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CME Article

Clinics in diagnostic imaging (111)

Chee N W C, Suhailee S, Goh J

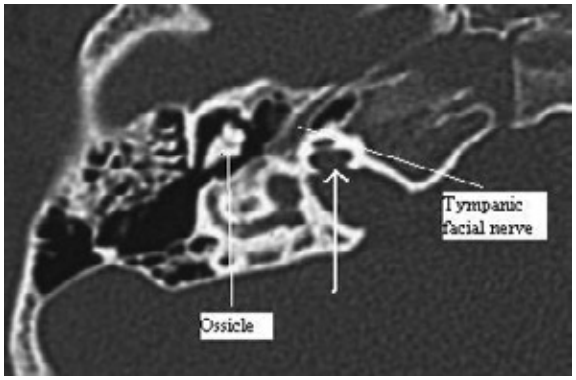

Fig. 1 Axial CT image of the right petrous temporal bone.

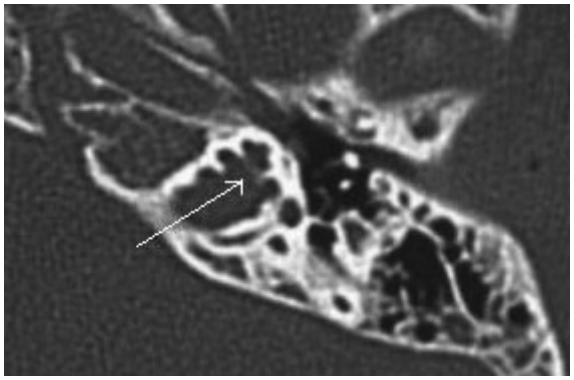

Fig. 2 Axial CT image of the left petrous temporal bone.

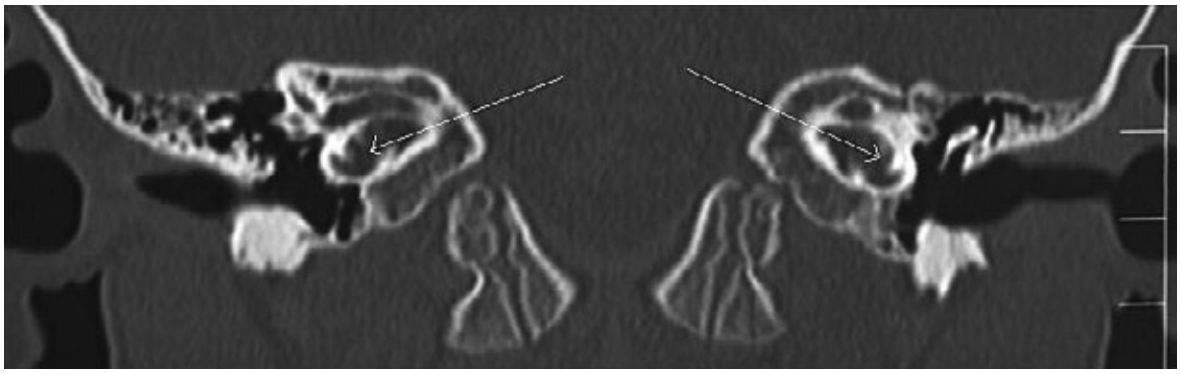

Fig. 3 Coronal CT image of the temporal bones.

CASE PRESENTATION

Two brothers, aged five and six years old, presented with congenital hearing loss and speech delay. Their other siblings, a nine-year-old sister, an eight-year-old brother and a four-year-old brother, all had normal hearing. Both parents also had normal hearing. The birth history was unremarkable. Otomicroscopical examination showed intact tympanic membranes and no middle ear effusion. Pure tone audiometry showed bilateral severe to profound mixed conductive and sensorineural hearing loss. High-resolution computed tomography (CT) of the temporal bones was performed. Both affected brothers had similar CT findings. What do the CT images (Figs. 1-3) show? What is the diagnosis?

Department of Otorhinolaryngology
Tan Tock Seng Hospital
11 Jalan Tan Tock Seng
Singapore 308433

Chee N W C, MBBS, FRCS, FAMS
Senior Consultant

Suhailee S, MBChB, MRCSE
Medical Officer

Department of Diagnostic Radiology

Goh J, MBBS, FRCR
Consultant

Correspondence to:
Dr Nelson WC Chee
Tel : (65) 6357 7742
Fax : (65) 6357 7749
Email: nelson_chee@yahoo.com

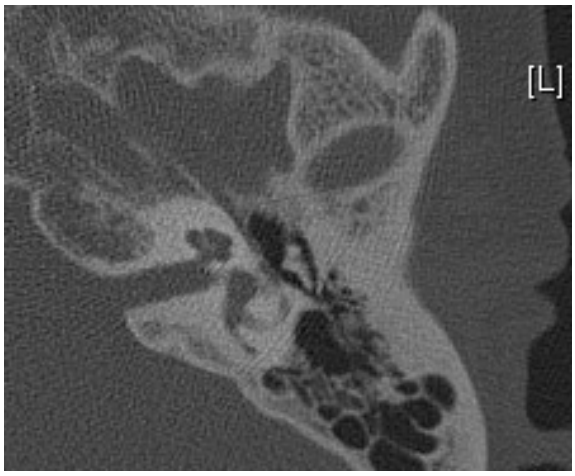

Fig. 4 Axial CT image of the left petrous temporal bone shows normal configuration of the internal auditory canal.

Fig. 5 Coronal CT image of the left temporal bone shows normal configuration of the internal auditory canal.

IMAGE INTERPRETATION

CT of the temporal bones (Figs. 1-3) showed bilateral and symmetric bulbous dilatation of the internal auditory canals. There was incomplete separation of the basal turn of the cochlea from the fundus of the internal auditory canal (white arrows). The middle ear cavities and ossicles appeared normal. The semicircular canals and vestibules were well developed. CT images of the normal internal auditory canal are shown for comparison (Figs. 4 & 5).

DIAGNOSIS

X-linked congenital mixed deafness syndrome.

CLINICAL COURSE

Both brothers were fitted with external digital hearing aids. The parents were counselled on the possibility of cochlear implantation and its attendant surgical risks.

DISCUSSION

Developmental disorders of the inner ear can be divided into malformations affecting only the membranous labyrinth, and those involving both the osseous and membranous labyrinth. The latter group, consisting of conditions such as Mondini's dysplasia, enlarged vestibular aqueduct syndrome and common cavity deformations, is of radiological interest as the conditions are readily diagnosable with modern imaging modalities. However, few are aware of a rare clinical entity known as X-linked progressive mixed deafness syndrome that also presents with pathognomonic CT findings.

The underlying developmental anomaly is an absence of the bony partition between the fundus of the internal auditory canal and the basal turn of the cochlea, allowing a direct communication between the subarachnoid space and the perilymphatic space. The intracranial pressure is transmitted into the perilymphatic space, and the raised perilymphatic pressure that is exerted on the cochlear duct and the stapes footplate results in a mixed pattern of sensorineural and conductive hearing loss respectively[1]. The underlying DFN3 gene (deafness with congenital stapes footplate fixation) has been mapped to the Xq21 region, and is caused by mutations in or around the POU3F4 gene[2]. Female obligate carriers may be normal, or have a milder form of the same anomaly associated with milder hearing loss[3].

Axial CT images of the temporal bones show a symmetric bulbous dilatation of the internal auditory canals in both ears. There is incomplete separation of the basal turn of the cochlea with the fundus of the internal auditory canals[3]. Others have also reported widening of the labyrinthine section of the facial nerve canals[3-5] and abnormality of the vestibular aqueduct[4], although these findings were not seen in our patients. The middle ear spaces and ossicles are normal, and the inner ear vestibular apparatus is well developed.

Preoperative CT diagnosis is important to rule out X-linked deafness in subjects presenting with childhood-onset and progressive conductive or mixed hearing loss. Because of the communication between the subarachnoid space in the internal auditory canal and the perilymphatic space in the cochlear, an unsuspecting surgeon attempting to correct the conductive hearing impairment with a stapedotomy operation would encounter a profuse egress of clear cerebrospinal fluid (gusher) when manipulating the stapes[6,7], and risk worsening the sensorineural hearing loss. External hearing aids and speech training should be recommended,

and surgery avoided, especially for those with a predominantly conductive hearing loss. For severe sensorineural deafness, cochlear implantation may be a viable option. Risks would include cerebrospinal fluid gusher, meningitis and the possibility of introducing the electrodes into the internal auditory canal. Intraoperative and postoperative imaging would be advised to ensure proper placement of the electrode array[5].

ABSTRACT

Two siblings, boys aged five and six years old, presented with mixed hearing loss. Computed tomography of the temporal bones showed bulbous dilatation of the internal auditory canals and incomplete separation with the basal turn of the cochlear, consistent with the diagnosis of X-linked congenital progressive mixed deafness syndrome. The diagnosis and management of this rare condition is discussed.

Keywords: computed tomography, deafness, hearing loss, mixed deafness, X-linked congenital progressive mixed deafness syndrome

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SINGAPORE MEDICAL COUNCIL CATEGORY 3B CME PROGRAMME
Multiple Choice Questions (Code SMJ 200609B)

	True	False
Question 1: Regarding congenital hearing loss:		
(a) Computed tomography (CT) of the temporal bones will always be diagnostic.	❑	❑
(b) Mondini's dysplasia can be identified on CT.	❑	❑
(c) Inner ear malformations affect only the osseous labyrinth.	❑	❑
(d) Common cavity inner ear deformity can be diagnosed on CT.	❑	❑
Question 2: Indicate whether the following statements regarding X-linked congenital mixed deafness syndrome are true or false:		
(a) It only affects males.	❑	❑
(b) It can be diagnosed on CT.	❑	❑
(c) It is a common cause of childhood deafness.	❑	❑
(d) It can be mistaken for middle ear ossicular fixation.	❑	❑
Question 3: Regarding CT for X-linked congenital mixed deafness syndrome:		
(a) It is of no practical purpose.	❑	❑
(b) It shows a widening of the internal auditory canals.	❑	❑
(c) It shows loss of partition between the cochlear and the fundus of the internal auditory canal.	❑	❑
(d) The middle ear structures are well developed.	❑	❑
Question 4: Hearing loss in X-linked congenital mixed deafness syndrome:		
(a) Can deteriorate with time.	❑	❑
(b) Can present as a conductive hearing loss.	❑	❑
(c) Sensorineural deafness is due to raised intracranial pressure exerted on the stapes footplate.	❑	❑
(d) Is reversible.	❑	❑
Question 5: Regarding treatment for X-linked congenital mixed deafness syndrome:		
(a) Surgery should always be considered first.	❑	❑
(b) Hearing aids should be tried, especially for a significant conductive hearing loss.	❑	❑
(c) Surgery on the stapes footplate carries a risk of cerebrospinal fluid leak.	❑	❑
(d) Cochlear implant can be considered to correct the conductive hearing loss.	❑	❑

Doctor's particulars:

Name in full:_____

MCR number:_____ Specialty:_____

Email address:_____

Submission instructions:

A. Using this answer form

1. Photocopy this answer form.
2. Indicate your responses by marking the "True" or "False" box ☑
3. Fill in your professional particulars.
4. Post the answer form to the SMJ at 2 College Road, Singapore 169850.

B. Electronic submission

1. Log on at the SMJ website: URL <http://www.sma.org.sg/cme/smj> and select the appropriate set of questions.
2. Select your answers and provide your name, email address and MCR number. Click on "Submit answers" to submit.

Deadline for submission: (September 2006 SMJ 3B CME programme): 12 noon, 25 October 2006

Results:

1. Answers will be published in the SMJ November 2006 issue.
2. The MCR numbers of successful candidates will be posted online at http://www.sma.org.sg/cme/smj by 15 November 2006.
3. All online submissions will receive an automatic email acknowledgment.
4. Passing mark is 60%. No mark will be deducted for incorrect answers.
5. The SMJ editorial office will submit the list of successful candidates to the Singapore Medical Council.