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Original Article

Congenital Retrosigmoid Cholesteatoma: Case Series and Literature Review

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BACKGROUND: This study aimed to discuss 3 cases of congenital cholesteatoma located posterior to the sigmoid sinus, with no/minimal involvement of mastoid, and compare them with cases presented in the literature to better define this rare entity.

METHODS: Retrospective chart analysis of 3 congenital cholesteatomas located posterior to the sigmoid sinus treated surgically in 2 skull-base centers and literature review. Though congenital cholesteatoma can arise outside the middle ear, only a few cases presenting in the retrosigmoid occipital bone have been described earlier.

RESULTS: In all 3 patients, there was a delay in the presentation, as symptoms were nonspecific or lacking, leading in 1 case to severe complications. Computed tomography and magnetic resonance imaging, especially diffusion-weighted imaging scans, allowed accurate diagnosis and surgical planning. Surgery happened to be challenging due to the tight adherence of the cholesteatoma to the thinned dural surface. Complete excision was achieved in all the cases.

CONCLUSION: Congenital cholesteatoma located posterior to the sigmoid sinus is a rare entity and is even more exceptional after a critical review of the literature. Complete excision is quintessential to prevent intradural extension or infection. The most important surgical issue is the management of the posterior fossa dura and the sigmoid sinus. We recommend meticulous dissection with slow peeling of the epithelial lining from the dura. Bipolar coagulation of the dura may help in avoiding recidivism. Moreover, cerebrospinal fluid (CSF) leak during dissection has to be avoided as long as possible, because the loss of tension of the already thinned dura makes its peeling particularly difficult.

KEYWORDS: Congenital, cholesteatoma, epidermoid, intradiploic, occipital, retrosigmoid

INTRODUCTION

Congenital cholesteatomas (CC) are rare lesions, especially the intradiploic ones (developing into the cancellous bone between the inner and outer tables of the calvaria) arising outside the middle ear and mastoid. Several cases of intradiploic CC of the occipital bone and temporo–parieto–occipital junction have been previously reported in the English-language literature, but only a few cases are reported in the retrosigmoid area. We discuss 3 cases of CC located posterior to the sigmoid sinus and critically review the literature.

MATERIAL AND METHODS

We conducted a retrospective chart review of 3 CCs exclusively located posterior to the sigmoid sinus, treated surgically at the otorhinolaryngology and otoneurosurgery department, University Hospital of Parma, Parma, Italy and the ENT and Skull Base Department, Sint-Augustinus Hospital, Wilrijk, Antwerp, Belgium. A literature review was performed using the PubMed database with the keywords "congenital," "cholesteatoma," "epidermoid," "intradiploic," and "occipital." This study did not need any approval by the Area Vasta Emilia Nord Institutional Review Board nor required patients' informed consent, as specifically indicated in the founding regulation (version 3, 07/05/2019).

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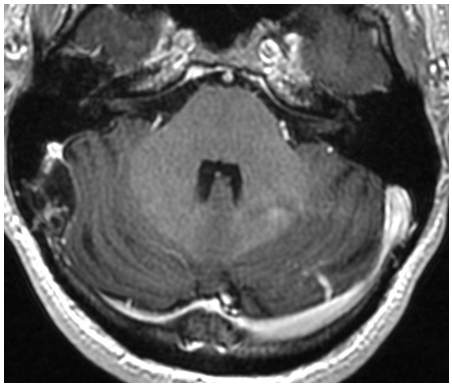

Figure 1. Axial MRI contrast-enhanced T1-weighted images in patient 1, showing a right hypo-intense retrosigmoid lesion surrounded by an irregular rim enhancement.

RESULTS

Case 1

A 35-year-old male presented with a history of right-sided headache of 2 months duration. There was no history of trauma, otorrhoea, or any other otologic symptoms. There were no cranial nerve palsies. There was no relief with conventional analgesics. Otoscopy was normal. Pure tone audiometry revealed normal hearing thresholds. CT showed right-sided lytic lesion behind the mastoid in the occipital region. Mastoid air cells and the middle ear were not involved. Contrast-enhanced T1-weighted images on magnetic resonance imaging (MRI) showed a hypo-intense lesion surrounded by an irregular rim enhancement (Figure 1). The patient did not undergo diffusion-weighted imaging as, at that time, it was not a widespread technique yet. Radiological features were suggestive for cholesteatoma and the diagnosis was confirmed intraoperatively (Figure 2A). During surgery, the lesion was closely adherent to the dura; this latter appeared extremely thinned (Figure 2B), as the external layer had disappeared. The lesion was peeled off from the dura and the posterior aspect of the sigmoid sinus. Mild bipolar coagulation was helpful in establishing a cleavage plane in the most adherent areas. A small dural tear during the removal of the last fragment of the cholesteatoma matrix produced a CSF leak with loss of tension of the dura with consequent increasing difficulty in final disease removal. Complete macroscopic excision of the cholesteatoma was achieved. To reduce the risk of skin remnants all the involved dural surface was gently coagulated at the end of the procedure. The dural tear was closed with a small fascia fragment stabilized with fibrin glue. Postoperative recovery was uneventful. Control imaging after 6 months showed no signs of residual lesion. The patient died for unrelated reasons 2 years after the surgery.

Case 2

A 52-year-old female was referred after the incidental discovery of a large lesion in the left occipital bone during an MRI performed for a left-sided persistent headache of 20 years duration. She complained of episodes of nausea and transient loss of consciousness of 6 months duration. There was no history of trauma, previous surgery, nor any other otologic or vestibular symptoms. Clinical examinations revealed no cranial nerve palsies and a normal otoscopy. Pure tone audiometry showed normal hearing thresholds bilaterally. Temporal bone CT showed left-sided occipital lytic lesion of $32 \times 14 \times 37$ mm dimension, without extension to the mastoid air cells and middle ear (Figure 3A). Contrast-enhanced T1-weighted images on MRI showed a hypo-intense lesion bounded by an irregular peripheral enhancement, with its anteromedial aspect in close contact with the sigmoid sinus (Figure 3B). Diffusion-weighted imaging (DWI) scans revealed very high signal intensity, due to diffusion restriction, strongly suggestive for cholesteatoma (Figure 3c). During surgery, the outer surface of the cortical bone appeared interrupted by the presence of a cholesteatoma. The latter was located posterior to the sigmoid sinus and closely adherent to it and to the posterior fossa dura (Figure 4a). The lesion was peeled off from the sigmoid sinus and the dura, even in this case, with the help of bipolar coagulation. Complete excision was achieved (Figure 4b), followed by mild bipolar coagulation of the involved dural surface. Postoperative recovery was uneventful, except for a minimal retroauricular seroma, and she was treated as an outpatient. Two years after the surgery, the patient was symptoms free and the control MRI was negative for residual lesions (Figure 5a and b).

Case 3

A 51-year-old female patient was first seen because of a pressure sensation in the left ear, with a severe headache. There was no history of

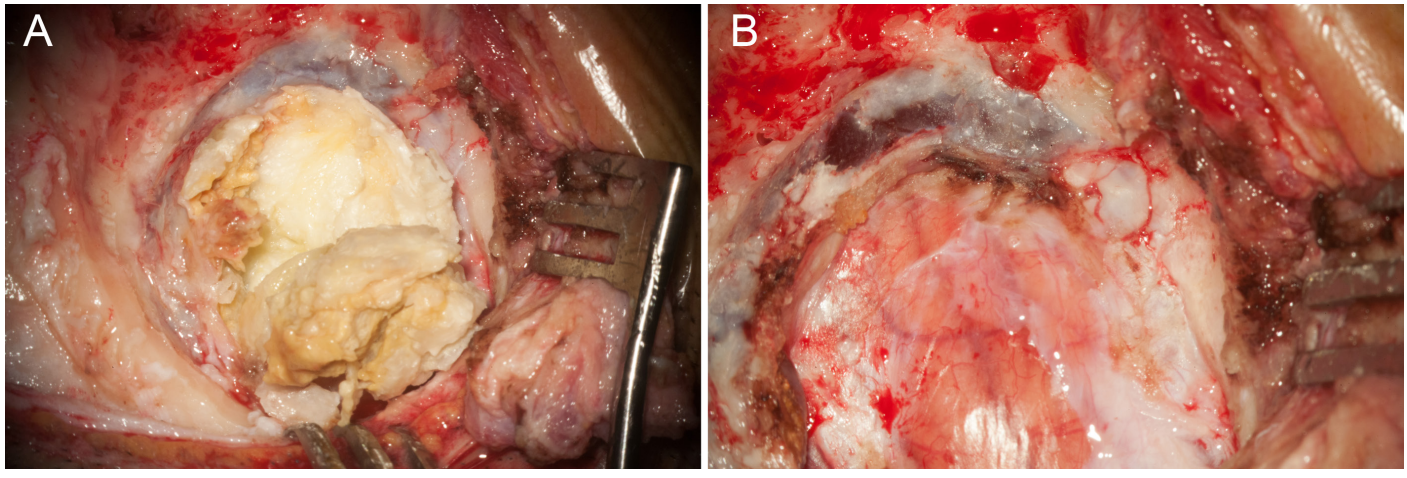

Figure 2. Intraoperative findings in patient 1. In panel (A) the lesion has the typical appearance of a cholesteatoma. Panel (B) shows that the lesion is closely adherent to an extremely thinned dura.

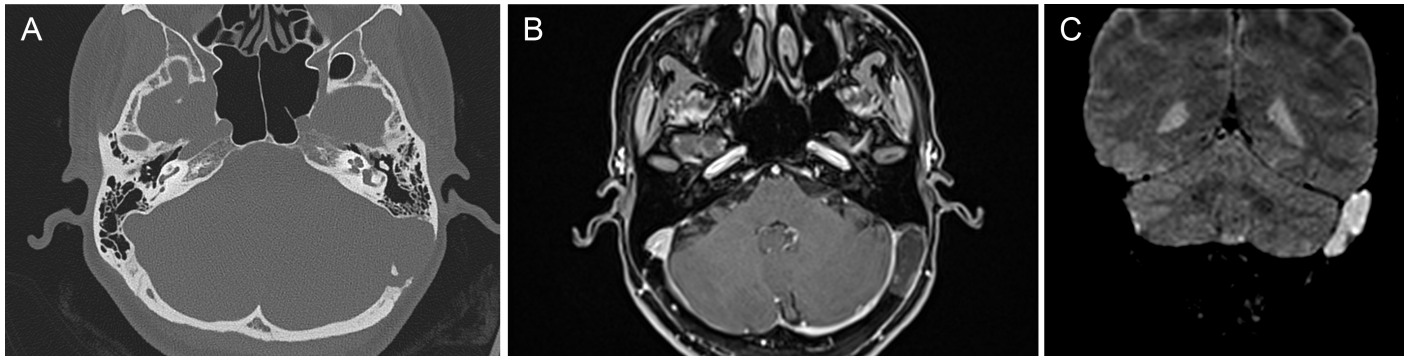

Figure 3. Radiological findings in patient 2. Panel (A) shows the typical lytic aspect of the left occipital lesion. In panel (B), MRI contrast-enhanced T1-weighted images showing a hypo-intense lesion bounded by an irregular peripheral enhancement, with its antero-medial aspect in close contact with the sigmoid sinus. Panel c reveals the high signal intensity in DWI scans, strongly suggestive of cholesteatoma.

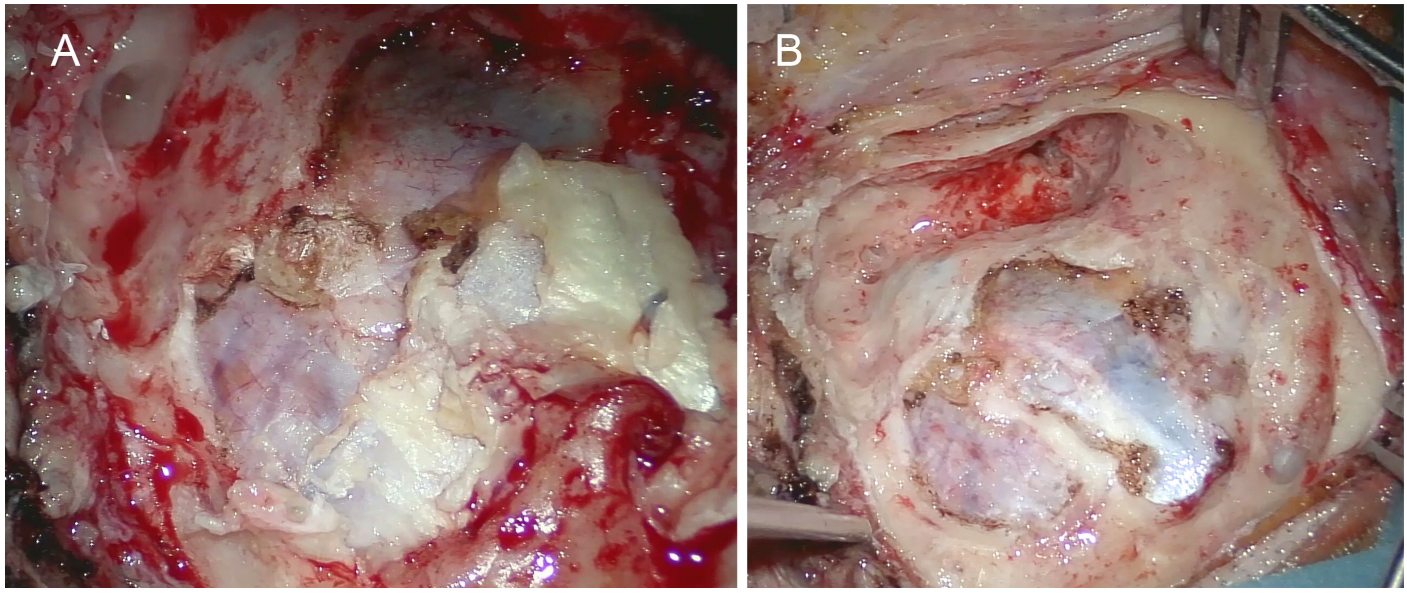

Figure 4. Intraoperative findings in patient 2. In panel (A) the cholesteatoma appears to be located posterior to the sigmoid sinus and closely adherent to it and to the posterior fossa dura. Panel B shows the surgical field after complete excision.

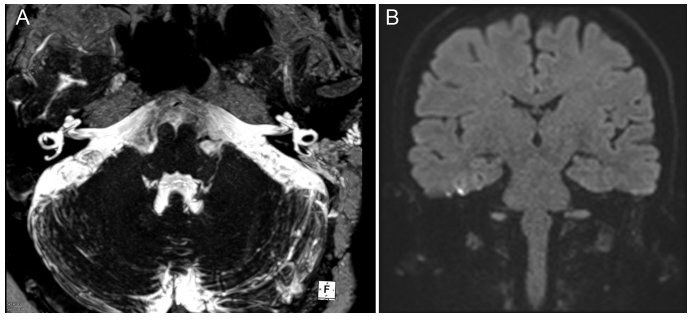

Figure 5. Postoperative radiological follow-up in patient 2. In panel (A), the axial MRI contrast-enhanced T1-weighted images and in panel (B), the coronal DWI scans were negative for residual lesion.

chronic otitis media nor any cranial trauma. The otoscopy, the audiogram, and impedance measurements were all normal. There were no neurological signs. No imaging was done at that time because of the normal hearing thresholds and the absence of neurological signs. Three months later, the patient was admitted to the hospital because of meningitis due to a Haemophilus influenzae infection, which was first treated with antibiotic therapy. An intra-cerebellar abscess was identified on MRI and subsequently drained by the neurosurgeons (Figure 6a). Because of the presence of an important erosion of the bone posterior to the mastoid, evident on CT, exploration of the retrosigmoid area was accomplished, showing a cholesteatoma. This latter had also perforated the dura in the posterior fossa and was in connection with the drained abscess, sticking to the cerebellar

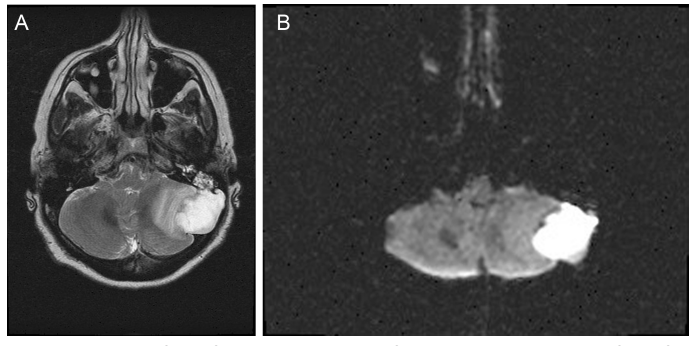

Figure 6. In panel (A), the preoperative axial MRI in patient 3. In panel (B), the high signal on DWI, suspicious for residual cholesteatoma, after neurosurgical abscess drainage.

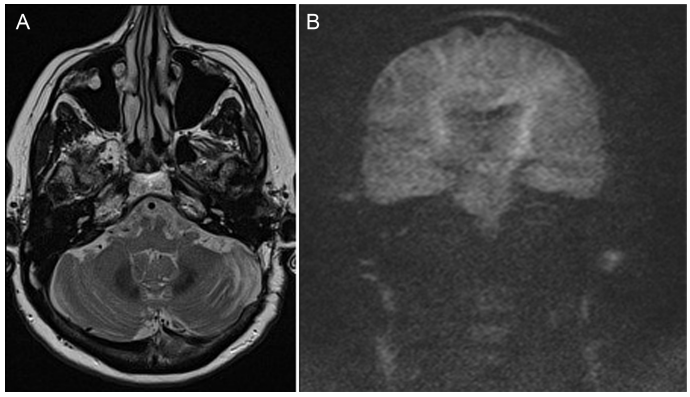

Figure 7. Postoperative radiological follow-up in patient 3, showed no residual cholesteatoma in axial (panel A) and DWI coronal (panel B) scans.

tissues. The patient was later referred to the ENT department because the postoperative MRI showed a high signal on DWI, suspicious for residual cholesteatoma at the transition zone between the mastoid and the occipital bone (Figure 6b). During surgery, residual cholesteatoma was found in the area between the mastoid and the occipital bone. After emptying, there was an extension of this cystic cavity toward the cerebellum where the abscess had been drained. The epithelium lining this cavity, the bone, and the dura was progressively removed micro-mechanically. Further, bipolar coagulation of the dura was performed in order to prevent any residual epithelial cells to regrow. No dural leak occurred. Abdominal fat was used to fill the cavity as well as the mastoid, in order to close any communication between the middle ear and the surgical cavity. A control MRI at 1, 3, 7, and, most recently, 14 years later showed no residual cholesteatoma (Figure 7A and B).

Every patient - except patient 1 - signed an informed consent for images/video collection and publication, and provided a verbal consent to be part of the study.

Literature Review

In our literature review, we found 71 articles, 37 of which were selected on the basis of their English language and their title. We chose 26 of them after reading the abstracts. Finally, 26 cases of retrosigmoid CC were identified in 21 articles (Table 1); additional cases in which it was impossible to exclude a mastoid origin were not considered for the study.

DISCUSSION

Among CC of the temporal bone, the middle ear variant is the most frequent type, followed by petrous bone and mastoid.[1-3] Intradiploic cholesteatomas are encountered occasionally. They may involve various sites within cranial bones: frontal, parietal, occipital, and temporal bones and their junction sites.[4,5] Intradiploic cholesteatomas of the cranial bones have been reported by various authors, since the early 20th century[6-8] to the last 30 years.[4,5,9,10] A meticulous literature review outlined a total of 26 CCs located across the temporo–parieto–occipital junction, as listed in Table 1. Only 14 of them are centered on the retrosigmoid area, as in our series. Seven have a more posterior position with a tendency to reach the median line, especially the bigger ones. The remaining 5 cases are presumed to have a similar origin to our cases, as they lack an iconographic documentation of the exact location.

History and Definition

The terms "cholesteatoma" (Mueller, 1838) and "epidermoid" (Von Remak, 1858) were coined for lesions containing cholesterol crystals, arising from embryonic epithelial cell rests. Since the first reports from Bailey's and Cushing's (1920 and 1922), other author's published cases of epidermoids, with a total number of 223 primary diploic locations.[4,6,7,11] From Miller's study,[4] very few cases have been found, with only 12 cases with a retrosigmoid location.

Whether the term "cholesteatoma" or "epidermoid" is to be used as a synonym is a matter of discussion already developed by Clark et al.[9] The term "cholesteatoma" has a tendency to be used more frequently by ENT surgeons,[1,5,9,12] while the term epidermoid is being used more often by neurosurgeons.[11,13-24] We agree with Clark et al[9] that histopathologically and radiologically, intradiploic CC and intradiploic epidermoid cysts are the same entity. However, in order to avoid confounding nomenclature, we feel that the extradural lesions should be referred to as "congenital cholesteatomas" while the term "epidermoid" should be used for the intradural locations.

Origin

The origin of CC is still unclear. The 4 main theories are implantation, invagination, metaplasia, and epithelial rest. The last one is the most accredited theory[2] and can be applied also to the retrosigmoid location.

Presentation

A retrosigmoid CC often lacks symptoms until it reaches a large size and involves the surrounding structures. In the asymptomatic stage, diagnosis occurs only as an incidental finding. When present, symptoms are nonspecific, with headache reported as the most common (it was present in all our patients). A high index of suspicion is necessary to reach the diagnosis; as a consequence, it is not uncommon that the disease is only diagnosed at an adult age. Sometimes a tender mass might be palpable; however, large lesions may cause complications such as intracranial hypertension, meningitis (our case 3), seizures, or focal neurological deficits.

Diagnosis

The diagnosis of CC is based on radiologic examination. A high-resolution CT scan demonstrates intradiploic bony erosion caused by a soft tissue mass, usually exposing surrounding structures such as sigmoid sinus, and posterior cranial fossa dura. On MRI, the lesion has the classic features of a cholesteatoma: being hypo-intense on T1-weighted images and hyper-intense on T2-weighted images. Gadolinium enhancement is absent or limited to peripheral rim enhancement due to surrounding inflammatory tissue. With DWI, cholesteatoma shows very high signal intensity due to diffusion restriction.

Surgical Key Points

Though benign in nature, cholesteatoma, with its bone destruction property, can produce serious complications. The long-lasting growth of the lesion, due to the lack of symptoms, can cause dural compression and consequent significant thinning, as happened in 2 of our cases. If undetected, the lesion can tear the dural barrier and become intradural. Cases of malignant conversion to squamous cell carcinoma have been reported. Therefore, complete excision should be the aim of surgery, as it is of utmost importance in order to avoid recurrences. Except for 4 cases,[5,7,9] all the other authors in

Table 1. Cases of Pure Congenital Retrosigmoid Cholesteatoma

Authors	Date	Age (y)	Sex	Site	Side	Size (cm)	Symptoms	Complete Excision	Residual/recurrent (MONTHS)
Cushing[6]	1922	40	M	TPO	L	13	Mass	Yes	NA
King[7]	1939	26	M	O	L	8	Mass, headache	**No**	NA
Rowbotham[8]	1939	35	F	O	L	NA	Headache	Yes	NA
Canale et al[24]	1974	46	M	TPO	R	14	Mass, headache	NA	NA
Rubin et al[21]	1989	21	F	TP	R	NA	Mass, headache	Yes	no (60)
		17	M	TP	R	NA	Mass	Yes	no (48)
		21	M	PO	R	NA	Mass	Yes	no (24)
Ciappetta et al[11]	1990	14	F	T	L	NA	Mass	Yes	no (180)
		40	M	O	R	NA	Headache	Yes	no (120)
Guridi et al[16]	1990	47	M	O	R	7	Mass	Yes	NA
Atabay et al[13]	1994	51	M	O	L	8	Mass, headache	Yes	NA
Miller et al[4]	1994	19	M	TO	L	2.5	Mass, headache	Yes	NA
Luntz et al[1]	1997	54	F	TO(M)	R	NA	Neck pain, instability	Yes	no (9)
Piotin et al[20]	1998	39	M	TO	R	NA	Non pulsatile tinnitus	Yes	no (4)
Canalis et al[5]	2002	63	M	TPO (M)	R	7.5	Mass	**No**	yes (14)
		35	M	TPO	R	3.5	Headache	**No**	NA
Bikmaz et al[14]	2005	46	F	O	L	NA	Headache, dizziness	Yes	NA
		35	F	TO (M)	L	5	Headache, disequilibrium	Yes	NA
Clark et al[9]	2008	52	F	TO	L	NA	Headache	**No**	NA
Kumaran et al[10]	2010	55	M	O	R	7.4	Headache	Yes	NA
Davidoss et al[12]	2014	64	M	TO(M)	L	12.7	Hearing loss	Yes	no (6)
Moreira-Holguin et al[18]	2015	42	M	O	R	NA	Headache	Yes	no (12)
Ylmaz et al[22]	2016	27	M	O	L	2.4	Mass	Yes	NA
Oommen et al[19]	2018	46	F	O	R	7	Mass, headache	Yes	no (2)
Dąbrowski et al[15]	2018	56	M	O	L	NA	Dizziness, memory disorder, disequilibrium	Yes	NA
Kuwano et al[17]	2020	49	F	TO(M)	L	NA	Otitis media with effusion	Yes	no (6)
Present series	2021	35	M	O	R	3.1	Headache	Yes	no (6)
		52	F	O	L	3.7	Headache, nausea, loss of consciousness	Yes	no (26)
		51	F	O	L	5.2	Headache, ear pressure, meningitis	Yes	no (168)

M, extension to the mastoid; NA, data not available; O, occipital; P, parietal; T, temporal.

the literature achieved a complete excision of the cholesteatoma (Table 1). Bipolar coagulation may be helpful in establishing a correct cleavage plane between cholesteatoma matrix and dura in the most sticking areas during mechanical dissection. To reduce the risk of any residual epithelial cells to regrow, a final dura coagulation could be performed as the last step of the surgery. Removal of the dura has been suggested by Constans et al[23] but seems unnecessary when using the technique described above, thereby avoiding CSF leak.

We conclude that CC located outside the temporal bone is infrequently encountered. We believe that only the extradural epithelial lesions should be referred to as "cholesteatoma," while the term "epidermoid cyst" has to be reserved to intradural location. A complete surgical excision should be tried in all cases to avoid recurrence. Management of the posterior fossa dura is the key point of the surgical treatment.

Ethics Committee Approval: This study did not need any approval by the Area Vasta Emilia Nord Institutional Review Board, as specifically indicated in founding regulation (version 3, 07/05/2019).

Informed Consent: Verbal informed consent was obtained from the patients who agreed to take part in the study.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – M.F.; Design – G.D.; Supervision – T.V.H., M.F.; Materials – G.D., T.S., T.V.H., M.F.; Data Collection and/or Processing – T.S., T.V.H., M.F.; Analysis and/or Interpretation – G.D., M.F.; Literature Search – G.D.; Writing – G.D., T.S., M.F.; Critical Review – T.S., T.V.H., M.F.

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