Congenital Cholesteatoma Involving the Eustachian Tube: A Rare Presentation

Thomas A Stewart, MD; Michael Sim, M4; Helen Xu, MD
Loma Linda University, CA

Introduction

Cholesteatomas are non-neoplastic, keratinizing squamous epithelial lesions that can affect many different areas of the skull base. They are most commonly found in the middle ear where they are categorized as either congenital or acquired. Traditionally, the clinical criteria for the diagnosis of congenital cholesteatoma (CC) included a white pearl-like mass behind an intact tympanic membrane (TM), a normal pars tensa and flaccida, no history of otorrhea or infection, no history of TM perforation, and no previous otologic procedure.[1] The most common presentation is an asymptomatic, white mass found behind an intact TM. A small percentage of patients can also present with conductive hearing loss. These lesions can cause serious complications by eroding nearby structures or by precipitating infection, either within the temporal bone or intracranially. The incidence of all cholesteatomas in children is 3 in 100,000 and the incidence of CC has been reported to be 0.12 in 100,000, but the actual incidence may be higher since the lesion can perforate the TM and be misdiagnosed as an acquired cholesteatoma (AC).[4]

The most common location of CC is in the anterior superior quadrant of the mesotympanum, followed by the posterior superior quadrant.[3] Recent studies however, have shown that extensive lesions at presentation are not uncommon, with cases of diffuse invasion of the mesotympanum, extension to the attic, aditus ad antrum, sinus tympani, facial recess, and eustachian tube (ET) being documented.[3,5-7] The theory of pathogenesis of CC remains an area of controversy, with multiple theories currently in contention. They include the acquired inclusion, migration, amniotic fluid contamination, metaplasia, and epidermoid formation theories. We present a first case of CC that originated from the ET with subsequent expansion into the mesotympanum and unusual presentation as chronic otorrhea and TM perforation.

Case Report

A four-month-old female with a history of congenital neutropenia presented with right chronic otorrhea that started when she was one month old. The patient underwent right attempted myringotomy and pressure equalization tube placement at age two and a half months since her otorrhea didn't respond to medical treatment. The patient was examined by a pediatric otolaryngologist and found to have significant granulation tissue in the external auditory canal with no normal TM landmarks identified. The surgery was aborted and the patient was referred to the otology service for further evaluation and management. Other than her right ear infection, and chronic neutropenia, the patient was healthy with no other local or systemic infections.

A high resolution computed tomography (CT) scan of the temporal bone was done initially with findings of opacification of the right middle ear and mastoid with intact ossicles and no bony erosion. A large soft tissue mass was also noticed around the right eustachian tube area (Fig 1.) MRI of the skull base with and without gadolinium was then ordered for further evaluation of the mass. The mass appeared to be inside the right eustachian tube, with low signal in T1 and high signal in T2 images (Fig 2.) There was no communication between the mass and the adjacent internal carotid.

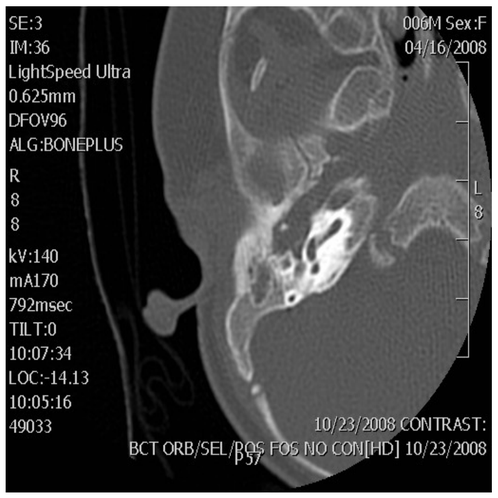
To this point, the underlying pathology of the otorrhea was not clear. Differential diagnoses included: recurrent otitis media, chronic otomastoiditis secondary to neutropenia, histiocytosis of the temporal bone, eustachian tube neoplasm, and congenital cholesteatoma. The patient underwent right middle ear exploration and tympanomastoidectomy at the age of 6 months. Intraoperatively the patient was found to have significant granulation tissue extruding from the middle ear through a large anterior/inferior perforation of the inflamed TM. In the middle ear, a small portion of cholesteatoma with keratin debris was limited to the anterior mesotympanum, with extension of cholesteatoma from the eustachian tube.

Further dissection revealed extensive cholesteatoma inside the eustachian tube. The eustachian tube seemed to be remodeled during the process of the disease with its size about double that of the middle ear. A complete removal of squamous epithelium and keratin debris was confirmed with a retro-insertion of a flexible fiber optic laryngoscope through the nasal orifice of the eustachian tube. The nasopharyngeal portion of the eustachian tube seemed normal. Other parts of the middle ear and mastoid were free of cholesteatoma although there was significant pockets of granulation tissue in other areas. The ossicular chain was intact and mobile. TM perforation was repaired with a medial graft after all disease was removed from the middle ear and eustachian tube. A silastic sheet placed in the middle ear.

The patient was doing well initially after surgery with an intact TM and no further otorrhea at the 6 week postop visit. Two months after surgery, the patient was admitted to the hospital for a second trial of bone marrow transplantation for her neutropenia. The patient was found to have a recurrence of right otorrhea and an inflamed TM with a small posterior perforation. A revision tympanomastoidectomy was scheduled to rule out any recurrence. The patient was found to have significant granulation tissue in the middle ear around the silastic sheet. A small residual cholesteatoma pearl was identified at the anterior mesotympanum. Otherwise, the eustachian tube and mastoid were free from cholesteatoma. The silastic sheet was removed and the TM was repaired with a medial graft.

The patient's facial nerve remained intact throughout the course. An audiogram was unable to be done secondary to her frequent hospital admissions for bone marrow transplantation secondary to congenital neutropenia. The patient was lost to follow-up after her revision surgery.

Radiographs



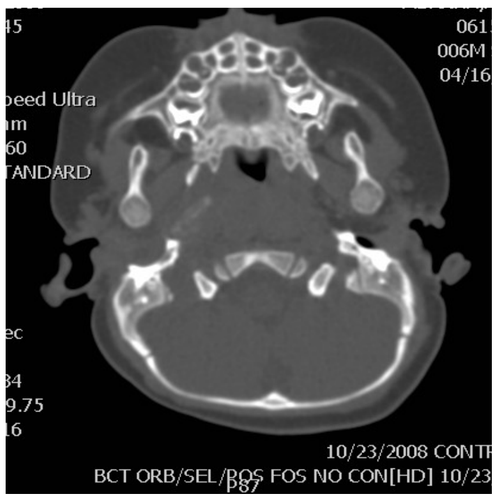


Fig 1. HRCT temporal bone without contrast showing a soft tissue mass completely filling the R middle ear with medial extension . There is reactive bone formation at sites of erosion including osseous portion of the EAC.

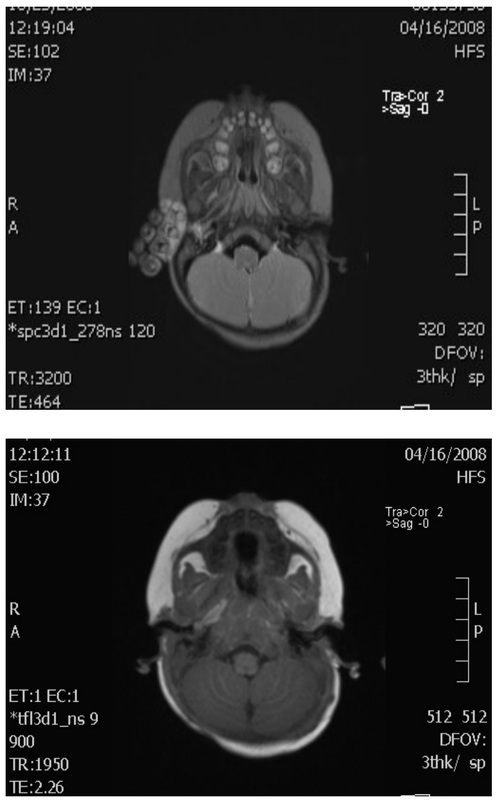


Fig 2. MRI T1 – hypointense T2 – hyperintense

Discussion

Otorrhea in an infant is not uncommon. The most common etiology is recurrent otitis media secondary to eustachian tube dysfunction. Other uncommon causes include histiocytosis, tuberculosis, acquired cholesteatoma and advanced congenital cholesteatoma.

The diagnosis of congenital cholesteatoma for this case was delayed because of the clinical presentation as chronic otorrhea and TM perforation, which would traditionally rule out CC. We consider this case to be CC, instead of acquired cholesteatoma, based on the following facts: 1) 90% of cholesteatoma content was identified inside the eustachian tube with only 10% limited to the mesotympanum just adjacent to the eustachian tube; The progression of middle ear cholesteatoma usually takes the pathway of least resistance. We would expect the mesotympanum to be full of cholesteatoma if the lesion had initiated in the middle ear such as in cholesteatoma secondary to TM perforation, prior to eustachian tube involvement; 2) significantly enlarged eustachian tube space, double the size of the middle ear, would suggest a chronic process of disease, including remodeling of the eustachian tube during pre-partum development; 3)The patient's symptoms started as early as one month after birth, which would not give sufficient time for development of such a massive cholesteatoma inside eustachian tube with such a significant change in eustachian tube size.

The imaging studies play a significant role in the diagnosis and management of this case. Initial CT scan of temporal bone helped to evaluate the middle ear/mastoid condition and pathology around the temporal bone. The soft tissue mass at the eustachian tube area was not initially identified by the radiologist who reviewed the case. It cannot be over emphasized that the surgeon should review all images on their own with attention to the surrounding tissue as well as the temporal bone. Although an MRI of the skull base with and without gadolinium did not help to reveal the final diagnosis of the mass, it helped to evaluate extension of the mass and association of adjacent structures, like the carotid. This relationship between the eustachian tube mass and major vessels is important prior to surgery because of the limited exposure of the eustachian tube during a middle ear exploration. It would be very difficult to control bleeding intraoperatively if it happened to be a vascular lesion.

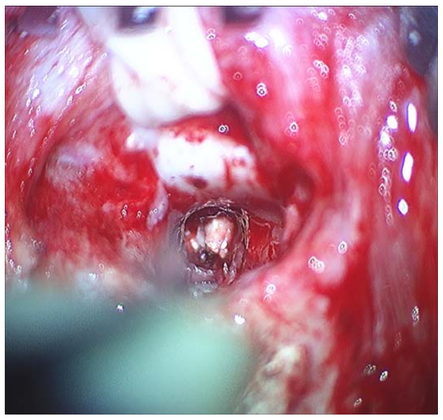
The principle in management of eustachian tube CC should be similar to cholesteatoma at other sites. Surgical management with complete removal of squamous epithelium is the main treatment. With such extensive disease, a second look at 12 months after surgery should be considered to rule out any recurrence or residual cholesteatoma. Our case was complicated with congenital neutropenia, which could have contributed to why the patient had a recurrence of her ear infection. Although a small residual cholesteatoma pearl was found during the revision surgery, it was very small and retained as an intact mass that would make it unlikely to trigger a major infection.

Another possible etiology for this patient's recurrence of otitis media is the possibility of acid reflux through the pathologically enlarged eustachian tube. We did consider eustachian tube occlusion during the first surgery to avoid acid reflux, but it was not done because of the following concerns: 1) it may cause difficulty in identification of a cholesteatoma recurrence in the eustachian tube during the second look; 2) the nasopharyngeal portion of the eustachian tube appeared normal, suggesting a certain degree of function of eustachian tube might be retained. If a patient with eustachian tube cholesteatoma continues to have recurrent otitis media with no evidence of recurrence or residual disease, acid reflux should be considered and middle ear fluid pH should be investigated. An alternative would be eustachian tube occlusion with pressure equalization tube placement to improve the outcome if the acid reflux into middle ear is confirmed.

The patient's audiologic function was never evaluated during the course of treatment. The audiology appointments were canceled several times because of frequent hospitalizations and for bone marrow transplantation. No intraoperative ABR was done since the patient was seen as an in-patient and both ear surgeries were performed on weekends, when ABR service was not available. Clinical observation suggested that the patient had at least a good hearing on the contralateral side.

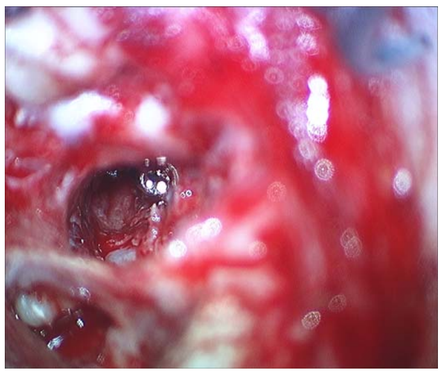
The epidermoid formation theory may help explain the pathogenesis of this case. The theory is based on the existence of ectodermal tissue in the first pharyngeal cleft of the fetus during development of the external auditory canal. The theory holds that this formation persists postpartum to become the precursor to CC.[4] There is evidence to support epidermoid formation as the pathogenic mechanism for CC formation. Liang et al.[12] identified varying numbers of CK14 immuno-positive, novel cell rests in the posterosuperior, posteroinferior, and anteroinferior regions of the middle ear. There was also further confirmation of four epidermal rests in a survey of 30 post-natal human temporal bones[14] and independent discoveries of keratinizing epidermoid formations considered to be intermediate forms of CC12 and 13 which were the missing links in the progression from the epidermoid formation to the CC. The eustachian tube lumen develops in the embryo by the lateral extension of the endoderm of the first pharyngeal pouch as it touches the inner surface of the ectoderm of the first branchial cleft. It is possible that some ectodermal tissue in the first branchial cleft was embedded during this fusion process and it slowly expanded during the development of the fetus, with expansion of the eustachian tube secondary to this mass effect. The cholesteatoma eventually expanded into the mesotympanum adjacent to the eustachian tube; subsequently the patient was born with eustachian tube dysfunction secondary to obstruction by cholesteatoma, causing middle ear infection which was followed with TM perforation and otorrhea.

Operative Photographs



Surgical Findings

1. Granulation in the medial EAC with no remnant TM
2. Cholesteatoma in the A-I quadrant
3. ET completely impacted and dilated with large cholesteatoma mass
4. Subtotal ossicular erosion with intact stapes superstructure
5. Granulation in the epi/hypotympanum, facial recess, antrum
6. Well pneumatized mastoid



Post-Surgical Findings

1. Complete cholesteatoma extirpation – confirmation by retrograde endoscopy
2. Silastic placed into middle ear
3. Medial Graft Tympanoplasty
4. Split thickness skin graft to anterior bony EAC defect

Conclusion

We present a first case of congenital cholesteatoma of the eustachian tube with extension into the middle ear in a 4 month old infant. The unique clinical presentation as chronic otorrhea and TM perforation challenges the traditional definition of middle ear congenital cholesteatoma. Imaging studies prior to surgery are essential for differential diagnosis of a eustachian tube mass and chronic otorrhea. While the principle of treatment is the same as for cholesteatoma in the middle ear, acid reflux into the middle ear should also be considered as a potential sequelae from a pathologically enlarged Eustachian tube causing a recurrence of ear symptoms.

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