

# Congenital Cholesteatoma: Delayed Diagnosis and its Consequences

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## SUMMARY

This is a retrospective review of congenital cholesteatoma cases that were managed surgically. There were 5 cases. The age of presentation ranged from 5 to 18 year old. Three patients presented with complication of the disease. Three patients had intact tympanic membrane, two had perforation at the anterior superior quadrant. All patients had cholesteatoma medial to tympanic membrane. Four cases had extensive ossicular erosion with preoperative hearing worse than 40 dB. Four cases underwent canal wall down mastoid surgery and one underwent canal wall up surgery. One patient had recurrence which required revision surgery. In conclusion, congenital cholesteatoma presented late due to the silent nature of disease in its early stage. Extensive disease, ossicular destruction with risk of complication at presentation were marked in our study. Hence, more aggressive surgical intervention is recommended in the management of congenital cholesteatoma.

## KEY WORDS:

*Cholesteatoma, Children, Congenital*

## INTRODUCTION

Congenital cholesteatoma of the middle ear was first described by Howard House in 1953. Later, Derlacki and Clemis in 1965 established the clinical criterias for the diagnosis which include a pearly white mass medial to an intact tympanic membrane, a normal pars tensa and flaccida, and no history of otorrhea, perforation, or previous otologic procedure. Levenson in 1986 then revised the criteria by adding that otorrhea and perforation should not be an exclusion criteria. Otitis media may stimulate the growth of congenital cholesteatoma. The pathogenesis of cholesteatoma growth is still poorly understood as evidenced by the multiple theories currently found in the literature. They include implantation, invagination or invasion, metaplasia and epithelial cell rest theories. The most accepted theory is the epithelial cell rest theory. This is based on Teed's observation of an epidermal structure found in 5 month fetus<sup>1</sup>. Failure to undergo involution at around 33 weeks explained the formation of congenital cholesteatoma.

The incidence of congenital cholesteatoma is estimated to be between 4% to 24% of all cholesteatomas in children<sup>2</sup>. The most common presentation of a congenital cholesteatoma is

an asymptomatic white retrotympanic ear mass<sup>3</sup> and they may be discovered incidentally during the time of routine otologic evaluation or during a myringotomy. Otagia and otorrhea are rare, but nearly 50% of patients describe episodes of previous otitis media<sup>4</sup>. The most common location is at the anterior superior quadrant compared to acquired type which is more common at the posterior superior quadrant of tympanic membrane. Although clinically it is possible to differentiate congenital from the acquired type, it is difficult to distinguish them in an advanced disease<sup>5</sup>.

The treatment for congenital cholesteatoma is surgery. The goal of surgery is complete removal of disease with restoration of hearing if possible. The options are tympanoplasty with intact canal wall or canal wall down.

## MATERIALS AND METHODS

A retrospective review of all cases of congenital cholesteatoma that were managed surgically were obtained from operating book records from July 1999 till December 2008. Clinical presentation, intraoperative disease extension and management outcome of the cases were reviewed. Pre and postoperative hearing level were determined using pure tone average at 500, 1000 and 2000 Hz.

## RESULTS

There were 5 patients (12.2%) identified as congenital cholesteatoma from a series of 41 cases of cholesteatoma in children. The age of presentation ranged from 5 to 18 year old. There was a male predominance in this series with 3 males and 2 females. All patients had whitish mass medial to tympanic membrane. Table I shows the clinical presentation of the cases of congenital cholesteatoma. Two cases presented with perforated anterosupero quadrant of the tympanic membrane. Two patients had facial nerve palsy and one had labyrinthine fistula. Majority of patients had at least moderate hearing loss. Table II shows the disease extension, status of ossicles and type of surgery performed. Four cases underwent canal wall down surgery in view of extensive disease. Figure 1 shows an extensive congenital cholesteatoma eroding tegmen tympani and medial wall of cochlea seen on High Resolution CT of temporal bone. According to Potsic staging system, 4 patients had stage 4 disease and 1 had stage 3. Duration of post operative follow up for these patients were between 6 months to 6 years with

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**Table I: Clinical features of congenital cholesteatoma**

Cases	Clinical presentation	Status of tympanic membrane	Pre operative hearing level
1	Incidental finding	Perforation at anterior superior quadrant	Moderate HL
2	Facial asymmetry, hearing loss	Intact	Moderate conductive HL
3	Vertigo, hearing loss	Intact	Moderate mixed HL
4	otorrhoea	Perforation at anterior superior quadrant	Mild conductive HL
5	Facial asymmetry, hearing loss	Intact	Profound HL

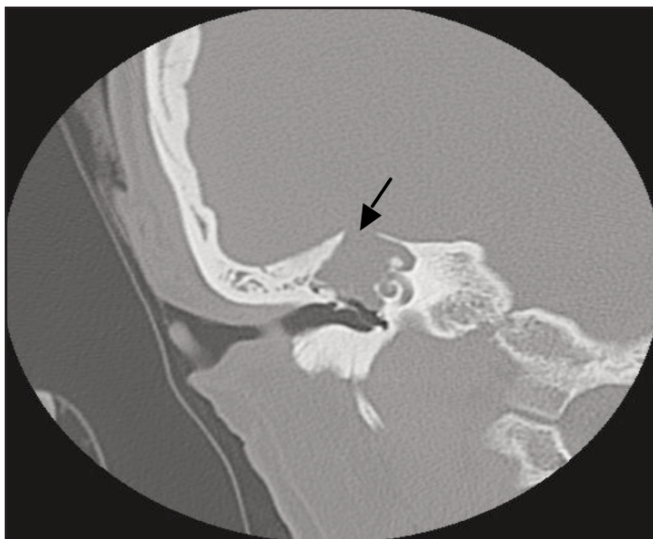
HL- hearing loss

**Table II: Type of surgery with intra operative finding**

Cases	Disease extension	Status of ossicular erosion	Type of surgery
1	Attic, middle ear	Incus, stapes crura	CWD + Tympanoplasty III
2	Attic, middle ear, mastoid, petrous apex. Facial nerve dehiscence	Maleus, incus, stapes suprastructures	CWD
3	Middle ear, mastoid. Lateral Semicircular Canal and tegmen eroded	Long process incus	CWD
4	Attic, middle ear, mastoid	incus	CWD
5	Attic, retrolabyrinthine. Tegmen eroded (Figure 1)	Stapes suprastructure	CWU

CWD- canal wall down

CWU- canal wall up



**Fig. 1:** Coronal view of HRCT temporal showing cholesteatoma at the epitympanum and mesotympanum with erosion of tegmen and medial wall of cochlea

mean of 46 months. Four patients had post operative hearing assessment which showed worse hearing in 3 cases and preserved hearing in 1 case. One patient developed recurrence disease in the petrous apex which required revision surgery via translabyrinthine approach.

**DISCUSSION**

The incidence of congenital cholesteatoma in our study is 12.2% (5/41 cases) which is consistent with the literature. There is also male preponderance in our study which is also similarly reported by Potsic *et al*<sup>6</sup>. The classical characteristics of congenital cholesteatoma as described by Levenson were not common in our review and most of patients had delayed presentation with hearing loss and complications. This is

probably due to asymptomatic disease in the middle ear in the early stage of disease.

There is a high proportion of patients presented with complication of disease in our reviews with two facial nerve palsy and one labyrinthine fistula. Darrouzet *et al* reported facial nerve palsy and labyrinthine fistula in congenital cholesteatoma are much more common than in acquired form. However, there is a lower incidence of intracranial complications with congenital cholesteatomas than the acquired type. An early diagnosis with improvement in health care and preventive medicine in children would decrease the rate of complication. The incidental finding during myringotomy for middle ear effusion for one patient in our study is not uncommon. Darrouzet *et al* and many studies report that pseudo-otitis media with effusion is the typical main finding at otoscopy, with myringotomy leading to diagnosis<sup>7</sup>.

Majority of our cases had intact tympanic membrane with whitish mass medial to it on otoscopic examination. The presence of perforation in two patients in our series can be considered as being congenital origin. Tympanic membrane perforation occurred in numerous cases and may be induced by cholesteatoma that initially develop in the middle ear. Koltai *et al* reported clinical evidence to suggest that it is reasonable for a sufficient growth of the congenital cholesteatoma to cause tympanic membrane perforation in a nontraditional location<sup>8</sup>. The traditional location of perforation for acquired form of cholesteatoma will be at the pars flaccid or the posterosuperior quadrant of pars tensa.

We notice that the pre operative pure tone average in this study correlate with age of patient; the younger the patient presented the better the hearing level. The oldest patient (18 year old) had profound hearing loss on presentation. This is probably due to the fact the older children had disease that had existed longer with a higher chance of developing complication.

The growth of congenital cholesteatoma follows a natural course as described by Koltai. It is believed that it started at the anterior superior quadrant of the tympanic membrane and continue growing in a posterior direction of spread to involve the incudostapedial joint or stapes superstructure, but usually spares the footplate. Continued expansion proceeds toward the facial recess, sinus tympani, and eventually the mastoid air cells. Majority of our patients had disease in posterior mesotympanum with erosion of the ossicles. The finding of labyrinthine fistula and facial nerve involvement intraoperatively was reported to be more common compared to acquired lesions.

In many series<sup>7,9,10</sup> canal wall up is recommended in view of normal or subnormal Eustachian tube function, well-developed temporal bone pneumatization, incidence of cystic localized lesions, and normal appearance of the middle ear mucosa. The need for canal wall down procedure is rare and should be considered when there is only labyrinthine involvement or concern about the reliability of follow-up. However, in our series canal wall down surgery was performed in most of the cases due to extensive disease with high complication at presentation.

Serial audiograms in children is difficult because they are prone to inconsistencies in their audiograms. The postoperative hearing was not possible to be improved in our review. However, our number of patient is limited and furthermore patients presented with at least moderate hearing loss in majority of cases.

Recurrence rates of congenital cholesteatoma should be lower than acquired forms of the disease since the pathophysiology does not involve Eustachian tube dysfunction. Bennet *et al* did not experience recurrence in the 32 patients with long-term follow-up. Because of low recurrence rates, he did not routinely stage his patients and only re explore them for obvious recurrence, worsening or poor hearing<sup>9</sup>. However, other series reported with highly variable recurrence rate

range from 8%-81% depending on the sample size, type of surgery performed and size of the lesion. Darrouzet *et al* reported the overall recurrence rate in his series was 29.2% which was considered satisfactory assuming the lesions were relatively extensive. In our review with limited number of cases, only 1 case had recurrence disease. Recurrence occurred in the case of petrous apex involvement during the first surgery which implies that very extensive disease is more likely to develop recurrence.

**CONCLUSION**

In our series, the classical presentation of congenital cholesteatoma is rare with most cases had delayed presentation with hearing loss and consequences. Hence, more aggressive treatment i.e. canal wall down surgery is recommended due to extensive disease.

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