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Congenital cholesteatoma

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Abstract

We present a British series of eleven patients with primary cholesteatoma, including one patient with bilateral disease. Eight children presented with a history of hearing loss, while one child had had recurrent otitis media and another had had earache. Operative findings were: in five ears, cholesteatoma confined to the antero-superior segment with intact ossicles, in a further four, cholesteatomas extending throughout the mesotympanum with ossicular erosion in one, and in two ears posterior disease throughout the middle ear and mastoid, which had eroded the ossicles in both cases. The five cases of antero-superior cholesteatoma lend most support to Michaels' concept of epidermoid formation as a possible source of congenital cholesteatoma. With a greater awareness of the problem and careful examination of the antero-superior quadrant of the tympanic membrane, earlier diagnosis may be possible enabling removal of small intact cholesteatoma sacs and preserving the structures of the middle ear and therefore the hearing.

A screening programme for infants included as part of their routine examination which would be undertaken by examiners who are trained to be more aware of the problem and skilled at otoscopy, would help in the earlier detection of such cases as is shown by reports from the U.S.A.

Introduction

Congenital or primary cholesteatoma is a cholesteatoma of the middle ear, behind an intact tympanic membrane where there is no history of instrumentation or disease of the pars flaccida. It is an uncommon condition.

Historically, in 1829, Cruveilhier described multicentric white 'Tumeurs perlées'. In 1838, Mueller first used the term 'cholesteatoma' (Peron and Shuknecht, 1975), to describe intracranial deposits of keratin and in 1922 Cushing speculated upon the possibility of a congenital origin of cholesteatoma of the ear—a hypothesis put forward by Korner in 1830. However, the classic description of a cholesteatoma of the middle ear behind an intact tympanic membrane was first reported as recently as 1953 by House.

Since then cases have been described with increasing frequency, (McDonald *et al.*, 1984; Levenson *et al.*, 1986; Cohen, 1987). Bilateral congenital cholesteatoma is rare (Wingert *et al.*, 1976; Curtis, 1979; Wang *et al.*, 1984). Most of the recent reports are from the United States.

In this paper, eleven cases are presented from four centres in England. In five cases, the cholesteatoma presented in the anterior superior part of the middle ear, supporting the theory of an epidermoid cell rest forming the basis of the condition, as described by Michaels (1986, 1988a,b).

Pathogenesis

Many theories have been put forward as to the origin of primary cholesteatoma.

They include the following:

1. Squamous epithelium from the external auditory meatus enters the middle ear by invasion through a pre-existing marginal perforation or retraction pocket (Ruedi, 1959).
2. Squamous metaplasia of the middle ear (Sadé *et al.*, 1983).
3. Laterally located epidermoid tumours arising from ectodermal implants in the fusion planes of the first and second branchial arches (Pararella and Rybah, 1978).
4. It has been suggested that the embroonic tympanic ring functions to control ingrowth of the canal wall skin epithelium into the middle ear cleft, before the development of the connective tissue annulus (Aimi, 1983).
5. Amniotic fluid squamous debris developing into primary cholesteatoma (Piza *et al.*, 1985).

None of these theories so far have been convincingly proven. Michaels (1986, 1988b) recently discovered cell rest—epidermoid formation, which is seen in many fetal ears at the junction of the Eustachian tube with the middle ear near the anterior limb of the tympanic ring at the exact point of epithelial transformation from the

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TABLE I

CASE REPORTS. CLINICAL DETAILS AND SITES OF CASES OF CONGENITAL CHOLESTEATOMA N = 12 EARS (11 PATIENTS)

Case no.	Age	500/2 kHz hearing loss	Otitis media	Otoscopy	Glue ear	Site of cholesteatoma: Antero-superior	Meso-tympanic	Post-erior	Mastoid	Ossicular erosion
1	9	40 dB+	–	Featureless tympanic membrane, white behind	–		+			+
2	6	50 dB+	–	Full tympanic membrane, white behind	–	+	+	+	+	+
3	5	30/40 dB+	+	White bulging tympanic membrane	+	+	+			+
4	5	30/60 dB+	–	Opaque and congested antero-superiorly	–	+	+			+
5R	4	20 dB+	–	White cyst anteriorly and secretory otitis media	+	+				–
5L	4	80/30dB+	–	Opaque tympanic membrane	–	+	+			+
6	7	60 dB+	–	white posteriorly not in contact with tympanic membrane	–			+		+
7	2	–	–	Opaque antero-superiorly	–	+				–
8	8	30 dB+	–	Retracted tympanic membrane, white mass posteriorly	–			+		–
9	7	40/20 dB	–	Discrete white area antero-superiorly	+	+				–
10	3	30 dB+	–	Discrete white area antero-superiorly	–	+				–
11	6	30 dB	–	White antero-superiorly	+	+				–

tympanic cavity to the Eustachian tube. It disappears at 33 weeks gestation in the normal fetus, but it may persist and have the potential to become a primary cholesteatoma. Recent studies by Levenson *et al.*, (1986) and Cohen (1987) show that two-thirds of primary cholesteatomas originate from the anterior superior quadrant of the middle ear. This report adds further support to Michaels' theory of epidermoid formation as to its likely origin.

Summary of case histories

Of the eleven children, aged two to nine with a mean age of six, all but two presented with hearing loss. The first exception, an eight-year-old girl, was referred for tonsillectomy but on direct questioning there was a history of hearing loss which was then confirmed on examination and audiometry. The second, presented with earache but had no hearing loss (case no. 7, Table I).

Only one child had had recurrent otitis media. At operation, a middle ear effusion was found in four ears. Five children had cholesteatoma confined to the antero-

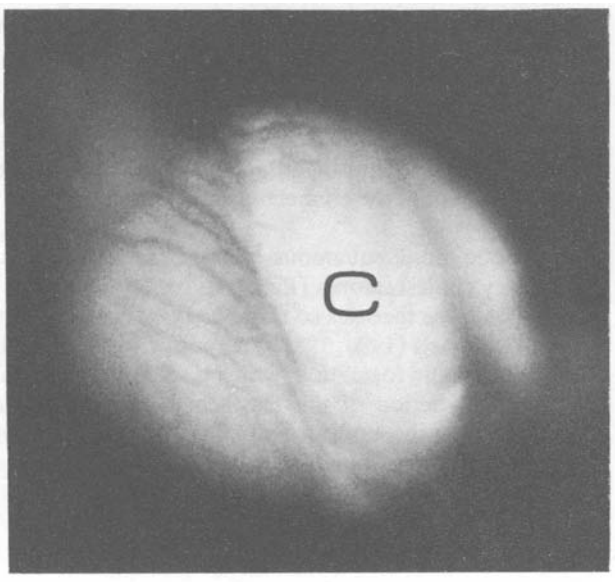

FIG. 1

Case 7: Congenital cholesteatoma (c) situated antero-superiorly in the middle ear.

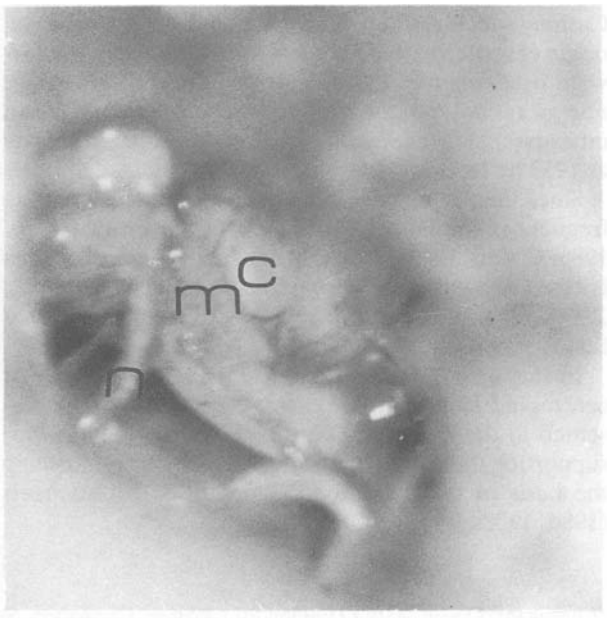

FIG. 2

Case 7: Operative appearances: congenital cholesteatoma (c) situated medial to the malleus handle (m). (n) = chorda tympani nerve.

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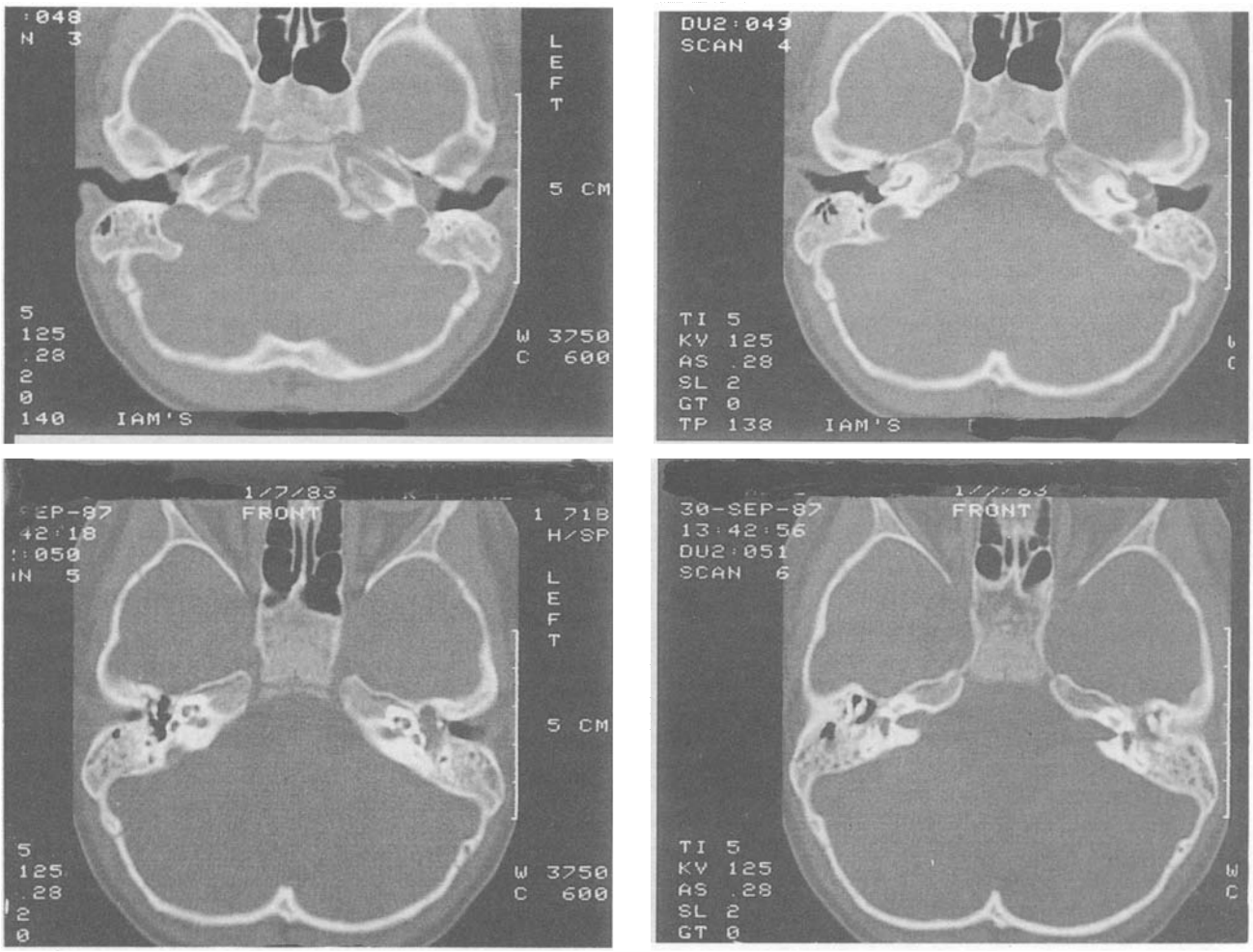

FIG. 3

Case 5: C.T. scan—the appearances are compatible with extensive cholesteatoma of the middle ear cleft with probable ossicular erosion on the left side, and a small localized cholesteatoma with no evidence of bone erosion on the right side.

superior segment (Figs. 1 & 2). Cholesteatoma was found bilaterally in one child (Fig. 3): on the right, there was a pearl in the antero-superior position, blocking the Eustachian tube opening with a middle ear effusion, and on the left it occupied the mesotympanum with ossicular erosion. In two cases, the cholesteatoma was situated posteriorly and in one case it extended into the mastoid.

All cases conformed to the definition of primary cholesteatoma as the tympanic membranes were intact and there had been no previous surgical intervention.

Discussion

All 12 ears fulfilled the criteria for congenital cholesteatoma. Five of the 12 cholesteatoma were antero-superior in the mesotympanum and lend most support to Michaels' concept of epidermoid formation as a possible source of congenital cholesteatoma (Michaels 1986, 1988a) as they are situated in precisely the same location. In the ears with disease throughout the mesotympanum, the site of origin cannot be determined; these were older children in whom one might speculate that the disease was at a later stage in the natural history. Two of the ears contained cholesteatoma confined to the posterior hypotympanum with normal mucosa antero-superiorly; these could not have arisen from the epidermoid formation, according to Michaels (1986, 1988a, b).

It may be asked why, on the one hand, numerous cases of small, antero-superior congenital cholesteatoma are being reported from the U.S.A., while on the other hand, the present study is the first one from the U.K. and consists of a majority of rather advanced lesions. It can be assumed that many more 'congenital' cholesteatomas are treated in the U.K. at a late stage, where the drum has already been breached. We are left with the possibility that the differences in the rate of diagnosis of congenital cholesteatoma between the two countries, and the stage of the disease when it first presents, may be the result of differences in the organization of paediatric medical practice. In the United States, screening of well children for otological disorders now often includes otoscopy carried out by paediatric physicians trained in that procedure. In the United Kingdom screening is solely by audiometric testing without otoscopy. Most of the patients in our series had hearing loss. In the American cases neither hearing loss nor impedance changes were a feature of the ear disease. There is similar dearth of cases of congenital cholesteatoma in other European countries where screening procedures are similar to the British and do not involve otoscopy.

Apart from the lack of perforation of the tympanic membrane, some of our cases show a resemblance to acquired cholesteatoma. It is feasible that this advanced type of congenital cholesteatoma may not only be a later

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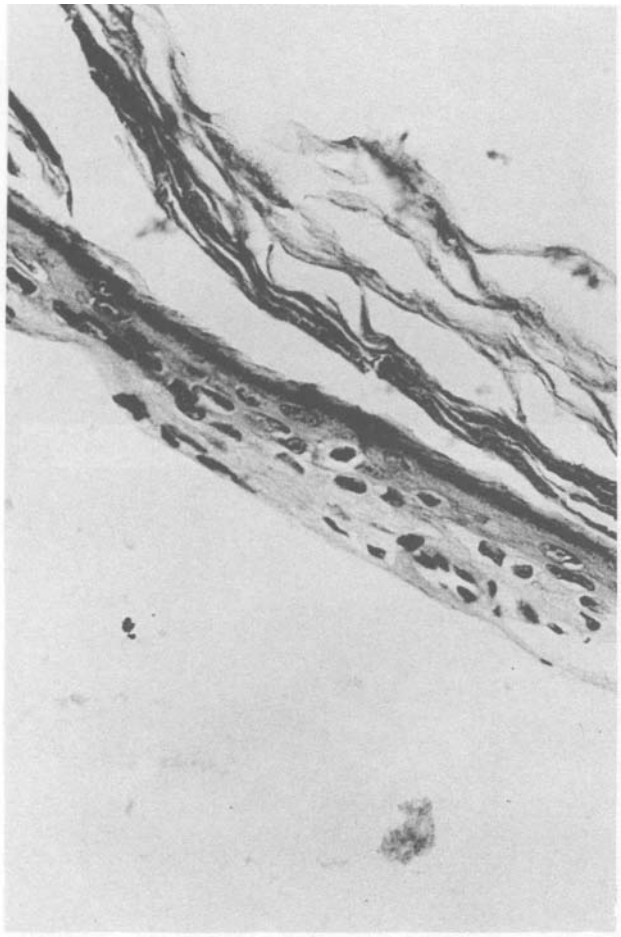

FIG. 4

Histological section of congenital cholesteatoma showing thin malphighian layer and hyperkeratotic squames on the surface. Haematoxylin and eosin ×700.

form of the small American type of the disease, but part of the spectrum of cholesteatoma that has hitherto been described as acquired (Fig. 4).

An otoscopic screening programme in young children, to be carried out routinely as part of preventative medical care is a logical conclusion of the above argument. Resources are not generally available for this to be a doctor-based procedure. Perhaps the utilization of trained nurse-otoscopists as reported by Ruben (1989) might be a measure in helping to reduce the serious morbidity associated with congenital cholesteatoma.

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Key words: Cholesteatoma, congenital.

References

Aimi, K. (1983) Role of the tympanic ring in the pathogenesis of congenital cholesteatoma. *Laryngoscope,* **93:** 1140–1146.

Cohen, D. (1987) Location of primary cholesteatoma. *American Journal of Otology,* **8:** 61–65.

Cruveilhier (1829) Anatomie Pathologique T. II.

Curtis, A. (1979) Congenital middle ear cholesteatoma: two unusual cases and a review of the literature. *Laryngoscope,* **89:** 1159–1165.

Cushing, H. A. (1922) A large epidermal cholesteatoma of the parieto-temporal region deforming the left hemisphere without cerebral symptoms. *Surgery, Gynaecology and Obstetrics,* **34:** 557–566.

House, H. P. (1953) An apparent primary cholesteatoma: case report. *Laryngoscope,* **63:** 712–713.

Levenson, M. J., Parisier, S. C., Chute, P., Wenig, S., Juarbe, C. (1986) A review of 20 congenital cholesteatomas of the middle ear in children. *Otolaryngology—Head and Neck Surgery,* **94:** 560–567.

McDonald, T. J., Cody, D. T. R., Ryan, R. E. (1984) Congenital cholesteatoma of the ear. *Annals of Otology, Rhinology and Laryngology,* **93:** 637–640.

Michaels, L. (1986) An epidermoid formation in the developing middle ear: possible source of cholesteatoma. *Journal of Otolaryngology,* **15:3:** 169–174.

Michaels, L. (1988a) Origin of congenital cholesteatoma from a normally occurring epidermoid rest in the developing middle ear. *International Journal of Pediatric Otorhinolaryngology,* **15:** 51–65.

Michaels, L. (1988b) Evolution of the epidermoid formation and its role in the development of the middle ear and tympanic membrane during the first trimester. *Journal of Otolaryngology,* **17:** 22–28.

Paparella, M. M., Rybah, L. (1978) Congenital cholesteatoma. *Otolaryngologic Clinics of North America,* **11:** 113–120.

Peron, D. L., Schuknecht, H. F. (1975) Congenital cholesteatomata with other anomalies. *Archives of Otolaryngology,* **101:** 498–505.

Piza, J., Northop, E. C., Eavey, R. (1985) Amniotic fluid contents in the ears of neonates and infants. *XIII World Congress of Otorhinolaryngology (Abstract),* **1:** 48.

Ruben, R. (1989) Efficacy of early diagnosis of primary cholesteatoma in infants. Proceedings of Third International Conference in Cholesteatoma and Mastoid Surgery.

Ruedi, L. (1959) Cholesteatoma formation in the middle ear in animal experiments. *Acta Otolaryngologica,* **50:** 233–242.

Sade, J., Babiacki, A., Pincus, G. (1983) The metaplastic and congenital origin of cholesteatoma. *Acta Otolaryngologica,* **96:** 119–129.

Wang, R., Zubick, H. H., Vernick, D. M., Strome, M. (1984) Bilateral congenital middle ear cholesteatomas. *Laryngoscope,* **94:** 1461–1463.

Wingert, R. H., McCurdy, J. A., Hays, L. L. (1976) Congenital tympanic cholesteatoma. *Ear, Nose and Throat Journal,* **55:** 323–327.

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