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Original Article

Congenital Mastoid Cholesteatoma

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BACKGROUND: Congenital cholesteatomas account for just up to 5% of all cholesteatomas and most commonly arise in the petrous apex and middle ear. Congenital cholesteatomas arising in the mastoid are rare and typically present late.

METHODS: In this study, we report a case series of 3 cases managed in our department between 2006 and 2021 and present a summary of the current literature.

RESULTS: Congenital cholesteatomas arising in the mastoid is a rare finding and even among reported cases, not all are clearly mastoid in origin. Their location allows for considerable growth before symptoms develop. Pain and localized swelling in the temporal area are the most common presenting symptoms which can lead to diagnostic challenges. Our cases show that although surgery is often appropriate, conservative management may be suitable in certain situations.

CONCLUSION: Congenital cholesteatoma of mastoid origin is rare and can present a diagnostic challenge. Greater awareness is important to facilitate early detection. A high index of suspicion is needed in those presenting with retro-auricular pain and swelling in the context of a normal ontological examination.

KEYWORDS: Congenital cholesteatoma, ear surgery, otology, temporal bone

INTRODUCTION

Cholesteatomas are benign, slow-growing lesions that result in progressive exfoliation of squamous epithelium.[1] They can be congenital or acquired,[1] with congenital cholesteatomas accounting for just 1%-5% of cases.[2] Congenital cholesteatomas mostly arise in the petrous apex and middle ear. However, they may originate elsewhere, including the fourth ventricle and cerebellopontine angle.[3,4] They have also been shown to rarely originate in the mastoid. However, of those reported to have originated in the mastoid, a number have been felt to not truly be mastoid in origin, making it an even rarer entity.[3]

Congenital cholesteatomas are expansive and locally destructive.[1] Presenting features depend on the location of the lesion. Those with origin in the mastoid may be asymptomatic and detected incidentally. Alternatively, they have been shown to present with post-auricular swelling, neck pain, and dizziness.[1] In this study, we report 3 rare cases of congenital cholesteatoma of mastoid origin which were managed with differing approaches and review the current literature.

MATERIALS AND METHODS

We conducted a retrospective case review of all congenital cholesteatomas of mastoid origin managed in our department between 2006 and 2021. This identified 3 cases that we presented here. A literature review was performed to identify previously reported cases of congenital cholesteatomas originating in the mastoid which were summarized and compared to our cases. This retrospective study protocol underwent local assessment and approval at Queen Elizabeth Hospital Birmingham (CARMS-18192). Written informed consent was obtained from all participants who participated in this study.

RESULTS

We identified 3 cases of congenital cholesteatoma of mastoid origin managed by our department between 2006 and 2021 which are described here.

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CASE 1

A 34-year-old woman was referred after imaging revealed a 15 mm × 18 mm lesion in the right temporal bone. She presented to her doctor with a history of temporal headaches, dizzy spells, and numbness in the right arm and had a recent diagnosis of HIV. The lesion in the posterior half of the right mastoid was consistent with a cholesteatoma with high signal on T2-weighted images, mixed low signal on T1-weighted images, and high intensity on diffusion-weighted magnetic resonance imaging (MRI) (Figure 1a). Imaging showed localized bony destruction but a normal middle and internal ear. On examination, her tympanic membrane was normal and pure tone audiometry revealed normal hearing thresholds. She opted to undergo right cortical mastoidectomy which confirmed that the middle ear was clear of disease and isolated to the mastoid. The lesion was removed down to dura and histology confirmed cholesteatoma. She recovered well, post-operative tinnitus settled, and she was soon asymptomatic with good residual hearing. At clinic follow-up 4 years post-procedure, she remained asymptomatic and was disease-free on the latest scan (Figure 1b). She was discharged from the routine follow-up.

CASE 2

A 73-year-old man was referred to his local Ear, Nose, and Throat department with a long history of intermittent painful episodes of swelling in the posterior and inferior left ear. These episodes were treated with oral antibiotics by his general practitioner and were typically resolved within days. There was no history of otorrhea, tinnitus, vertigo, trauma, or previous ear surgery. His hearing was subjectively bilaterally equal. A magnetic resonance imaging scan as requested by his GP showed a well-defined cystic lesion involving the mastoid and occipital bone and lying very close to the left hypoglossal canal.

Otoscopy was normal, revealing intact tympanic membranes bilaterally. Cranial nerve examination was normal. Pure tone audiometry demonstrated a bilateral, high-frequency sensorineural loss in keeping with age-related hearing change (presbyacusis).

Following the initial MRI, the referring team felt that the most likely diagnosis was a mastoid mucocele, but due to the unusual location and extensive skull base involvement, the patient was referred to the regional lateral skull base service for further management.

Additional scans were performed to establish the diagnosis and to aid with surgical planning. Computed tomography (CT) (Figure 2a) showed an extensive soft tissue mass of low density occupying most of the mastoid portion of the temporal bone, markedly thinning the bone overlying the lesion, without any erosive or permeative process and a structurally normal and well-aerated mastoid with some displacement of the sigmoid sinus medially. To help distinguish it from other differential diagnoses, a diffusion-weighted MRI was performed (Figure 2b). This confirmed the presence of a large mass (approximately 4 cm × 4.3 cm × 4.3 cm) and provided the added information of high T2 signal on b1000 with restricted diffusion and non-enhancement on the post-contrast sequences, indicating the diagnosis of cholesteatoma.

The management of cholesteatoma can be conservative with serial imaging. However, for those who are symptomatic, surgery is the only effective management. For our symptomatic patient, a postauricular approach was performed. Elevation of the subperiosteal flap and uncapping of the mastoid revealed the extensive cholesteatoma (Figure 2c) with a 2 mm breach in the cortex of the temporal bone. The lesion was expanding to the mastoid bone and in contact with the middle fossa and posterior fossa, dura, sigmoid sinus, and an exposed vertical segment of the facial nerve extending inferiorly toward the digastric ridge, the mastoid tip, and jugular bulb. It was found to be extending anteriorly toward the sinus tympani, posteriorly to the retrosigmoid dura, medially toward the internal carotid

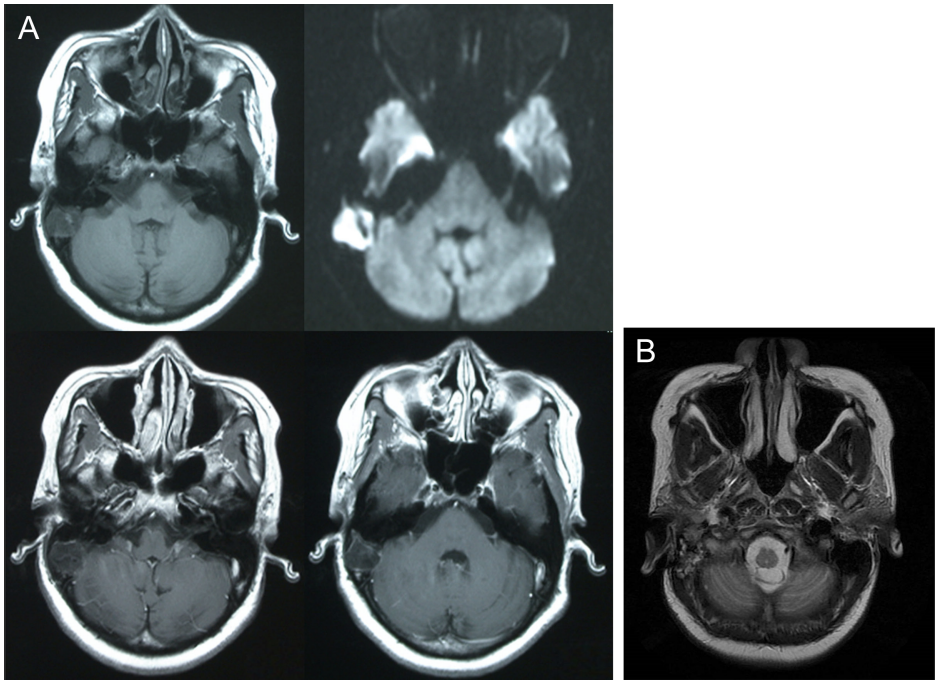

Figure 1. a, b. (a) Pre-operative scans showing cholesteatoma isolated to the posterior mastoid bone. (b) MRI of 3.5 years post-procedure showing no residual or recurrent disease. MRI, magnetic resonance imaging.

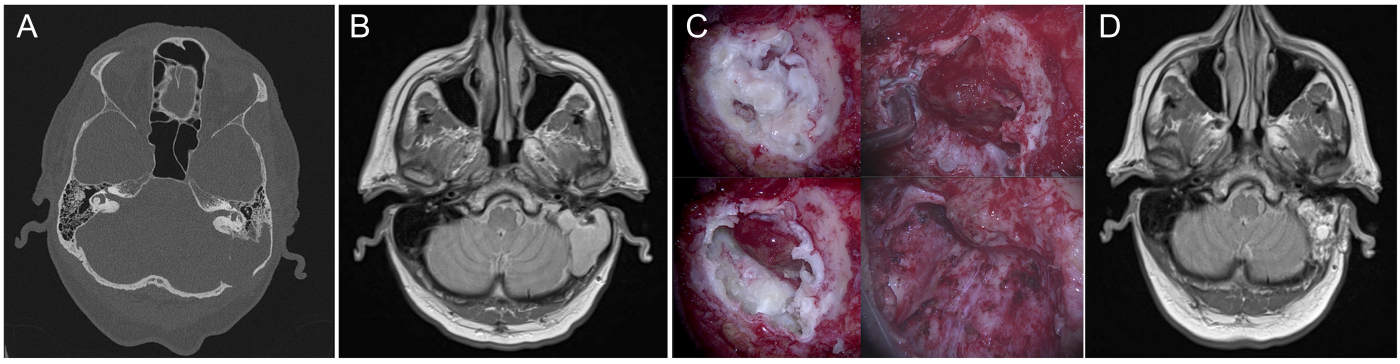

Figure 2. a-d. (a) CT scan demonstrating normal middle ear and an absence of middle ear origin/involvement in the cholesteatoma. (b) - Diffusion-weighted MRI showing lesion with high T2 signal, high signal on the b1000, restricted diffusion (ADC 500), and non-enhancement on the post-contrast sequences, consistent with a congenital cholesteatoma. (c) - Intra-operative images showing removal of cholesteatoma. (d) Post-operative MRI showing no evidence of residual or recurrence. CT, computed tomography; MRI, magnetic resonance imaging.

artery, and vertically to the facial nerve. New bone formation was seen over the mastoid antrum obscuring the view of the middle ear structures and the second genu of the facial nerve.

The cholesteatoma was gently removed from the dura and facial nerve. The mastoid tip was excised due to disease involvement. As the middle ear was normal on CT imaging, the new bone over the mastoid antrum, labyrinth, and second genu of the facial nerve was not disturbed. The cholesteatoma was then traced along the middle fossa dura, retrosigmoid area, and posterior fossa dura. The epithelium was cleaned off the vertical facial nerve, and dissection was continued toward the digastric ridge. Potassium titanyl phosphate laser was used at 1 w continuous setting to polish the cavity before being obliterated with abdominal fat.

The histopathologic assessment confirmed the diagnosis of cholesteatoma. Post-operatively, the patient recovered well and retained full function of the facial nerve. Repeat diffusion-weighted imaging (DWI) MRI (Figure 2d) was performed which did not reveal any residual or recurrent disease. At the 6-month follow-up, the patient was well with no post-operative complications. A further scan is planned after 12 months.

CASE 3

A 48-year-old man under the care of the hematologists with mantle cell lymphoma, managed with intensive chemotherapy followed by autologous stem cell transplantation, developed headaches and visual disturbance 20 days post-transplantation. A CT head (Figure 3a) showed increased thickness of a lytic occipito-mastoid lesion and the right convexity dura. An MRI head (Figure 3b) then confirmed the presence of an enhancing right temporal lesion. The patient's symptoms settled, and he was asymptomatic. Multidisciplinary team (MDT) review of the Positron emission tomography (PET) scan showed that the lesion was present in the pre-treatment scan and had a low standard uptake value, suggesting a benign lesion. A DWI MRI was performed which confirmed congenital cholesteatoma measuring $29 \times 24 \times 44$ mm originating from the mastoid. Given that the patient is asymptomatic, immunosuppressed, and recovering from chemotherapy, he is being followed up with surveillance MRI. Repeat MRI after 12 months (Figure 3c) has shown a stable appearance and the patient remains asymptomatic.

Table 1 summarizes the current literature describing congenital cholesteatomas of mastoid origin. Sixteen previous cases are described in relation to their presenting symptoms, imaging performed, size, management, and outcomes.

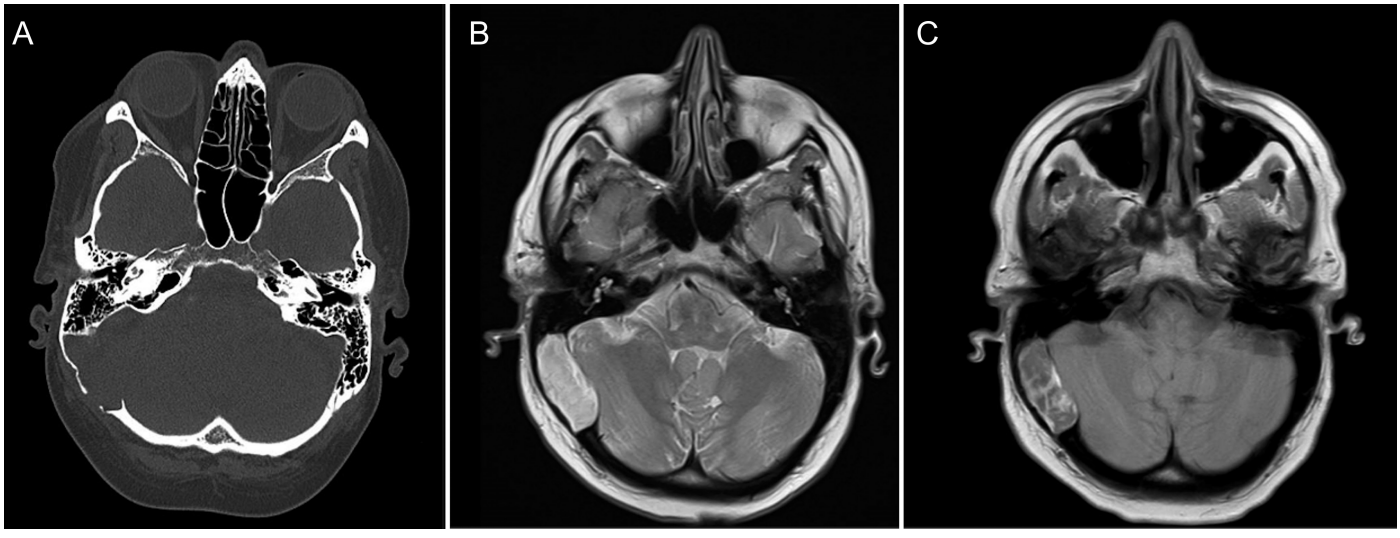

Figure 3. a-c. (a) CT showing a lytic occipito-mastoid lesion. (b) MRI showing the enhancing right temporal lesion. (c) - Stable appearance of lesion at 12-month follow-up. CT, computed tomography; MRI, magnetic resonance imaging.

Table 1. Summary of Literature Review

Reference	Age	Presenting Symptoms	Imaging	Size	Management	Outcomes/Complications
Derlacki & Clemis (1965)[9]	24	Intermittent pain and swelling over right mastoid for 2 years	Mastoid x-rays – 7 × 11 mm cyst in mastoid (1948) Repeat x-rays in 1955 – enlargement in the area of translucence	Not given	Surveillance followed by surgery	No post-operative complications or recurrence of cholesteatoma
Mevio et al (2002)[10]	36	1-year history of recurrent episodes of positional vertigo	CT – expanding lytic mass in the posterior part of the mastoid process DW MRI — mass in the left mastoid cavity that was eroding the posterior and superior walls of the mastoid, invading the middle and posterior cranial fossa.	Not given	Surgery	Resolution of vestibular symptoms on day 1 post-op. No follow-up details were given.
Warren et al (2007)[11]	30	Incidental	CT and MRI – no further details	Not given	Surgery	No post-operative complications reported
	13	Neck mass	CT and MRI – no further details	Not given	Surgery	No post-operative complications reported
	9	Unilateral hearing loss	CT – no further details	Not given	Surgery	No post-operative complications reported
Giannuzzi et al (2011)[5]	71 F	Unilateral mastoid and neck pain	CT – expansile destructive lesion in left mastoid process with a small erosion of the external cortex MRI – hypointense on T1, hyperintense on T2	Not given	Surgery	No reported post-operative complications at 18-month follow-up. Patient declined radiologic follow-up
	77 M	2-year history of vertigo	CT – destructive lesion confined to the right mastoid process. MRI - hypointense on T1, hyperintense on T2. Occupying entire mastoid process	Not given	Surgery	No reported post-operative complications and no recurrence at 1-year follow-up
	60 M	Asymptomatic – incidental finding	CT – lytic lesion in the left mastoid process MRI – classical for cholesteatoma	Not given	Surgery	No reported post-operative complications and no recurrence at 18-month follow-up
Cvorovic et al (2011)[6]	29 F	3-month history of retro-auricular pain, tinnitus, and unilateral mild hearing loss	CT – destruction of the position canal wall by a lesion of soft tissue density in the mastoid cavity. Destruction of bony plates of the posterior fossa and sigmoid sinus with perilabyrinthine propagation	Not given	Surgery	No reported post-operative complications and no recurrence at 3-year follow-up
Hidaka et al (2011)[8]	65 M	2-week history of unilateral post-auricular swelling and pain. Gradual hearing loss	CT – expansile, lytic lesion in the mastoid eroding the bony plate of the posterior fossa and the sigmoid sinus. Abscess in the post-auricular region.	Not given	Surgery – initially an emergency mastoidectomy revealed a post-auricular abscess connecting to the mastoid. The peripheral mastoid was filled with cholesteatoma	2 months after the emergency mastoidectomy for acute mastoiditis, surgery to remove the entire cholesteatoma was undertaken. No further follow-up details are known.

Nagato et al (2012)[12]	10 M	Stricture of left side external auditory canal identified 6 months earlier.	CT temporal bones - destruction of the posterior wall of the external auditory canal by a lesion showing soft tissue density in the left mastoid cells MRI - hypointense on T1 and hyperintense on T2 and hypointense on gadolinium-enhanced T1	Not given	Surgery - destruction of the posterior bony wall of the external auditory canal was found. After removal of mastoid cortical bone, cholesteatoma was observed in the mastoid cavity. CT at 1 year – no cholesteatoma reoccurrence; CT at 2 years – no restriction reoccurrence.	No post-operative complications or recurrence at 2 years follow-up
Kotsiopoulos et al (2012)[13]	52 M	2-week history of left ear otorrhea	CT — an expansile, lytic lesion in the left mastoid process that had eroded the bony plates of the posterior fossa and the bony plate covering the sigmoid sinus and eroded the posterior wall of the external auditory canal	Not given	Surgery – mastoidectomy.	Satisfactory with no post-operative complications reported at 1-month follow-up
Hong et al (2014)[14]	59 M	5-month history of right side ear discharge with no previous right side history.	CT — mass occupying the mastoid tip extending to the posteroinferior wall of the EAC	Not given	Surgery – mastoidectomy. The air cell in the mastoid tip was filled with cholesteatoma.	No post-operative complications. Satisfactory at 3-month follow-up
Davidoss et al (2014)[3]	64 M	Mild hearing loss	CT – extensive soft tissue mass involving the left middle ear cavity, left mastoid, petrous bone, and occipital bone. MRI – well-circumscribed mass in the left occipital bone with contiguous involvement of the mastoid and petrous temporal bones DWI – cholesteatoma confirmed	12.7 × 8.9 × 2.5 cm	Surgery Due to the extent of the mass, it was not possible to determine accurately the site of origin	Surgical bed disease-free at 6 months. Diffusion-weighted MRI also accurately showed 2 small intra-calvarial foci of residual disease that were out of the resection site pending further excision
Fowler et al (2018)[15]	35 M	1-year history of recurrent vertigo, associated with nausea, lasting 10 s and <30 attacks per day	CT temporal bones – large expansile mass originating from mastoid, resulting in erosion of bony posterior semi-circular canal. MRI confirmed extra-axial mass originating from the temporal bone with local mass effect on the cerebellum. T1 weighted images – no enhancement; T2 weighted images – mass was hyperintense	4.5 × 3.1 × 4.3 cm	Surgery	No post-operative complications
Sepehri et al (2018)[7]	87 F	Incidental	CT – expansive, destructive lesion posterior in mastoid process MRI – hypointense on T1 and hyperintense on T2	1.8 × 2.0 × 3.0 cm	Surveillance	Stable at 1-year follow-up

DISCUSSION

Congenital cholesteatomas are rare, accounting for just 1%-5% of cholesteatoma cases.[2] They are thought to develop after there is a failure to reabsorb embryonic epidermoid residue leading to the development of a cyst behind an intact tympanic membrane.[3]

Presentation is dependent on the location of the cholesteatoma. The mastoid is the least frequently reported site for the origin of congenital cholesteatoma.[3] A review of the literature identified 16 cases of mastoid cholesteatoma (Table 1). However, it is not always clear if these are truly mastoid in origin as identified by Giannuzzi et al.[5] Davidoss et al[3] acknowledged that due to the size of the lesion, it was not possible to accurately determine the site of origin. All of our cases involved the mastoid with no involvement of the mesotympanum or epitympanum, distinguishing them from cholesteatomas of middle ear origin. However, it is not possible to determine the precise site of origin, particularly in case 3. Both cases 1 and 2, managed surgically, were extradural and the assumption from the imaging is that case 3 is also extradural. Giannuzzi et al[5] suggested the following definition to distinguish those of clear mastoid origin: (1) a normal tympanic membrane with no previous ear surgery and no history of otorrhea and (2) no involvement of the middle ear, attic, or aditus as confirmed by imaging and intraoperative examination.

When arising in the middle ear, patients often present in childhood with subtle hearing loss. However, when developing in the mastoid, such as in these cases, hearing is typically unaffected and may only be picked up incidentally.[3] This was the case in 4 of the previously reported cases. Congenital cholesteatomas originating in the mastoid are often larger than those seen elsewhere as their location enables significant growth before detection.[6] They are also typically identified in adulthood such as in our cases. Review of the literature revealed an average age of presentation of 47 years when our cases are included, with a range of 9-87 years of age. When symptoms existed, the main presenting features were pain and swelling localized to the temporal area, this was also seen in our second case. Other cases reported neck pain, hearing loss, and acute mastoiditis at presentation.[1] Cvorovic et al[6] suggested that neck pain occurred secondary to inflammation of the muscles inserting into the mastoid process and post-auricular pain due to erosion involving the periosteum.

All but one[7] of the published cases of congenital cholesteatoma of the mastoid were managed surgically. No cases reported any postoperative complications. There is variable information about postoperative follow-up in the literature. The oldest case was reported by Derlacki and Clemis[9] in 1965, prior to the use of surveillance imaging. Giannuzzi et al[5] had 1 patient who declined radiologic follow-up. One case, reported by Hidaka et al[8], was initially managed for acute mastoiditis which revealed the cholesteatoma. They subsequently underwent further surgery to remove the entire cholesteatoma 2 months later.[8] Where information exists, maximal follow-up was 3 years, with no cases reporting any recurrent disease.[6] Our first case was followed up for 4 years, with no evidence of recurrent disease. The patient with the largest reported mastoid cholesteatoma, reported by Davidoss et al[3], which measured 12.7 × 8.9 × 2.5 cm, was found to have a disease-free surgical bed at 6-month follow up with DW-MRI, but it revealed 2 small intra-calvarial foci of residual disease which necessitated further surgery.[3]

Although the surgery was the mainstay of management, as demonstrated by Sepehri et al[7] who reported the incidental finding of a 1.8 × 2.0 × 3.0 cm cholesteatoma in an 87-year-old woman, surveillance is a justifiable option in some patients. Our third case was also managed conservatively as the patient was asymptomatic and had recently undergone a stem-cell transplant.

CONCLUSION

Our cases highlight the diagnostic challenge and management considerations in rare cases such as these. The symptoms and characteristics of congenital cholesteatoma of the mastoid differ in a number of ways from congenital cholesteatoma of the middle ear making this rare entity easy to miss and leading to late diagnosis. It is important to maintain a high index of suspicion in patients presenting with atypical symptoms of retro-auricular pain, with or without dizziness and hearing loss in the context of a normal otological examination.[6] While chronic ear disease is typically painless, secondary inflammation may cause pain. Early diagnosis is best to prevent potentially serious complications[2] and an understanding of differentials based on anatomy and selection and interpretation of imaging is imperative.

Ethics Committee Approval: This retrospective study protocol underwent local assessment and approval at Queen Elizabeth Hospital Birmingham (CARMS-18192).

Informed Consent: Written informed consent was obtained from all patients who participated in this study.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – R.I., P.M.; Supervision – R.I., P.M., W.S.C., J.M.; Data Collection – E.R., J.M., R.I.; Analysis and/or Interpretation – E.R., J.M.; Literature Review – E.R., J.M., W.S.C.; Writing Manuscript – E.R., J.M., R.I.; Critical Review – W.S.C., P.M., R.I.

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