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Congenital Cholesteatoma

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Cholesteatoma means the presence of exfoliated keratin in any pneumatic space of the temporal bone originating from keratinizing stratified squamous epithelium (Schuknecht, 1993). It is defined as either acquired or congenital. Acquired cholesteatoma can be primary, when perforation is confined to the region of the *pars flaccida*, or secondary, when perforation is usually marginal and located in the *pars tensa* of the tympanic membrane.

Congenital cholesteatoma is a rare type of keratoma that originates from the same ectoderm that forms the primitive notochord. Embryonic cell-rests from this ectodermal structure may occur in any of the cranial bones. When they occur in the petrous portion of the temporal bone they may spread in and around the labyrinth and extend into the middle ear, mastoid, or cranial cavity (Paparella *et al.* , 1991). Possible sites of origin within the temporal bone include the middle ear, petrous apex adjacent to the facial canal, external auditory canal, and mastoid. Congenital cholesteatoma may be intradural and may present as a mass in the cerebellopontine angle. The middle ear is the most prevalent site, and a mastoid primary is the rarest (Thakkar *et al.* , 2006).

The classic presentation of congenital cholesteatoma is as a pearl behind an intact tympanic membrane in the anterior-superior quadrant, and most of the time there is no history of otorrhea, perforation, or previous otologic procedure. A history of otitis media or effusion does not exclude a case from being considered as congenital cholesteatoma (Kazahaya and Potsic, 2004, Warren *et al.* , 2007). Cushing in 1922 expressed the opinion that it was not improbable in many of his recorded cases that the keratoma itself had been responsible for the otitis media rather than the reverse.

The incidence of cholesteatoma in childhood is estimated to be three to six per 100,000 people. Nearly 30 % of pediatric cholesteatomas are congenital (Kazahaya and Potsic, 2004, Shirazi *et al.* , 2006). These keratomas usually lie dormant for years. If they become infected, intratemporal complications such as facial palsy (usually the first sign that appears), mastoiditis, acute labyrinthitis or petrositis may occur quickly. Intracranial complications can also occur very fast, such as meningitis, encephalitis, cerebral abscess, hydrocephalus oticus, sigmoid sinus thrombosis, and subarachnoid and subdural abscess (Paparella *et al.* , 1991).

As a result of the prevalence in children of chronic serous otitis media, rapid

growth of tissue, and dysfunction of the Eustachian tube, cholesteatomas in children may have a more aggressive pattern of growth relative to that in adults. The incidence of both residual and recurrent disease is higher in children compared to that in adults (Shirazi *et al.* , 2006). Therefore, the complications of this disease in childhood must be diagnosed as soon as possible, and the etiologic diagnosis established as well. In cases of intratemporal or intracranial complications of congenital cholesteatoma, clinical endovenous treatment alone does not solve the problem. The patient may die. Early surgical intervention is mandatory to save the life of the patient.

The number of reported cases of congenital cholesteatoma has increased considerably in recent decades probably because of heightened awareness leading to timely diagnoses, improved otoscopes and microscopes, and refined radiologic examination of the temporal bone. Recognition of the disease by otolaryngologists, pediatricians, and neurologists plays an important role in earlier diagnosis of and intervention in this disease (Potsic *et al.* , 2002).

Cholesteatoma needs to be distinguished from cholesterol granuloma and from anomalous sigmoid sinus or neoplasms. Cholesterol granulomas histologically consist of crystals of cholesterol formed by degradation of components of the blood and can be differentiated by their bright signal-intensity on both T1- and T2 weighted images. Cholesteatomas do not produce increased signals when using gadolinium on an MRI, unlike meningiomas, schwannomas, or metastatic lesions (Thakkar *et al.* , 2006).

Taking into account the severity of complications of congenital cholesteatoma, its morbidity/mortality, and its variable and nonspecific clinical presentation (the most common presentation being an incidental finding), we decided to establish a protocol for early identification of this threatening disease. In our clinical practice we order computed tomography of the temporal bone every time a child presents one of the following circumstances. The possibility of having a congenital cholesteatoma decreases from first to last item listed.

- A whitish mass behind an intact and normal tympanic membrane
- Facial palsy
- Signs of mastoiditis (postauricular subperiosteal abscess)
- History of episodes of fetid otorrhea, and an intact tympanic membrane at the time of examination
- Unilateral conductive or mixed hearing loss
- Persistent discharge from the ear after insertion of ventilation tube
- Persistent effusion in the middle ear
- Diagnosis of meningitis
- Vertigo and imbalance
- Sudden deafness
- Diagnosis of intracranial abscesses
- Headache and fever of undetermined origin
- Ear or neck pain of undetermined origin

Throughout the radiologic evaluation of the temporal bone (CT scan), we look for the presence of a mass in the middle ear or mastoid (hypodense expansive lesion), destruction of the ossicular chain, exposure of the *tegmen tympani* dural plate, sigmoid sinus, or fallopian canal, and erosion into the semicircular canals. An MRI may also be useful in ruling out intracranial complications (dural involvement, brain herniation, sigmoid sinus thrombosis, intracranial abscesses and so forth).

This protocol was suggested on the basis of a retrospective study in which we evaluated our series of congenital cholesteatomas in the middle ear/mastoid comprising 37 children who underwent surgery to remove the disease. Information regarding clinical presentation and temporal images from computed tomography were recorded from their charts. Data from the literature also were considered to produce this protocol.

Children with congenital cholesteatoma have abnormal vestibular anatomy, such as a larger endolymphatic fossa and vestibular aqueduct, and there was a trend toward the vestibule of the semicircular canal being smaller as compared with that in controls (Propst *et al.* , 2008). We can wonder if these malformations facilitate the intracranial complications.

The treatment of choice for congenital cholesteatoma is surgical. The otologist has to choose among three main surgical procedures for resection of the disease. These include tympanoplasty, canal-wall-up tympanomastoidectomy (closed cavity technique), and canal-wall- down tympanomastoidectomy (open cavity technique) (Kazahaya and Potsic, 2004, Shirazi *et al.* , 2006). The great dilemma lies in deciding between a closed or an open cavity technique. The main goals to be reached with surgery are: complete eradication of the disease, preservation and optimization of hearing results, and a trouble-free ear. It is advisable to monitor the facial nerve intraoperatively. Early treatment decreases the extent of the disease and reduces the risk of recurrence and complications.

Closed cavity tympanomastoidectomy has been recommended as the technique of choice in children, but each case should be evaluated separately and the appropriate technique tailored to the individual patient. When dealing with a case of more extensive disease, very sclerotic mastoids, disease in an only hearing ear, the possibility of labyrinth fistula, or a case of recurrence of the disease, an open cavity technique may be a better choice. Regardless of the technique used to remove the cholesteatoma, the patient should receive long-term follow-up to monitor the ear for recurrent disease.

Recommended readings

1. Schuknecht HF. Pathology of the Ear. Philadelphia: Lea & Febiger; 1993. Paparella MM, Shumrick DA, Gluckman JL, Meyerhoff WL. Otolaryngology. 3rded. Philadelphia: W. B. Saunders; 1991.
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