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Case Report

Atypical Presentation of Congenital Cholesteatoma in an Adult Case

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ABSTRACT

Cholesteatoma congenita is a recognized anatomoclinical entity. Adult forms are rare, with only a handful of documented cases. Making the diagnosis of congenital cholesteatoma in adult patients still pose a challenge even among experienced otologists. The etiopathogenesis of this lesion is still unknown; however, when cholesteatoma develops in subjects with no history of ear inflammation, as in the case we report here, an embryological origin is strongly suspected. An acquired origin is assumed in patients with a history of inflammatory processes in the outer or middle ear, due to proliferation of the basal cell layer of the tympanic membrane epithelium.it can occur in atypical places and at atypical ages. Many authors prefer tympanomastoidectomy of the canal wall. But it can also be successfully treated by intact canal wall tympanomastoidectomy, with good hearing results. We report a rare case of congenital cholesteatoma in an adult patient.

Keywords: Congenital cholesteatoma; Adult; Atypical presentation; Conductive hearing loss; Mastoidectomy

Introduction

The first definition of congenital cholesteatoma with criteria to distinguish it from acquired cholesteatoma was proposed by Derlacki and Clemis[1] in 1965 and completed by Levenson et al. in 1989[2]. They proposed the following criteria:

- The existence of a white mass on the medial aspect of a normal eardrum[1,2];
- A normal pars flaccida and pars tensa[1,2];
- Absence of antecedent otorrhea or perforation[2];
- No history of myringotomy or middle ear surgery[2];
- Exclusion of intratympanic or giant cholesteatoma[2];
- Antecedent otitis media is no longer an exclusion criterion[2].

Congenital cholesteatoma (CC) is thought to be caused by inadequate folding of the epidermoid formation inside the middle ear cleft. Stratified squamous epithelium increases during the third and fifth weeks of embryonic life during the development of the middle ear mucosa. Cholesteatomas generally arise in childhood due to the sluggish growth of squamous cells that stay in the middle ear cavity from this time[3]. It usually appears as a "pearl" under the tympanic membrane in the anterosuperior quadrant. The prognosis for congenital cholesteatoma-induced conductive hearing loss is considered to be poor[4].

It may be discovered incidentally by otoscopy. The typical appearance is that of a whitish retrotympanic mass. Its location is variable, with, in order of frequency, the anterosuperior mesotympanum (65%), the posterosuperior meso tympanum (15%), the two upper quadrants (17%) and the anteroinferior

mesotympanum (2-5%)[2].The otoscopic appearance may also be strictly normal[5]. It can also be diagnosed by aspiration of epidermal debris during paracentesis for OSM or suspected in the presence of conductive hearing loss or unilateral seromucous otitis. The persistence of conductive hearing loss in a child after placement of a tympanic should raise the possibility of congenital cholesteatoma. Facial paralysis (9%)[5] and rarely vertigo or tinnitus may complete the clinical picture. Many reports of pediatric cholesteatomas exist but studies in adult population are relatively less. We report one description in adults.

Case Report

A 41-year-old lady presented to our Ear, Nose and Throat (ENT) department with progressively worsening right hearing loss and chronic right otalgia, patient without any particular pathological history. There was no history of otitis media, ear trauma or otologic surgery. The history of her illness goes back to 3 years with the installation of o-progressive right hypoacusis with intermittent rotatory vertigo and intermittent headaches without other associated signs in particular no facial palsy no tinnitus. The evolution was marked by the appearance of intense and intermittent chronic homolateral otalgia. In the examination, normal otoscopy **(Figure 1)**, there was no facial palsy, no spontaneous nystagmus.

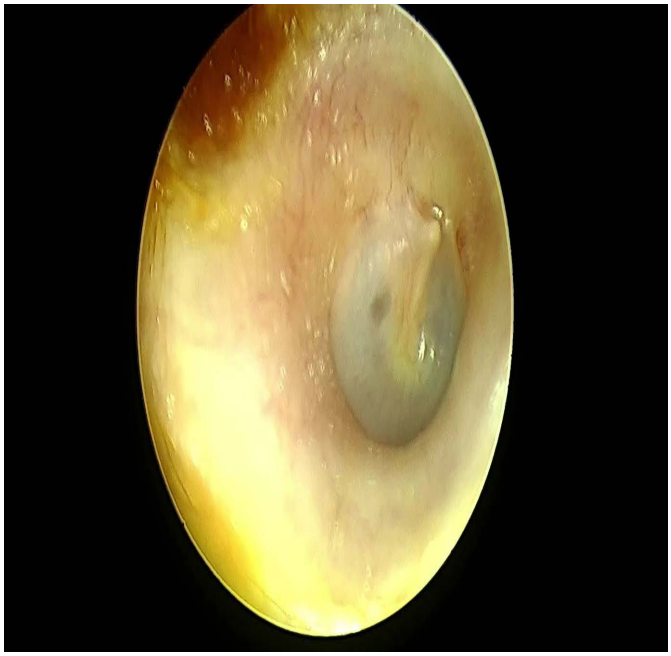

Figure 1: Otoscopy of the Right Ear.

The audiogram showed the conductive hearing loss on the right ear with a loss of 70 db and Weber was lateralized to the right (**Figure 2**). The tympanogram curve is flat on the right ear. The video-head impulse test schowed a right lateral deficit (**Figure 3**).

On the CT scan of the temporal bone, the right ear cavity had a soft tissue density, incomplete filling of the epitympanum and mesotympanum, by well-defined, encapsulated tissue formations, one of which is pedicled opposite the Jacobson's nerve, thining of the tegmen tympani, ossicular lysis, fistula between the upper semicircular canal and the tympanic cavity and mastoid cell filling (**Figure 4**).

In the MR imaging of the temporal bone schowed abnormal signal intensity within the right middle ear cavity which appeared hypointense on T1-weighted, hyperintense on T2-weighted images with minimal peripheral enhancement post-gadolinium, coincide with the diagnosis of cholesteatoma. Mastoid cell filling are also filled in, with loss of the annular appearance of the right semicircular canal (**Figure 5**).

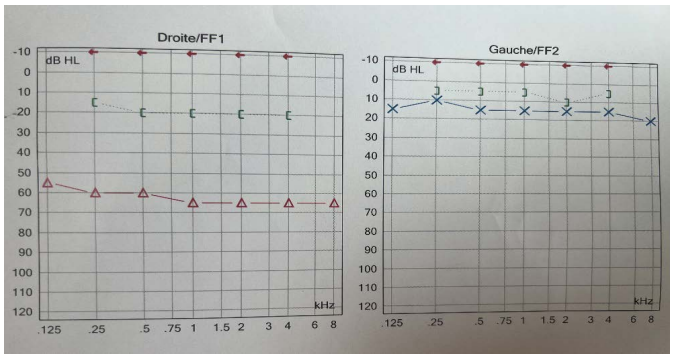

Figure 2: Right ear showed a moderate conductive hearing loss with air-bone gap of 45 dBH.

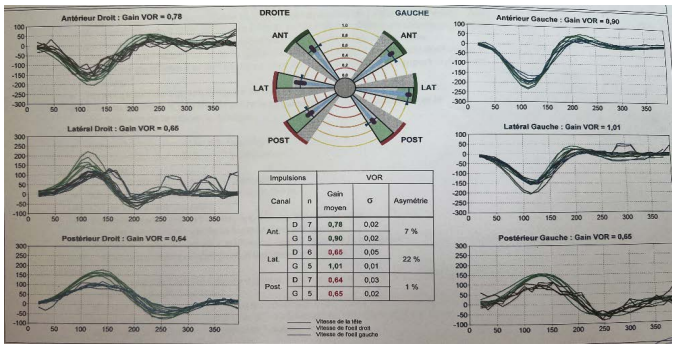

Figure 3: The video-head impulse test schowed a right lateral deficit.

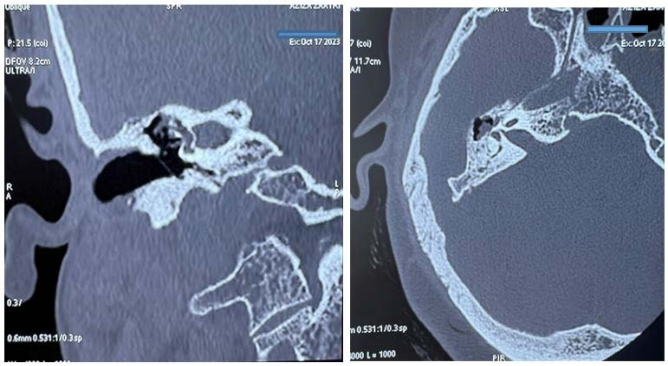

Figure 4: CT scan of the temporal bone in coronal and axial cuts: showing soft tissue density within the right middle ear cavity, pedicle tissue formation opposite the jacobson nerve, ossicular lysis, fistula between the upper semicircular canal and the tympanic cavity.

The patient underwent middle ear exploration, which confirmed the presence of a congenital cholesteatoma. The lesion was removed using a retroauricolar approach, following which a miringoplasty (underlay techinique) was performed using an autologous temporalis muscle fascia graft). Intra- or postoperative complication was seen in the patient. The diagnosis of cholesteatoma was confirmed histologically. The pure-tone average thresholds obtained at the 6th week of the operation were 10 dB (air/bone thresholds respectively). Follow-up 3 months after surgery revealed no signs of recurrence.

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

All diagnostic and therapeutic procedures were performed with patient's consent and respecting ethical principles of our institutional and national committee.

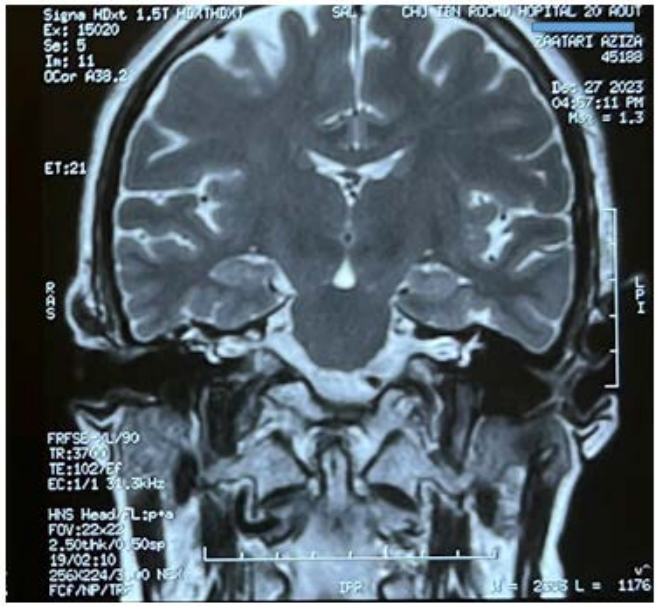

Figure 5: MR imaging of the temporal bone schowed a loss of the annular appearance of the right semicircular canal.

Discussion

Congenital cholesteatoma (CC) can appear at any age from infancy to adulthood (mean 16.7 years of age) and it is caused by congenital epithelial remains in the temporal bone[6]. Its histological appearance is comparable to acquired cholesteatoma; nevertheless, some characteristics of the patient, such as the absence of ear illness or surgical history, support the diagnosis of CC[7].

Otorrhea, the clinical sign of acquired cholesteatoma in children, is absent in congenital cholesteatoma[5]. Location is not a diagnostic criterion, but according to Duclos et al[5], Potsic et al[8] and Zappia and Wiet[9], two clinical forms of cholesteatoma can be described cholesteatoma: localized anterosuperior encapsulated forms, asymptomatic and without hearing loss; diffuse forms with invasion of the posterosuperior quadrant or mesotympanum, frequently associated with ossicular erosion. Potsic et al point out that the finding of two different clinical forms is correlated with two population groups of different ages[8]. Encapsulated anterosuperior forms are more frequent in young children, whereas diffuse forms with invasion of the posterosuperior quadrant or mesotympanum are found in a wider population of children of varying ages[8]. Conductive hearing loss is an inevitable consequence of CC involving the posterior tympanum[10]. In the adults and elderly, the disease may involve an unusual localization and conductive hearing loss is the most common symptom in these patients.

Congenital cholesteatoma in adults is rare, but there are a few descriptions:

- Zappia and Wiet report a case of congenital cholesteatoma in a 30-year-old man.A case of posterosuperior congenital cholesteatoma with erosion of the incus and head of the malleus has been reported[9].
- Mc donald et al describe five cases in patients aged 22 to 59 with conductive hypoacusis, intact tympanic membrane, facial or dural dehiscence, labyrinthine fistulae or multiple localizations[10].
- Mendoc¸a Cruz et al report a case of congenital cholesteatoma in a 26-year-old woman presenting clinically with hypoacusis and associated facial palsy[11].
- Galli et al. report a case of congenital cholesteatoma in a 34-year-old man, revealed by temporomandibular joint pain[10].
- Soderberg and Dornhoffer describe a case of congenital cholesteatoma in a 24-year-old woman with 55 dB of hearing loss, which developed progressively over several years[12].
- With the exception of the case described by Zappia et al[11], adult forms are extensive and late-onset. In these cases, there is always labyrinthine involvement, facial paralysis and diffuse involvement of the middle ear or even the temporomandibular joint.

Most descriptions in adults are symptomatic of a diffuse, progressive form and the absence of otorrhea, perforation, myringotomy or middle-ear surgery, together with an intact eardrum, allow the diagnosis to be made, but the encapsulated form in the posterior-superior quadrant is not described.

More severe consequences such labyrinthine fistula, facial paralysis, meningitis, cranial abscess and even death could result from CC if it is discovered too late[13].

Imaging is crucial to the diagnosing process, particularly when it comes to differentiating between lesions that impact the mastoid and the petrous apex. In magnetic resonance imaging (MRI), CC have low signal on T1 and high signal on T2, they do not augment with contrast and they exhibit restricted diffusion on diffusion weighted imaging (DWI) sequences[16]. Computerized tomography (CT) displays expansile well circumscribed mass. Another function of DWI is to detect recurrent or residual disease, particularly in situations where the canal wall is preserved during surgery or when cartilage is utilized to reconstruct the tympanic membrane and direct otoscopic visualization is insufficient to determine the condition. As a result, DWI may prevent needless second opinion procedures[17].

Congenital cholesteatoma is a condition primarily affecting children. 73% of patients who underwent surgery over a 24-year period were 15 years of age or younger, according to a sizable retrospective study[14]. The disease is generally in a more advanced stage when a patient is diagnosed, generally speaking[15]. When our case was diagnosed and operated on, he was 35 years old.

The unusual condition known as congenital cholesteatoma of the middle ear is caused by squamous epithelial cells in the tympanic cavity that were left over from embryonic development. Despite being innocuous at first, it worsens over time and can lead to potentially fatal complications. The prognosis for middle ear anatomy-related hearing loss is typically poor. These factors make early diagnosis and treatment of this illness crucial.

Recurrence rate and hearing improvement after canal wall down or wall up mastoidectomy have not been reported different in the literature in cases with CC[18,19].

Conclusion

To sum up, CC is capable of a variety of presentations. A successful surgical outcome is the complete eradication of the

disease. Surgery must, however, be customized to the precise location and severity of each patient's illness.

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