the extent of the disease and predict postoperative issues. The classification of CC is important for consistent findings and comparison.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Bennett M, Warren F, Jackson G, Kaylie D. Congenital cholesteatoma: theories, facts, and 53 patients. Otolaryngol Clin N Am. 2006;39:1081-94.
2. Persaud R, Hajioff D, Trinidade A, Khemani S, Bhattacharyya MN, Papadimitriou N et al. Evidence-based review of aetiopathogenic theories of congenital and acquired cholesteatoma. J Laryngol Otol. 2007;121(11):1013-9.
3. Swain SK, Agrawala R. Mastoid surgery: a high-risk aerosol generating surgical procedure in COVID-19 pandemic. Int J Otorhinolaryngol Head Neck Surg. 2020;6(10):1941-6.
4. Swain SK, Behera IC, Sahu MC. Role of Betadine irrigation in chronic suppurative otitis media: Our experiences in a tertiary care teaching hospital of East India. Int J Heal All Sci. 2019;8(1):29-32.
5. Kazahaya K, Potsic WP. Congenital cholesteatoma. Curr Opinion Otolaryngol Head Neck Surg. 2004;12(5):398-403.
6. Karmody CS, Byahatti SV, Blevins N, Valtonen H, Northrop C. The origin of congenital cholesteatoma. Am J Otol. 1998;19(3):292-7.
7. House HP. Management of congenital ear canal atresia. Laryngoscope. 1953;63(10):916-46.
8. Potsic WP, Korman SB, Samadi DS, Wetmore RF. Congenital cholesteatoma: 20 years' experience at The Children's Hospital of Philadelphia. Otolaryngol Head Neck Surg. 2002;126:409-14.

9. Levenson MJ, Parisier SC, Chute P, Wenig S, Juarbe C. A review of twenty congenital cholesteatomas of the middle ear in children. Otolaryngol Head Neck Surg. 1986 ;94(5):560-7.
10. Benhammou A, Nguyen DQ, El Makhloufi K, Charachon R, Reyt E, Schmerber S. Long term results of congenital middle ear cholesteatoma in children. Ann Otolaryngol Chir Cervicofac. 2005;122(3):113-9.
11. Levenson MJ, Michaels L, Parisier SC. Congenital cholesteatomas of the middle ear in children: origin and management. Otolaryngol Clin N Am. 1989;22(5):941-54.
12. Dornhoffer J. Cartilage tympanoplasty: indications, techniques, and outcomes in a 1,000-patient series. Laryngoscope. 2003;113:1844-56.
13. Aimi K. Role of the tympanic ring in the pathogenesis of congenital cholesteatoma. Laryngoscope. 1983;93:1140-6.
14. Persaud R, Hajioff D, Trinidade A. Evidence-based review of aetiopathogenic theories of congenital and acquired cholesteatoma. J Laryngol Otol. 2007;121:1013-9.
15. Tos M. A new pathogenesis of mesotympanic (congenital) cholesteatoma. Laryngoscope. 2000;110:1890-7.
16. Swain SK, Nahak B, Mohanty JN. Pediatric tympanoplasty: Our experiences in a tertiary care teaching hospital of Eastern India. Med J Dr. DY Patil Vidyapeeth. 2020;13(3):229-34.
17. Kojima H, Miyazaki H, Tanaka Y, Shiwa M, Honda Y, Moriyama H. Congenital Middle Ear Cholesteatoma Experience in 48 Cases. Nippon Jibiinkoka Gakkai Kaiho. 2003;106(9):856-65.
18. Huang TS, Lee FP. Congenital cholesteatoma: review of twelve cases. Am J Otol. 1994;15:276-81.
19. Nelson M, Roger G, Koltai PJ, Garabedian EN, Triglia JM, Roman S et al. Congenital cholesteatoma: classification, management, and outcome. Arch Otolaryngol Head Neck Surg. 2002;128(7):810-4.
20. Koltai PJ, Nelson M, Castellon RJ, Garabedian EN, Triglia JM, Roman S et al. The natural history of congenital cholesteatoma. Arch Otolaryngol Head Neck Surg. 2002;128(7):804-9.
21. El-Bitar MA, Choi SS, Emamian SA, Vezina LG. Congenital middle ear cholesteatoma: need for early recognition-role of computed tomography scan. Int J Pediatr Otorhinolaryngol. 2003;67(3):231-5.
22. Inokuchi G. Congenital cholesteatoma: posterior lesions and the staging system. Annal Otol Rhinol Laryngol. 2010;119:5.
23. Swain SK, Janardan S, Mohanty JN. Endoscopy guided eustachian tube balloon dilation: Our experiences. Iran J Otorhinolaryngol. 2020;32(112):287-94.
24. Swain SK, Behera IC, Sahu MC. Tinnitus among children–Our experiences in a tertiary care teaching hospital of eastern India. Pediatria Polska. 2017;92(5):513-7.
25. Gilberto N, Custódio S, Colaço T, Santos R, Sousa P, Escada P. Middle ear congenital cholesteatoma: systematic review, meta-analysis and insights on its pathogenesis. Eur Arch Oto-Rhino-Laryngol. 2020;277(4):987-98.
26. Rohlfing ML, Sukys JM, Poe D, Grundfast KM. Bilateral congenital cholesteatoma: a case report and review of the literature. Int J Pediatr Otorhinolaryngol. 2018;107:25-30.
27. Swain SK, Das A, Mohanty JN. Acute otitis media with facial nerve palsy: Our experiences at a tertiary care teaching hospital of eastern India. J Acute Dis. 2019;8(5):204-7.
28. Swain SK, Das A, Munjal S. A rare cause of bilateral facial nerve paralysis due to acute otitis media in a 52-year-old man. Med J Dr. DY Patil Vidyapeeth. 2020;13(6):688-91.
29. Lazard DS, Roger G, Denoyelle F, Chauvin P, Garabédian EN. Congenital cholesteatoma: risk factors for residual disease and retraction pockets-a report on 117 cases. Laryngoscope. 2007;117(4):634-7.
30. Denoyelle F, Simon F, Chang KW, Chan KH, Cheng AG, Cheng AT et al. International Pediatric Otolaryngology Group (IPOG) consensus recommendations: congenital cholesteatoma. Otol Neurotol. 2020;41(3):345-51.
31. Swain SK, Behera IC, Sahu MC. Role of Betadine irrigation in chronic suppurative otitis media: Our experiences in a tertiary care teaching hospital of East India. Int J Health Allied Sci. 2019;8(1):29-32.
32. Darrouzet V, Duclos JY, Portmann D, Bebear JP. Congenital middle ear cholesteatomas in children: our experience in 34 cases. Otolaryngol Head Neck Surg. 2002;126(1):34-40.
33. Sahu MC, Swain SK. Surveillance of antibiotic sensitivity pattern in chronic suppurative otitis media of an Indian teaching hospital. World J Otorhinolaryngol-Head Neck Surg. 2019;5(02):88-94.
34. Sahu MC, Swain SK, Kar SK. Genetically diversity of Pseudomonas aeruginosa isolated from chronic suppurative otitis media with respect to their antibiotic sensitivity pattern. Ind J Otolaryngol Head Neck Surg. 2019;71(2):1300-8.
35. Kojima H, Tanaka Y, Shiwa M, Sakurai Y, Moriyama H. Congenital cholesteatoma clinical features and surgical results. Am J Otolaryngol. 2006;27(5):299-305.
36. Mcgill TJ, Merchant S, Healy GB, Friedman EM. Congenital cholesteatoma of the middle ear in children: a clinical and histopathological report. Laryngoscope. 1991;101(6):606-13.
37. Aimi K. Rule of the tympanic ring in the pathogenesis of congenital cholesteatoma. Laryngoscope. 1983;93:1140- 6.
38. Mcgill TJ, Merchant S, Healy GB, Friedman EM. Congenital cholesteatoma of the middle ear in children: a clinical and histopathological report. Laryngoscope. 1991;101(6):606-13.

39. Anthwal N, Thompson H. The development of the mammalian outer and middle ear. J Anat. 2016;228(2):217-32.
40. Swain SK, Samal R, Pani SK. Effect of smoking on outcome of tympanoplasty. Indian J Otol. 2011;17(3):120-2.

Cite this article as: Swain SK. Congenital cholesteatoma-a review. Int J Adv Med 2022;9:1072-7.