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Congenital Mastoid Cholesteatoma: Case Series, Definition, Surgical Key Points, and Literature Review

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Objectives: We evaluate 3 new cases of congenital cholesteatoma confined to the mastoid process, and compare them with cases presented in the literature in order to better define this rare lesion.

Methods: We performed a retrospective chart analysis of all congenital cholesteatomas treated surgically in a tertiary referral and skull base center. We performed a complete analysis (history, radiologic, and surgical) of all patients with congenital cholesteatoma confined to the mastoid process; we then performed a literature review and compared our findings with the presented cases.

Results: The results of preoperative imaging were in line with the surgical findings. The most important surgical issue in this type of lesion was the management of the sigmoid sinus and the jugular bulb. Half of the cases previously reported in the literature appeared not to fulfill the definition criteria of a congenital cholesteatoma of the mastoid process.

Conclusions: Congenital cholesteatoma confined to the mastoid process is a rare lesion, and is even more exceptional upon critical review of the literature. Symptoms are often lacking or nonspecific, and although cases have a congenital origin, the diagnosis often is not made until adulthood. A combined congenital cholesteatoma group with middle ear and mastoid features seems to fill in the gap in the definition. Management of the sigmoid sinus and the jugular bulb is the most demanding surgical key point.

Key Words: congenital cholesteatoma, definition, ear surgery, mastoid disease, review.

INTRODUCTION

Congenital cholesteatoma (CC) is a rare lesion that may originate in various sites within the temporal bone, including the middle ear, the external auditory canal, the mastoid process, and the petrous apex.¹ Congenital cholesteatoma of the middle ear is the most frequent type and, because of the early occurrence of conductive hearing loss, is usually diagnosed in children.^{2,3} Congenital cholesteatoma of the external auditory canal is normally a result of congenital aural stenosis,⁴ whereas the existence of a primary form is uncertain.⁵ Cholesteatoma of the petrous bone occurs frequently as a consequence of medial extension of middle ear cholesteatoma. In a minority of cases, however, it can originate from congenital remnants trapped in the petrous bone, usually in the petrous apex or the supralabyrinthine area.^{6,7} Of all the locations, the occurrence of CC in the mastoid process is the least reported.⁸ Several cases of CC of the mastoid process have been previously reported in the English-language literature, but it seems that many cases were not strictly confined to the mastoid process. In defining CC located

in the mastoid process, any other possible location of origin should be ruled out. We present 3 cases of CC specifically confined to the mastoid process and critically review the literature to discuss the definition of certain locations of CC.

MATERIALS AND METHODS

We conducted a retrospective database review of CCs exclusively located in the mastoid process among all cholesteatomas treated surgically at the Gruppo Otologico, Piacenza, Italy. Among the 64 cases of CC identified, 46 were located in the middle ear and 15 in the petrous bone; only 3 were confined to the mastoid process. The latter were all treated surgically by one of the authors (M.F.). A literature review was performed with the PubMed database in November 2010 with the key words “congenital,” “cholesteatoma,” and “mastoid.”

RESULTS

Patient 1. A 71-year-old woman presented with a many-year history of pain in the region of the left mastoid process and upper neck. Otoscopy showed

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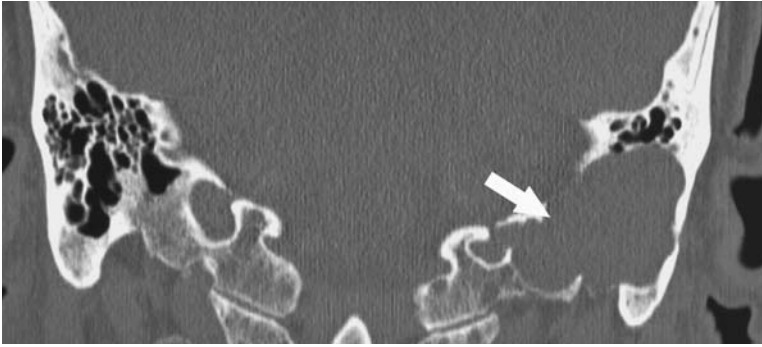


Fig 1. (Patient 1) Computed tomographic (CT) scan shows that lesion has exposed superolateral surface of jugular bulb (arrow).

a normal tympanic membrane, and audiometric examination demonstrated bilateral symmetric sensorineural hearing loss at the high frequencies.

A high-resolution computed tomographic (CT) scan of the temporal bone revealed an expansile destructive lesion in the left mastoid process with a small erosion of the external cortex. The middle ear, attic, aditus, and antrum were not involved by the lesion. The sigmoid sinus and the presigmoid posterior fossa dura were exposed, as were the lateral surface of the jugular bulb (Fig 1) and the third portion of the facial nerve. On magnetic resonance imaging (MRI), the lesion appeared hypointense on T1-weighted images and hyperintense on T2-weighted images, without enhancement after gadolinium infusion. Angiographic sequences showed no flow into the sigmoid sinus, but showed preserved flow into the jugular bulb.

A mastoidectomy with extension to the jugular bulb was planned to remove the cholesteatoma. The massive involvement of the sigmoid sinus and the jugular bulb required neck extension for control of the jugular vein and ligation, if required. During surgery, cholesteatoma was seen to involve the complete mastoid process with a small erosion of the cortical bone and exposed the third portion of the facial nerve and the lateral wall of the jugular bulb. The lesion compressed the sigmoid sinus, thinning its lateral wall, and interrupted blood flow. The presigmoid posterior fossa dura was completely exposed. Total removal was carefully performed to allow preservation of the facial nerve, the jugular bulb, and the posterior fossa dura. The absence of a clear cleavage plane between the cholesteatoma matrix and the lateral wall of the sigmoid sinus required removal of the wall itself. The sinus was plugged with Tabotamp (Johnson & Johnson, Gargrave, England) just lateral to the jugular bulb and medial to the opening of the superior petrosal sinus. Preservation of the jugular bulb made ligation of the jugular vein unnecessary. Finally, the mastoid cavity was filled with abdominal fat to protect the exposed delicate structures; the ear canal and ossicles were

left untouched.

The postoperative period was uneventful; facial nerve function remained at grade I (House-Brackmann grading system⁹), and the preoperative hearing level was preserved. The patient did not complain of any symptoms at 18 months; her facial nerve function and hearing remained unchanged. However, the patient refused radiologic follow-up.

Patient 2. A 77-year-old man presented with a 2-year history of dizziness. The results of an otoscopic examination were normal, and pure tone audiometry revealed a bilateral symmetric sensorineural hearing loss at the high frequencies. A CT scan of the temporal bone showed a destructive lesion confined to the right mastoid process without involvement of the middle ear, attic, aditus, or antrum (Fig 2). The lesion partially exposed the sigmoid sinus, the presigmoid posterior fossa dura, and the third portion of the facial nerve (Fig 3). A small area of the jugular bulb was also exposed. Magnetic resonance imaging confirmed a nonenhancing, expansile, smooth lesion that was hypointense on T1-weighted images and hyperintense on T2-weighted images, occupying the entire mastoid process and

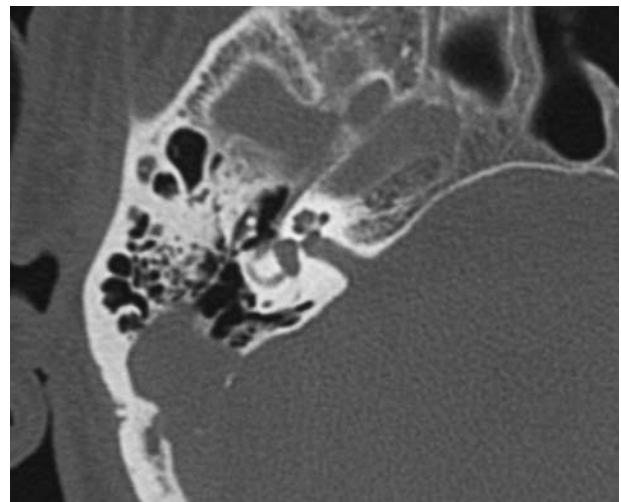


Fig 2. (Patient 2) CT scan shows lesion on right side confined to mastoid process. Attic and antrum are free of disease.

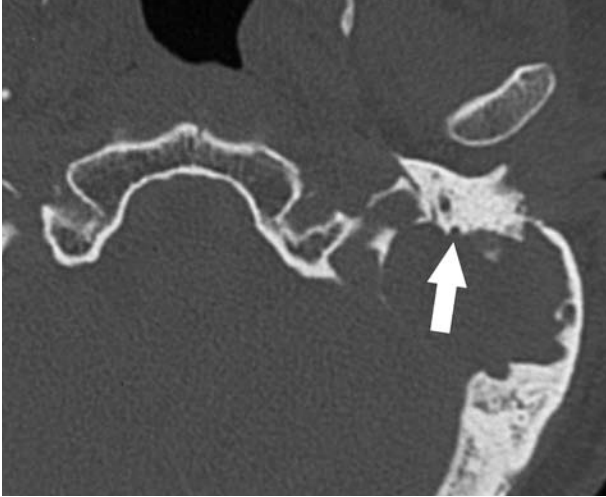


Fig 3. CT scan shows that posterior surface of mastoid portion of facial nerve (arrow) has been exposed by lesion.

making contact with the sigmoid sinus.

Right transmastoid removal of the cholesteatoma was planned. During surgery, we found that the mastoid process was filled by a huge cholesteatoma exposing the posterior cranial fossa dura and eroding the third portion of the fallopian canal. The sigmoid sinus and the jugular bulb were also exposed. Complete removal was carefully performed to allow preservation of the facial nerve, sigmoid sinus, jugular bulb, and posterior fossa dura. The mastoid cavity was filled with abdominal fat; the ear canal and ossicles were left untouched. The postoperative period was uneventful. The patient's facial nerve function and preoperative hearing remained unchanged at 1 year.

Patient 3. A 60-year-old man was referred to our clinic after incidental discovery of a large lesion in the left mastoid process during MRI performed for unrelated reasons. The results of an otoscopic examination and pure tone audiometry were normal. A high-resolution CT scan of the temporal bone revealed a lytic lesion in the left mastoid process without involvement of the middle ear, the attic, or the antrum, but with erosion of the external cortex. A large area of the sigmoid sinus, the presigmoid posterior fossa dura, and the third portion of the facial nerve were exposed (Fig 4A). Magnetic resonance imaging showed a lesion with the classic features of a cholesteatoma. Magnetic resonance angiographic sequences revealed no blood flow into the left sigmoid sinus.

A left transmastoid removal of the cholesteatoma was planned. During surgery, the cholesteatoma could be seen eroding the mastoid cortical bone and occupying the whole of the mastoid process without reaching the antrum. The cholesteatoma matrix was gently elevated from the epineurium of the third portion of the facial nerve, as well as from the dura of the posterior cranial fossa. The lateral dural wall of the sigmoid sinus was destroyed by the lesion, and the matrix was directly in contact with the vessel endothelium. It was impossible to establish a cleavage plane between the matrix and the fragile endothelium, so the sinus was sacrificed to accomplish total removal. The sinus was plugged with Tabotamp lateral to the jugular bulb and at the junction with the superior petrosal sinus. Complete removal was carefully performed to allow preservation of the jugular bulb and the posterior fossa dura. At the end of the

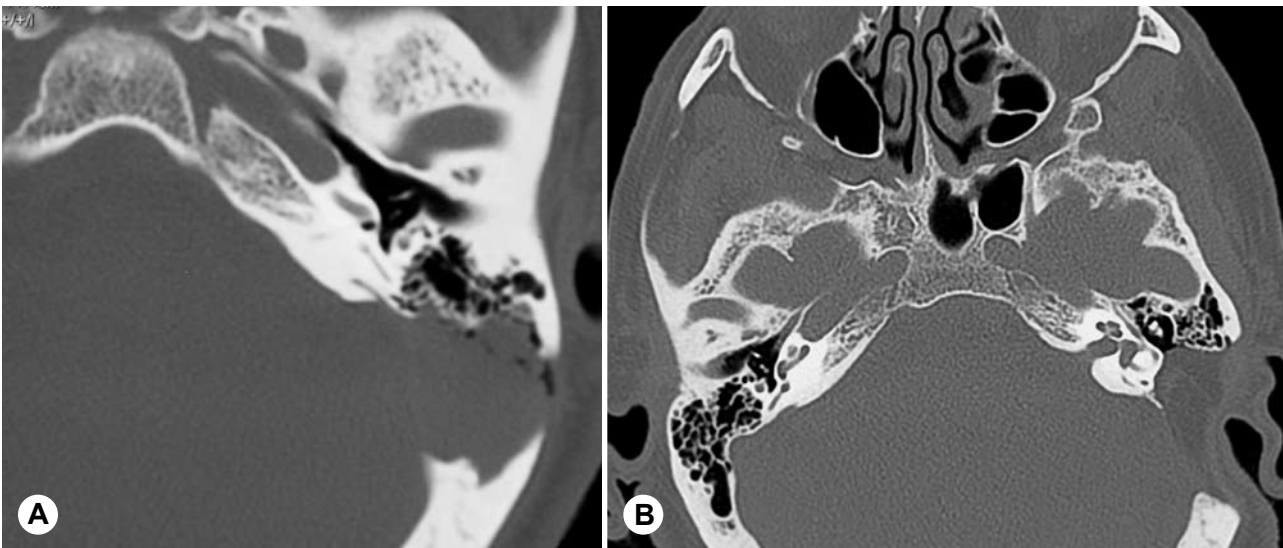


Fig 4. (Patient 3) Axial CT scans. **A)** Preoperative scan shows cholesteatoma confined to mastoid region. Posterior fossa dura, sigmoid sinus, and endolymphatic sac have been exposed by lesion. **B)** Postoperative scan shows fat obliteration of mastoid cavity.

TABLE 1. CASES OF PURE CONGENITAL MASTOID CHOLESTEATOMA

Authors	Pt No.	Pt Age (y)	Symptoms and Signs	Involvement of Structures				Surgery
				Sigmoid Sinus	Posterior Wall of EAC	Posterior Fossa Dura	Facial Nerve	
Hidaka et al ¹		65	Retroauricular swelling and pain, retroauricular abscess	Exposed	Intact	Exposed	NA	Mastoidectomy
Derlacki and Clemis ¹⁰		24	Mastoid pain and swelling	Exposed	Intact	Not exposed	NA	Atticomastoidectomy
Luntz et al ¹¹		54	Neck pain, instability	Exposed	Intact	Exposed	NA	Canal wall-up technique
Cüreoglu et al ¹²		70	Neck pain, retroauricular swelling	NA	Eroded	Exposed	NA	Modified radical mastoidectomy
Mevio et al ¹³		36	Dizziness	Occluded	Intact	Exposed	NA	NA
Thakkar et al ¹⁴		NA	NA	Exposed	NA	NA	NA	NA
Warren et al ¹⁵	3	30	Incidental finding	Exposed	Intact	Exposed	Exposed	Transmastoid approach
	4	13	Neck mass	NA	Eroded	NA	Exposed	Canal wall-down technique + neck extension
	6	9	Ear pain	Intact	Intact	Exposed	Intact	Canal wall-up technique
Present study	1	71	Mastoid and neck pain	Occluded	Intact	Exposed	Exposed	Mastoidectomy + sigmoid sinus packing
	2	77	Dizziness	Exposed	Intact	Exposed	Exposed	Mastoidectomy
	3	60	Incidental finding	Exposed	Intact	Exposed	Exposed	Mastoidectomy + sigmoid sinus packing

EAC — external auditory canal; NA — data not available.

surgery, the mastoid cavity was obliterated with abdominal fat; the ear canal and ossicles were left untouched.

At 18 months after surgery, the patient was doing well, his facial nerve function was grade I (House-Brackmann grading), and his hearing was preserved; MRI and a CT scan (Fig 4B) did not reveal a residual lesion.

Literature Review. In our literature review we found 437 articles, of which 111 were selected on the basis of their title and because they were in English. Of these, we selected 41 after reading the abstracts. From these, 20 cases of CC of the mastoid process were identified in 11 articles (Table 1^{1,10-15} and Table 2¹⁵⁻¹⁹).

DISCUSSION

Congenital cholesteatoma has been reported to originate in the petrous apex, the middle ear, the mastoid process, and the external auditory canal. The middle ear and the petrous apex are well established as sites of origin for CC.^{2,3,6,7} In contrast, CC of the external auditory canal is usually caused by congenital aural stenosis,⁴ and the true existence of primary CC in this site is uncertain.⁵ The mas-

toid process is undoubtedly the least-reported site for the onset of CC.⁸ We have found 20 cases of CC arising in the mastoid process in the literature; however, in our opinion, only 9 may be defined as pure CC of the mastoid process (Table 1). In all of the other cases, there was involvement of the middle ear and/or the petrous apex, or there were some features that made it impossible to differentiate them from acquired cholesteatoma (Table 2).

Definition. We believe that CC of the mastoid process should be defined as cholesteatoma with 1) all the features of a CC (normal tympanic membrane, no previous ear surgery, no history of ear discharge) and 2) no involvement of the middle ear, attic, or aditus as confirmed by radiologic and intraoperative findings.

Congenital cholesteatoma of the middle ear is usually diagnosed at an early stage (before posterior growth into the mastoid process) because of the appearance of conductive hearing loss produced by ossicular chain involvement. It is reasonable to believe in the existence of a third group of CC that originates from the antrum and/or aditus, which could grow into both the middle ear cleft and the mastoid process, and may include some cases previously re-

TABLE 2. PREVIOUSLY REPORTED CASES THAT DO NOT FULLY FIT ALL CRITERIA FOR CONGENITAL CHOLESTEATOMA OF MASTOID PROCESS

Authors	Patient No.	Reason for Exclusion	
Warren et al ¹⁵	1	Epitympanum involvement, ossicular erosion	
	2	Hearing loss, epitympanum involvement, ossicular erosion	
	5	External auditory canal destruction, ossicular erosion	
	7	Hearing loss, epitympanum involvement, ossicular erosion	
	8	Hearing loss	
	9	Epitympanum involvement, ossicular erosion	
	Borgstein et al ¹⁶	1	Hearing loss, petrous bone cholesteatoma
	Rashad et al ¹⁷	1	Hearing loss, middle ear involvement
		2	Otorrhea, external auditory canal fistula
Adjibabi et al ¹⁸	1	Otorrhea, polyp in external auditory canal	
Lee et al ¹⁹	1	Hearing loss, otorrhea, external auditory canal destruction	

These cases represent kind of combined middle ear and mastoid process congenital cholesteatoma, as they have characteristics of both locations of origin.

ported in the literature.^{15,17,18} In our opinion, these cases of CC are hard to classify, as they could be cholesteatoma of the mastoid process involving the middle ear or vice versa, or antrum and/or aditus cholesteatoma growing in both directions. A combined CC of the middle ear and the mastoid process seems to be a definition with characteristics from both groups (Table 3).

Origin. The origin of CC is still unclear. The four main theories are implantation, invagination, metaplasia, and epithelial rest. The last is the most accredited theory, even though epithelial rests in the fetus have been described in the middle ear but never in the mastoid process.²⁰

Presentation. The presentation of CC of the mastoid process differs from that of CC of other locations because of the lack of symptoms for a long period. The disease may not be diagnosed until adulthood, unlike the young age of diagnosis of CC at the other sites.¹³ In fact, because of its particular location, CC of the mastoid process produces no symptoms until it reaches a large size and involves the surrounding structures. In the asymptomatic stage, diagnosis occurs only as an incidental finding (as in patient 3); once there are any symptoms, they are nonspecific, such as retroauricular and/or neck pain (as in patient 1), or retroauricular swelling or

TABLE 3. DIFFERENCES BETWEEN CONGENITAL CHOLESTEATOMA OF MIDDLE EAR AND THAT OF MASTOID PROCESS

	Middle Ear	Mastoid
Presentation	Hearing loss	Temporal area and neck pain, dizziness
Age at diagnosis	Childhood	Adulthood
Findings of otoscopy	Cholesteatoma behind eardrum	No abnormalities
Findings of imaging	Restricted to middle ear	Restricted to mastoid process

dizziness (as in patient 2). Retroauricular pain and swelling seem related to mastoid cortex erosion and periosteum involvement, whereas neck pain is probably due to the inflammation of the insertion of the muscles into the mastoid process. Dizziness has been attributed to ipsilateral cerebellar hemisphere compression,¹³ which in our opinion seems unlikely. Mild extradural compression usually does not create any dizziness. The dizziness in these cases is probably related to endolymphatic sac compression, which was present in all of our patients. Only in 1 case presented by Derlacki and Clemis¹⁰ was erosion of the bony labyrinth considered responsible for the vestibular symptoms. The hearing is usually normal in patients with CC of the mastoid process (as in all of our patients), because the middle ear is not involved. Clinical examination findings are usually negative except for cases with retroauricular swelling.

Diagnosis. Confirmation of the diagnosis of CC is based on radiologic examination. A high-resolution CT scan demonstrates a soft tissue mass that has created a regular bony erosion and usually exposes surrounding structures such as the facial nerve, sigmoid sinus, jugular bulb, posterior cranial fossa dura, and endolymphatic sac.¹⁴ On MRI, the cholesteatoma is hypointense or isointense on T1-weighted images and hyperintense on T2-weighted images; either the lesion does not enhance after gadolinium infusion or there is only some rim enhancement due to surrounding inflammatory tissue.²¹ The differential diagnosis includes tumoral lesions, cholesterol granulomas, and histiocytosis of the temporal bone. However, all temporal bone tumors enhance after gadolinium infusion, whereas cholesterol granulomas can be distinguished by a bright signal intensity on both T1-weighted and T2-weighted images. Histiocytosis may resemble cholesteatoma of the mastoid process, but there is often variable enhancement after administration of gadolinium. In addition, the age of presentation is frequently less than 15 years, whereas CC confined to the mastoid process is discovered mostly in adulthood. The recent introduction of non-echo planar imaging diffusion-weighted

sequences represents a very helpful and accurate additional tool in doubtful cases.²²

Surgical Key Points. As for all cholesteatomas, surgery represents the only treatment method. In cases of CC of the mastoid process, an enlarged mastoidectomy is required, without extension to the middle ear cleft. The only reason to perform subtotal petrosectomy or a canal wall-down technique would be significant thinning of the posterior wall of the external auditory canal. These approaches, as well as a canal wall-up technique, may also be required if there is involvement of the middle ear or attic,¹⁵ although these cases cannot be considered pure mastoid process CC and are more a combined middle ear-mastoid process CC.

During surgery, there are several items to be aware of because of frequent involvement of the facial nerve, sigmoid sinus, jugular bulb, and posterior cranial fossa dura. The facial nerve is usually exposed in the third portion (as in our 3 patients); preoperative knowledge of the facial nerve exposure and the relatively easy dissection of the cholesteatoma matrix from the epineurium make management of this complication not particularly dangerous. The posterior fossa dura is treated as in petrous bone cholesteatoma.⁴ In the majority of cases, dissection of the matrix from the dural surface is accomplished; when it is not feasible, bipolar coagulation of the involved dura is preferred to its removal, in order to reduce the risk of cerebrospinal fluid leak.

Involvement of the venous system of the temporal bone, as often reported,^{1,10,11,13-15} is what requires the most careful planning. Preoperative magnetic resonance angiographic sequences may be used to evaluate the patency of the sigmoid sinus and the jugular bulb on both sides. When the sigmoid sinus and the jugular bulb are patent before operation, it is important to know which side is dominant. If they are patent, only the nondominant vein can be obliterated during surgery without any complication. However, in cases with an absence of flow, it is extremely difficult to distinguish simple sinus compression from thrombosis. The surgeon should be prepared to man-

age bleeding from the sinus, which can also require intraluminal packing. In the presence of massive involvement of the jugular bulb, it may be advisable to isolate the jugular vein into the neck, in order to be ready to ligate it in case of significant bleeding. In our patients it was possible to preserve the integrity of the venous system in 1 case, the sigmoid sinus was already thrombosed in 1 case, and it was necessary to pack it in 1 case. Anatomically, the sigmoid sinus, like all the intracranial sinuses, is an endothelium-lined, trabeculated venous channel encased within a double (periosteal and meningeal) dural layer.²³ The external periosteal layer is what gives the sigmoid sinus wall its resistance and allows surgical manipulation. This layer progressively disappears during the transition between the sigmoid sinus and the jugular bulb, so that at this level the vessel is much more fragile. If the dural layer is destroyed or thinned by the disease, preservation of the anatomic integrity of the sinus becomes almost impossible (as in patient 3). In our opinion, the extensive bony erosion frequently created by the lesion makes it necessary to obliterate the surgical cavity with fat, instead of leaving important structures exposed.

CONCLUSIONS

Congenital cholesteatoma purely located in the mastoid process is very rare; it appears even more exceptional when the literature is critically reviewed. The symptoms and characteristics of CC in the mastoid process clearly differ from those of CC in the middle ear. Congenital cholesteatoma of the mastoid process often appears in adulthood, is not seen on otoscopy, and has pain or dizziness as its first symptom; some asymptomatic cases are even found by chance. The diagnosis is made by imaging; MRI is currently superior, but a CT scan may better show details of the eroded bone and exposed structures. Surgery is the only treatment method, and careful manipulation of the exposed delicate structures is required in order to avoid complications. Some previously reported cases have characteristics of both middle ear and mastoid process CC and may be considered a combination group.

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