
PICTORIAL ESSAY

Congenital Inner Ear Malformations

KS Tse, KM Chu, LF Chiu, TW Fan, TK Tsang, TL Kwan

Department of Radiology and Imaging, Queen Elizabeth Hospital, Kowloon, Hong Kong

ABSTRACT

Hearing impairment is defined as a person's hearing threshold being above the normal range of -10 to 15 decibels. It has significant impact on a child's language, cognitive, socioemotional and behavioural development. Therefore early recognition of hearing impairment is of utmost importance. Congenital inner ear malformation is an important cause of sensorineural hearing loss in children. It consists of labyrinthine aplasia, cochlear aplasia, common cavity deformity, cystic cochleovestibular anomaly (incomplete partition type I), cochlear hypoplasia, Modini deformity (incomplete partition type II). The imaging manifestations of these conditions on computed tomography examination are discussed in this article. Semicircular canal malformation and internal auditory canal deformities, namely: atresia of the internal auditory canal and X-linked progressive hearing loss, are also illustrated.

Key Words: Cochlear implantation; Congenital abnormalities; Ear, inner; Hearing loss, sensorineural

中文摘要

先天性內耳畸形

謝健榮、朱嘉敏、趙朗峰、范子和、曾子勤、關鼎樂

聽覺障礙是指聽力閾值超過正常範圍的-10至15分貝。聽覺障礙對於小童的語言能力、認知、社會情緒及行為發展都有相當重要的影響，所以應盡早確定小童是否患有聽覺障礙。先天性內耳畸形是造成小童感覺神經性聽力損失的一個重要原因，分為迷路發育不全、耳蝸發育不全、共同腔畸形、囊性耳蝸前庭異常（未完全分隔一型）、耳蝸發育低下、和Modini內耳發育不全（未完全分隔二型）不同種類。本文討論這些病變的CT表現。此外，亦討論半規管畸形及內聽道畸形，即內聽道閉鎖和與X染色體有關的漸進式聽力損失。

INTRODUCTION

Globally, hearing impairment occurs in about 1 to 2 per 1000 live births. In Hong Kong, about 650 children under the age of 15 years were registered as having significant hearing impairment in the Central Registry for Rehabilitation in 2004.¹ As hearing

loss has adverse effects on language, cognition, behavioural and socioemotional development, it is crucial to recognise this problem promptly. This article illustrates the computed tomography (CT) findings associated with various congenital inner ear malformations.

Correspondence: Dr KS Tse, Room 117, Block K, Department of Radiology and Imaging, Queen Elizabeth Hospital, 30 Gascoigne Road, Kowloon, Hong Kong.

Tel: (852) 2958 2697; Fax: (852) 2760 4022; Email: sunnytks@gmail.com

Submitted: 16 May 2011; Accepted: 28 Jun 2011.

Bony labyrinthine development occurs primarily between the 4th and 8th gestational weeks, and as it continues ossification ensues between the 8th and 24th gestational weeks. Development of sensory epithelium including the organ of Corti occurs simultaneously with the growth and ossification of the bony labyrinth between the 8th and 24th gestational weeks. Insults occurring beyond the 8th week generally result in membranous labyrinthine deformities, which usually have no manifestations on anatomical imaging, i.e. CT and magnetic resonance imaging (MRI). In fact, a large majority of patients with congenital sensorineural hearing loss have defects restricted to the membranous labyrinth, whereas only 20% have bony labyrinthine deformities demonstrable by CT.²

In this context the embryogenesis of inner ear should be appreciated, at least in outline. Thickening of the ectoderm between the first branchial groove and rhombencephalon forms the otic placode at the 3rd gestational week, which then invaginates into the mesenchyme forming the otic pit at the 4th week. It further enlarges to become the otocyst at the end of the 4th week. The otocyst then differentiates to form dorsal and ventral pouches. By the end of the 8th week, the morphology of the labyrinth is generally identifiable. The otic capsule develops as a cartilaginous condensation of the mesenchyme from the 4th week onwards, and subsequently grows and ossifies to form the bony labyrinth. In general, the saccule, endolymphatic duct and utricle are completed by the 11th week, and the semicircular canals are

completed between the 19th and 22nd week, the lateral semicircular canal being the last to form. Therefore when the insults occur early during the 4th to 8th gestational week, cochlear deformity results. By contrast, injury ensuing after the 8th week results in malformation of the vestibule and semicircular canals, while the cochlea is preserved.²

Notably, that middle and external ear have a separate embryological origin from that of the inner ear. This explains the low frequency (about 10%) of associated middle or external ear anomalies in inner ear dysplasia (Figure 1). On the other hand, the development of the facial nerve canal has a complex interaction with the otic capsule. Therefore it is important to look for an anomalous course of the facial nerve in inner ear dysplasia (Figure 1).²

COCHLEAR MALFORMATION

Congenital sensorineural hearing loss can be classified based on its aetiology. Broadly, causes can be divided into: non-genetic (including infective, metabolic, traumatic, and teratogenic) and genetic. As mentioned, only a small proportion of patients with congenital sensorineural hearing loss have bony labyrinthine deformity. Thus, the classification system of congenital cochlear malformations suggested by Jackler et al³ and Sennaroglu and Saatci⁴ which entails a correlation with the early stages of bony labyrinth embryogenesis is practical from the radiologist's perspective. The classes include: labyrinthine aplasia, cochlear aplasia, common cavity deformity, cystic cochleovestibular anomaly

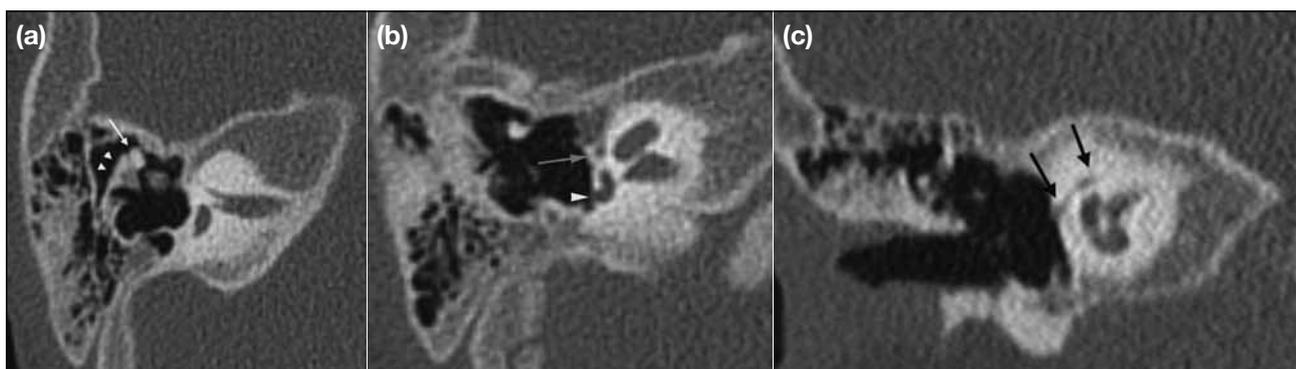


Figure 1. Semicircular canal aplasia with middle ear deformity.

(a) An axial computed tomographic (CT) image in bone window shows a narrowed and deformed middle ear cavity. The malleus (arrow) and incus (arrowheads) are rotated laterally, and abut on the lateral wall of the middle ear cavity. (b) The CT shows a rudimentary vestibule (arrowhead) with a small featureless cavity at the posterior labyrinth. The semicircular canals are aplastic. The labyrinthine portion of the facial nerve canal (arrow) is posterolaterally located. (c) The coronal CT image also confirms the posterolaterally located labyrinthine and tympanic portion of the facial nerve (arrows). The tympanic portion should normally run inferior to the lateral semicircular canal, which does not do so in this case.

(incomplete partition type I), cochlear hypoplasia and Mondini deformity (incomplete partition type II). Moreover, they depend on the timing of the insult, with respect to fetal development. The earlier it takes place, the more severe the hearing impairment.

Arrest of the otic placode's development at the 3rd gestational week leads to labyrinthine aplasia. Labyrinthine aplasia, also known as Michel deformity, is an extremely rare and very severe anomaly. It is characterised by the absence of the cochlea, vestibule and semicircular canals, there being no inner ear structures in the petrous bone on CT or high-signal membranous labyrinth on T2-weighted MRIs. The internal auditory canal (IAC) can vary from being hypoplastic to completely absent. The petrous bone is often hypoplastic or even absent (in more severe cases).³⁻⁵

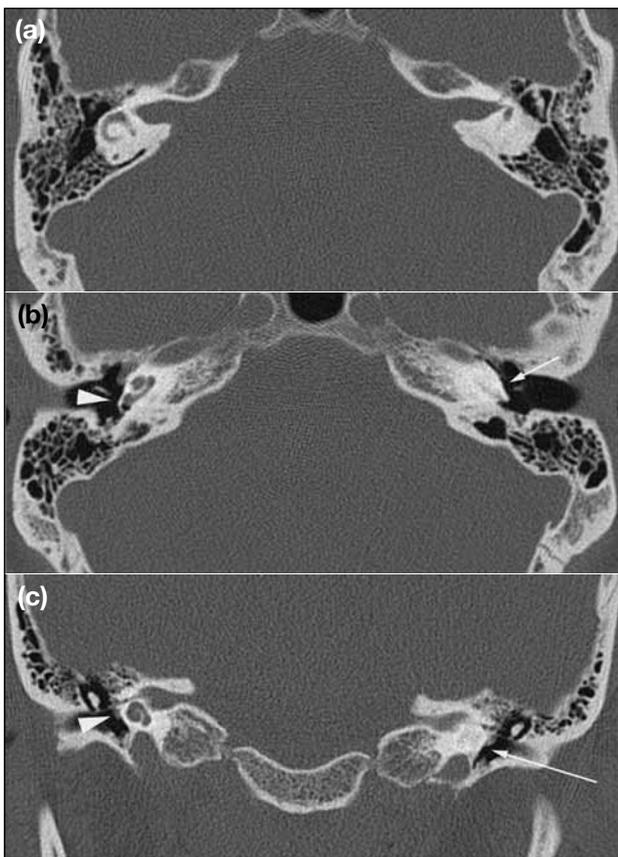


Figure 2. Cochlear aplasia. Axial computed tomographic images in the bone window shows (a) an absence of semicircular canals, and the presence of a rudimentary vestibule on left side. The left internal auditory meatus is slightly hypoplastic. (b) At the anterior aspect of bony labyrinth, a normal cochlea is not seen on left side as compared to the right side. The lateral wall of the inner ear is flat on left side (as indicated by the arrow), in contrast to the normal convexity of the lateral wall of cochlea (arrowhead) on right side in (b) and (c).

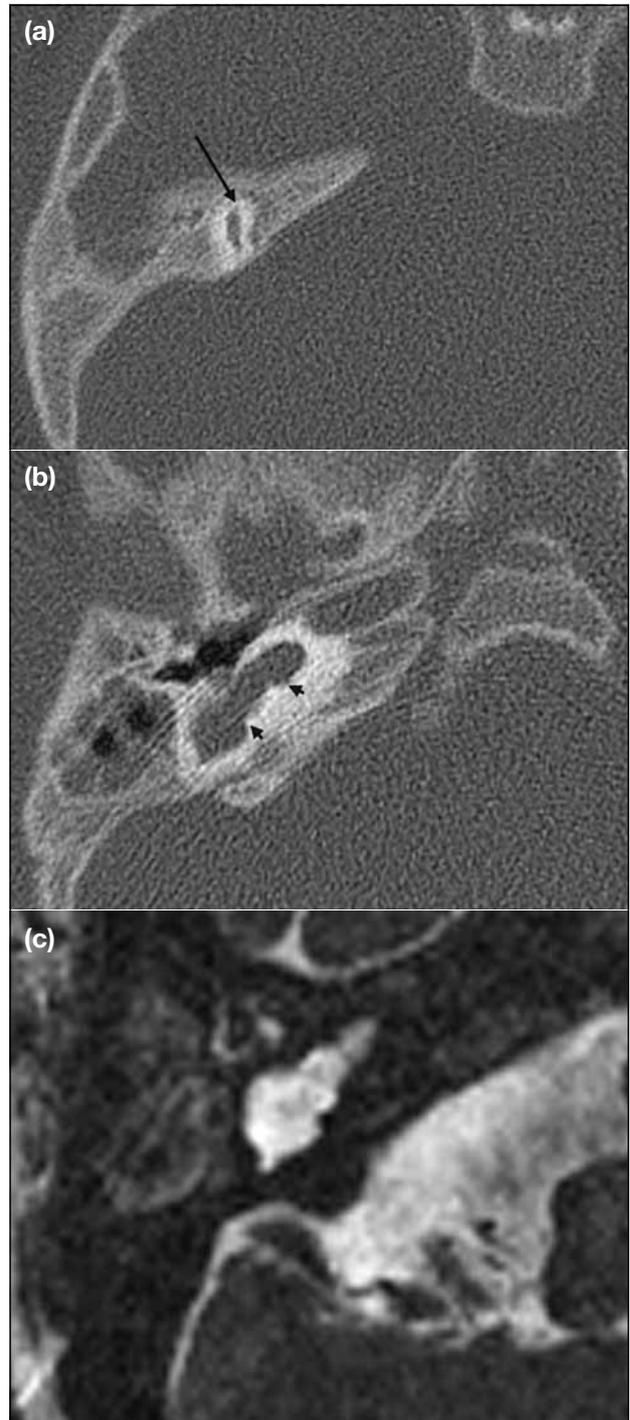


Figure 3. Common cavity deformity. Axial computed tomographic images in (a) bone window shows there is dilatation of the right superior semicircular canal (arrow). There is an ovoid-shaped featureless cystic cavity (short arrows) at the bony labyrinth, with absence of a normal configuration of a cochlea, vestibule, posterior and lateral semicircular canal, as shown in (b). Normal modiolus is also absent. Features are suggestive of common cavity deformity with a dysplastic superior semicircular canal. Correlation with a high-signal intensity structure suggestive of a featureless cystic cavity is illustrated on the axial magnetic resonance T2-weighted image (c).

Cochlear aplasia results from the arrest of otic development at the late 3rd gestational week. It is characterised by absence of the cochlea, and variable development of vestibule and semicircular canals. On CT, the anterior bony labyrinth is hypoplastic, with absence of cochlear development or normal convexity of the cochlear promontory. The posterior labyrinthine structures are usually dysplastic, and the IAC may be normal, hypoplastic or dilated (Figure 2). Associated absence of the cochlear nerve is common. It differs from labyrinthine ossificans, which is an acquired condition in which the cochlea has been formed but the lateral wall of anterior labyrinth is flat (instead of having a convex in configuration). As for labyrinthine aplasia, cochlear aplasia is an absolute contraindication for cochlear implantation.³⁻⁶

When the otocyst fails to differentiate into the primordia of cochlea, vestibule and semicircular canals between the 4th and 5th week of gestation, common cavity deformity of the inner ear occurs. There is a single common cystic cavity encompassing the cochlea, vestibule and semicircular canals, with the IAC entering the cavity at its mid portion. The cystic cavity is usually ovoid in shape but has no internal architecture. Its size varies with the stage of developmental arrest. The semicircular canals are typically absent, but sometimes

normal or dysplastic. The IAC is often abnormal (Figure 3). The presence of a featureless and undifferentiated ovoid cystic cavity spanning over the bony labyrinth is the key imaging finding in this entity. The position of the cystic cavity in relation to the IAC is crucial in making the correct diagnosis. In common cavity deformity, the IAC lies at the central portion of the cyst. On the contrary, in cochlear aplasia, the cyst is related to dysplastic vestibule and the semicircular canals are situated posterior to the IAC.³⁻⁵

Cystic cochleovestibular anomaly, now named 'incomplete partition type I', demonstrates better otic development than the common cavity deformity. Instead of a single common cavity, it entails two separated cystic cavities that represent the rudimentary cochlea and vestibule, and assumes a 'figure of 8' or 'snowman' appearance. The cavity at the anterior labyrinth represents the cystic unpartitioned cochlea. As in common cavity deformity, the modiolus is absent, and the fundus of the IAC is defective. The underdeveloped vestibule manifests as a cystic cavity at the posterior labyrinth. Semicircular canals are usually present but dysplastic. The vestibular aqueduct is usually normal. The 8th nerve is either deficient or absent, and the latter situation is also an absolute contraindication for cochlear implantation.³⁻⁵

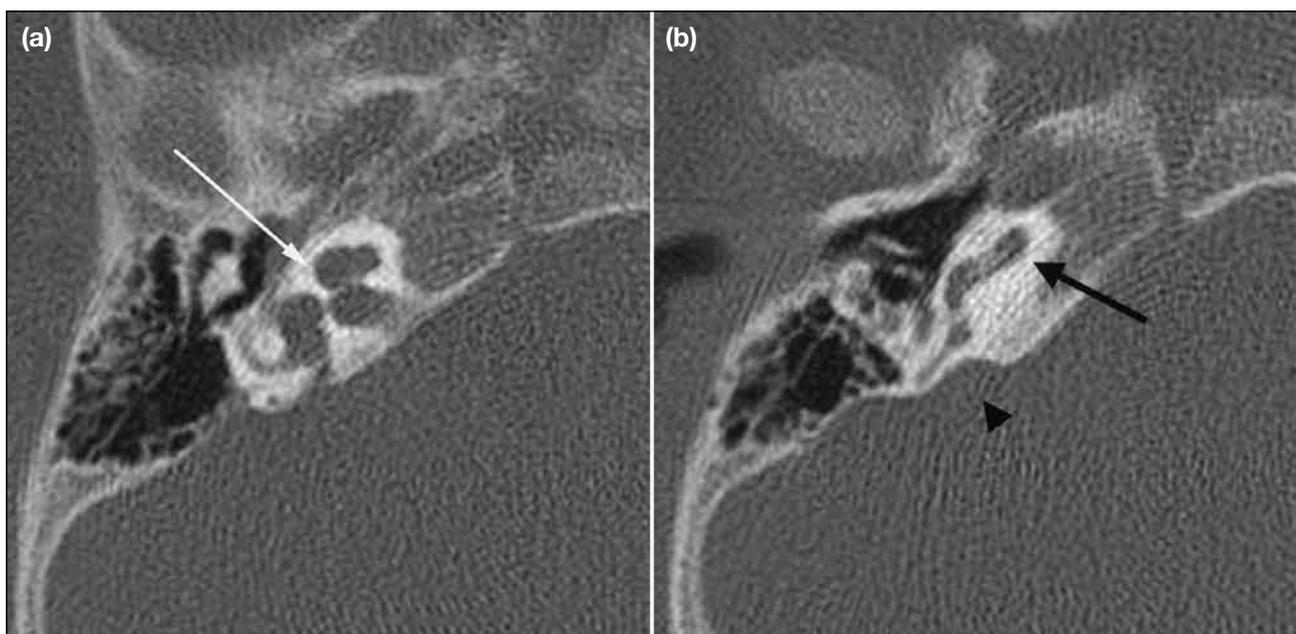


Figure 4. Incomplete partition type II.

Axial computed tomographic images in bone windows. (a) There is absence of normal partition between the apical and middle turns of right cochlea, as indicated by arrow. (b) The basal turn of the cochlea is preserved (arrow). An associated enlarged vestibular aqueduct (arrowhead) is evident.

According to Sennaroglu and Saatci,⁴ cochlear hypoplasia occurs when there is defective development of the cochlea during the 6th week. The cochlea and vestibular structures are separated from each other. However the cochlea is smaller than normal, and has only one or less than one turn. The vestibule and semicircular canals are usually small and malformed. The vestibular aqueduct is often normal.⁴

The Mondini deformity is now better called ‘incomplete partition type II’. It is the second most common imaging finding in children with sensorineural hearing loss. The

original description in 1791 classically included the triad of: (1) absence of a partition between the apical and middle turns of cochlea, (2) a large vestibule, and (3) an enlarged vestibular aqueduct. The pathology of incomplete partition type II lies at the absence of interscalar septum, which separates the apical and middle cochlear turns, resulting in a baseball cap–like cochlear apex. The basal turn is intact. Therefore there are one and a half cochlear turns, instead of normal two and a half. The cochlea is of normal size (Figure 4). Patients usually present with variable sensorineural hearing loss, with high-frequency hearing preserved.

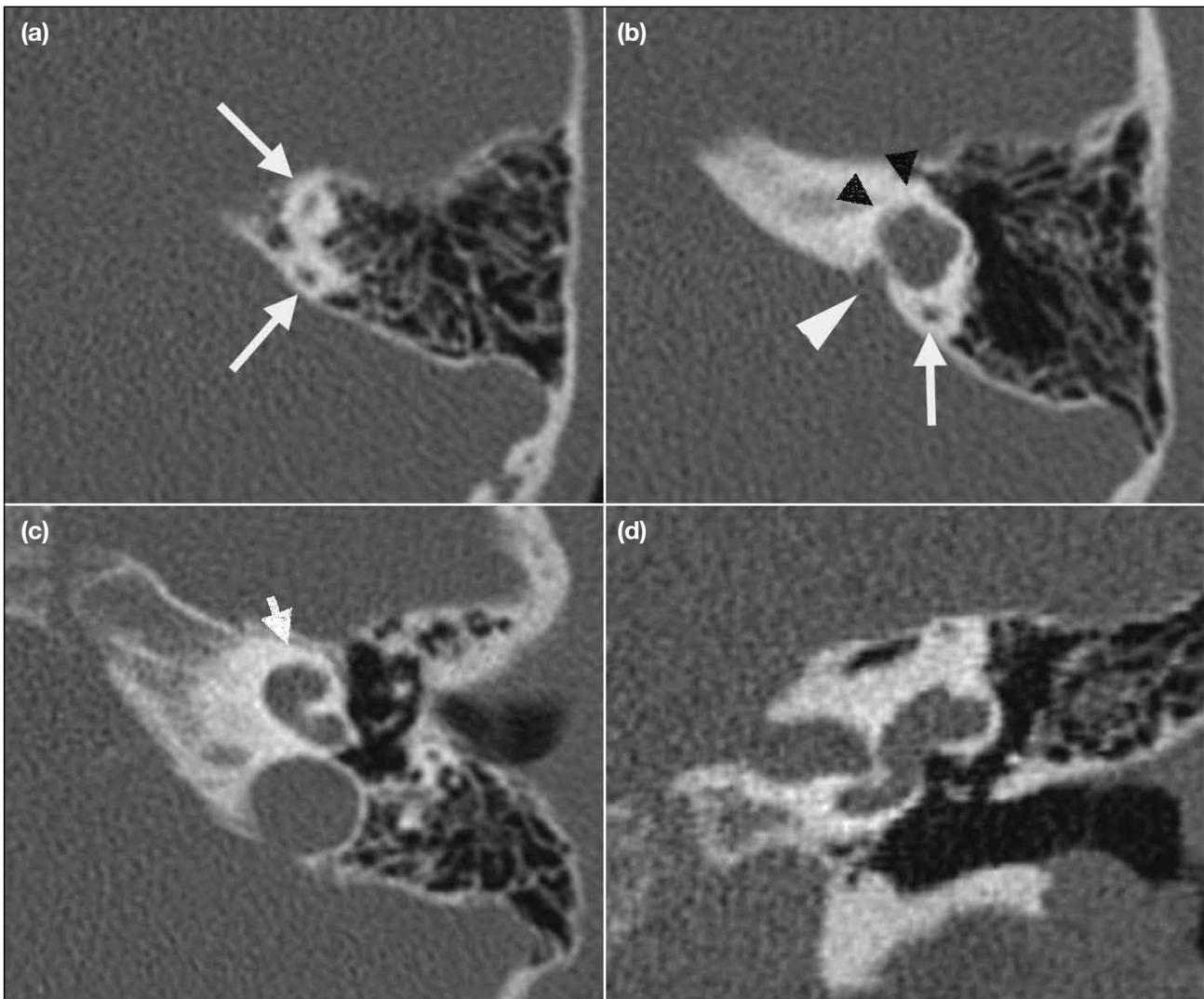


Figure 5. Semicircular canal, vestibule, and cochlear dysplasia.

(a and b) The left superior semicircular canal (white arrows) is dilated. The normal posterior and lateral semicircular canals, as well as the vestibule are absent. There is formation of a featureless cystic cavity (dark arrowhead in b) at the expected position of vestibule and lateral semicircular canal. Features are suggestive of semicircular canal and vestibule dysplasia. Associated enlarged vestibular aqueduct (white arrowhead) is noted. (c and d) Cochlear dysplasia (short white arrow) is also observed, with dilated cochlear turns, and incomplete apical and middle turn partition.

Perilymphatic fistula (giving rise to otorrhea and recurrent meningitis) is also possible, due to a defective modiolus. This condition is commonly associated with an enlarged vestibular aqueduct, which is not seen in earlier, more primitive developmental arrests. Anomalies of the vestibule, semicircular canals, and vestibular aqueduct occur in about 20% of cases. Anomalies of the stapes (absence or dysplasia of its footplate) are also common, and may also occur in many syndromes.^{3,4}

It is important to understand the above entities represent the prototypes of various stages of developmental arrest. In real life, many deformities do not fit readily into any of the above categories, for which reason it is more appropriate to describe these anomalies separately in anatomic terms. This includes the cochlear morphology, including: the number of cochlear ducts, any absence of modiolus and osseous spiral lamina; and the septations between the turns, vestibule, semicircular canal. In addition, the vestibular aqueduct has to be evaluated carefully by CT (Figure 5). The IAC should also be assessed for its cross-sectional dimension and morphology, particularly any dilatation of the fundus and dehiscence between the fundus of the IAC and the basal turn of the cochlea.²

SEMICIRCULAR CANAL MALFORMATION

As mentioned before, the semicircular canals start to develop from the vestibular anlage between the 6th and 8th week, with the superior one being the first, followed by posterior and then the lateral canal. The development

is not completed until the 19th to 22nd week. Semicircular canal anomaly is often associated with cochlear dysplasia, but may rarely occur in an isolation. Semicircular canal aplasia refers to the absence of a semicircular canal (Figure 6). This condition is also one of the manifestations of the so-called CHARGE association (syndrome). Semicircular canal dysplasia is a more common anomaly; isolated lack of development of posterior semicircular canal is implicated in the Alagille and Waardenburg syndromes.²

ENLARGED VESTIBULAR AQUEDUCT

The vestibular aqueduct is another important structure to examine in case of congenital sensorineural hearing loss. This aqueduct is a bony canal, extending behind the labyrinth from the vestibule to the posterior surface of the petrous bone. It contains the endolymphatic duct, which continues as a blind-ended endolymphatic sac. It represents the non-sensory part of membranous labyrinth, and is involved in normal endolymph resorption, digestion of foreign bodies, and pressure equalisation between the cerebrospinal fluid (CSF) and endolymph. It is regarded as enlarged if the size of the mid portion is greater than 1.5 mm, or its diameter is larger than that of the semicircular canal. Enlargement of vestibular aqueduct is the most common imaging abnormality in children with congenital sensorineural hearing loss. It is usually bilateral (Figure 7). According to Robson,⁷ identification of vestibular aqueduct enlargement should prompt careful evaluation of cochlea for any associated malformation, ranging from subtle asymmetry of the modiolus to the Modini

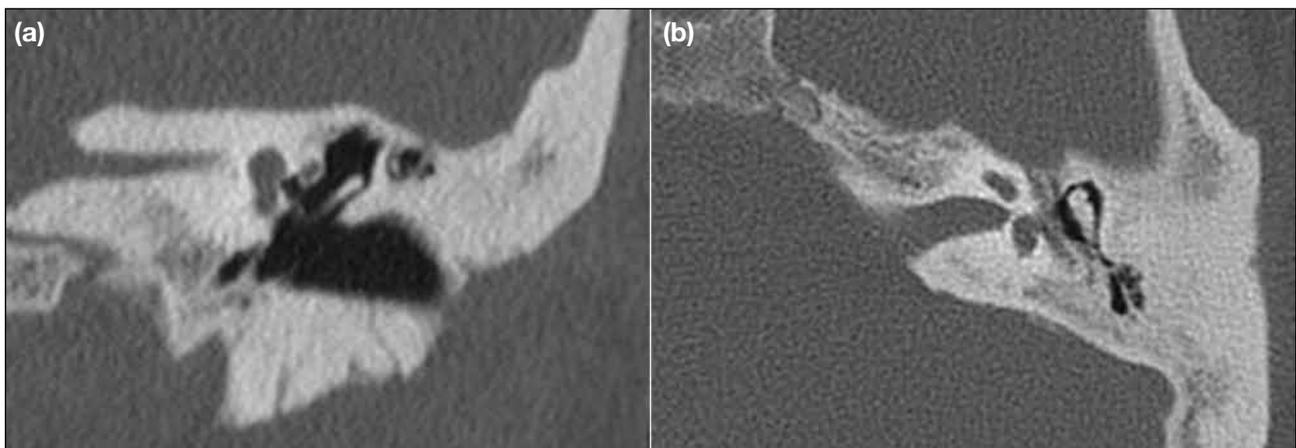


Figure 6. Semicircular canal aplasia.

The superior, posterior and lateral semicircular canals are absent. A rudimentary vestibule in the left petrous temporal bone are seen in (a) the coronal and (b) axial computed tomographic images.

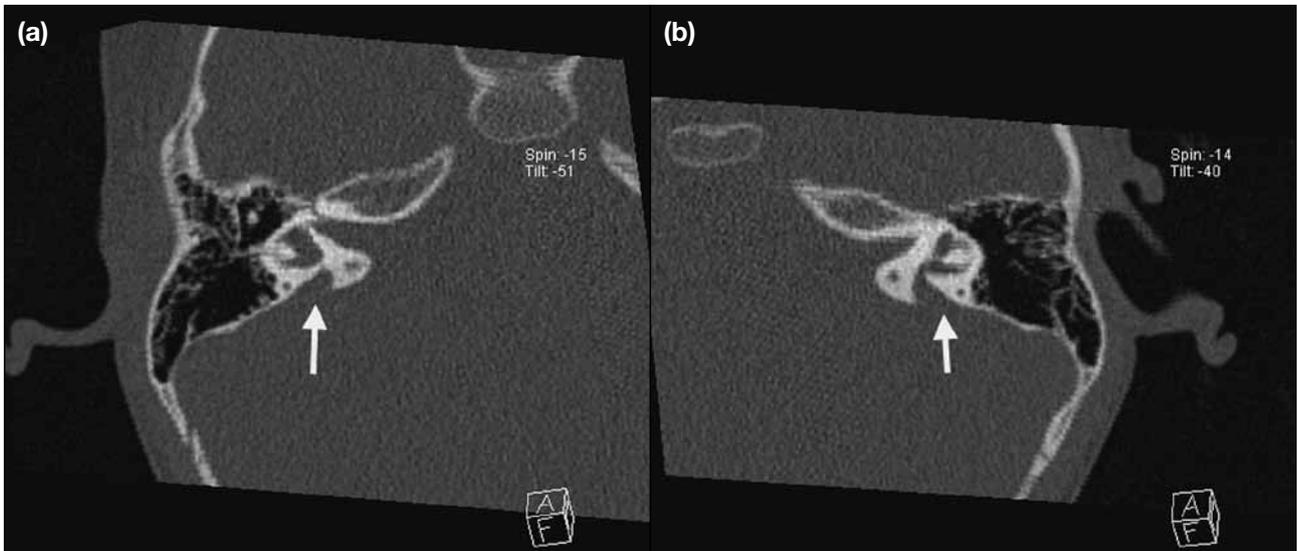


Figure 7. Enlarged vestibular aqueduct.
(a and b) Bilaterally the vestibular aqueducts are enlarged (arrows), measuring more than 1.5 mm in their mid portion.

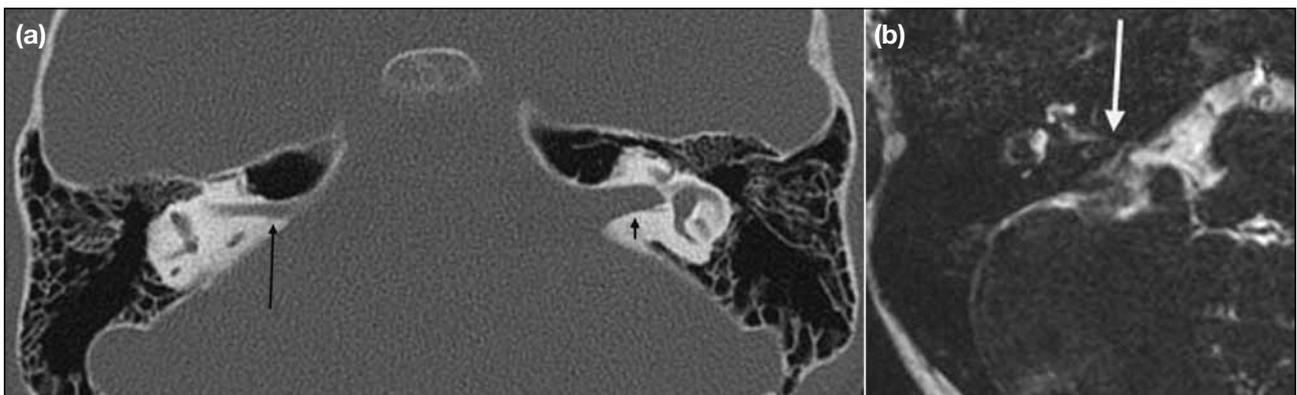


Figure 8. Absent vestibulocochlear nerve.
(a) An axial computed tomographic image demonstrates a narrowed right internal auditory canal (long dark arrow), as compared to the normal left internal auditory meatus (short dark arrow). (b) The absence of the 8th cranial nerve was subsequently confirmed by magnetic resonance imaging (white arrow).

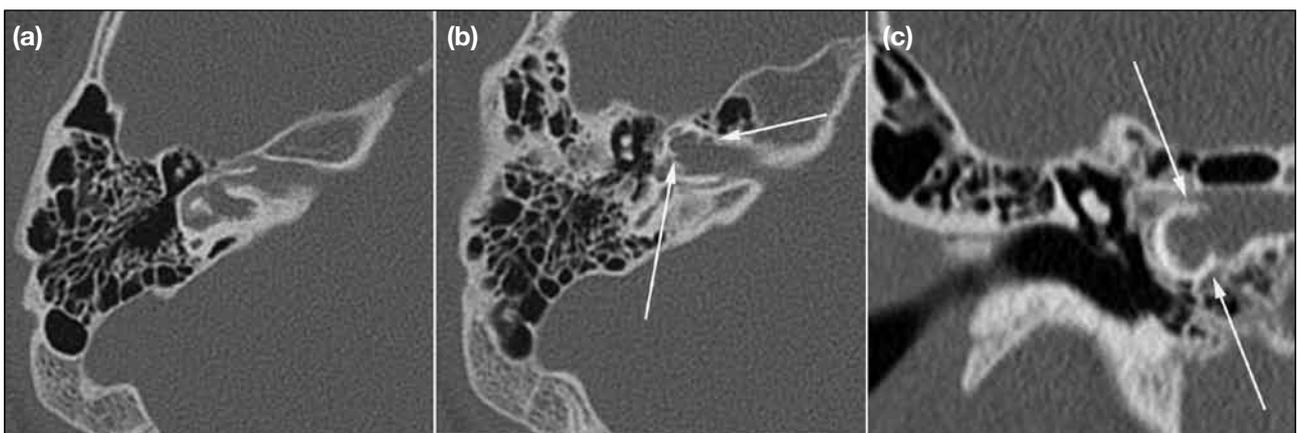


Figure 9. X-linked progressive hearing loss.
Axial computed tomographic images (a) in a bone window shows normal calibre of the right internal auditory canal (IAC), except that there is bulbous dilatation of its fundus. (b) The partition between the basal turn of the cochlea and IAC is deficient, and the modiolus of the cochlea is absent (arrows). (c) The absence of modiolus and the dehiscence between the fundus and the basal turn of cochlea is again demonstrated by the arrows in the coronal reformatted image.

deformity. It seldom occurs in earlier malformations (common cavity deformity, cystic cochleovestibular anomaly). Deformity of semicircular canals, vestibule and IAC is also common.⁸ As this entity implies a potential for deterioration in hearing after minor head trauma, early and prompt diagnosis is necessary.

INTERNAL AUDITORY CANAL MALFORMATION

IAC abnormalities include absence, stenosis, and enlargement. A stenotic IAC (down to about 1 to 2 mm in diameter) is often associated with the absence of the cochlear nerve (Figure 8). As this is a contraindication for cochlear implantation, it is essential to ascertain the status of vestibulocochlear nerve. The nerves in the cerebello-pontine angle and IAC may be better assessed on T2-weighted MRI, and are best examined in the plane perpendicular to the nerves and IAC.^{9,10}

The morphology of the IAC should also be assessed. In X-linked progressive hearing loss, there is dehiscence between the partition between the fundus of IAC and basal turn of the cochlea. Patients with such hearing problems have mixed sensorineural and conductive loss. The characteristic finding on CT is the bulbous dilatation of the fundus of IAC (Figure 9), and there may be an associated deficient or absent modiolus. Due to the defect between the IAC and cochlea, CSF pulsation can be transmitted from the IAC to the perilymph in the cochlea and vestibule. The CSF pressure can also be transmitted to the geniculate ganglion and tympanic segment of the facial nerve canal. In these patients, it may be possible to see enlargement of the facial nerve canal and vestibular aqueduct.^{9,11} Recognition of this condition is essential, as there is a potential risk of a CSF gusher during stapes surgery.

Although congenital inner ear malformations account for a small proportion of children with sensorineural hearing loss, for a radiologist it is of paramount importance to understand the imaging manifestations of these conditions. Being familiar with the prototype of various stages of developmental arrest, and coupled with careful description of anomalies in anatomic terms allows proper communication with clinicians and facilitates multi-disciplinary management.

REFERENCES

1. Developmental Disorder Series: Hearing Impairment. Child Assessment Service, Department of Health, Hong Kong. Child Assessment Service, Department of Health, Hong Kong website: http://www.dhcas.gov.hk/textonly/english/health_pro/public_edu_11.html. Accessed May 2011.
2. Swartz JD, Mukherji SK. The inner ear and otodystrophies. In: Swartz JD, Loevner LA. Imaging of the temporal bone. New York: Thieme Medical Publisher; 2008. p 298-411.
3. Jackler RK, Luxford WM, House WF. Congenital malformations of the inner ear: a classification based on embryogenesis. *Laryngoscope*. 1987;97(3 Pt 2 Suppl 40):2-14.
4. Sennaroglu L, Saatci I. A new classification for cochleovestibular malformations. *Laryngoscope*. 2002;112:2230-41.
5. Hudgins PA. Inner ear, congenital. In: Harnsberger HR. Diagnostic imaging: head and neck. Amrisys; 2004. I-2-96-111.
6. Witte RJ, Lane JI, Driscoll CL, Lundy LB, Bernstein MA, Kotsenas AL, et al. Pediatric and adult cochlear implantation. *Radiographics*. 2003;23:1185-200.
7. Robson CD. Congenital hearing impairment. *Pediatr Radiol*. 2006;36:309-24.
8. Koesling S, Rasinski C, Amaya B. Imaging and clinical findings in large endolymphatic duct and sac syndrome. *Eur J Radiol*. 2006;57:54-62.
9. Casselman JW, Offeciers EF, De Foer B, Govaerts P, Kuhweide R, Somers T. CT and MR imaging of congenital abnormalities of the inner ear and internal auditory canal. *Eur J Radiol*. 2001;40:94-104.
10. Veillona F, Ramos-Taboada L, Abu-Eid M, Charpiot A, Riehm S. Imaging of the facial nerve. *Eur J Radiol*. 2010;74:341-8.
11. Altay H, Savaş R, Oğüt F, Kirali T, Alper H. CT and MRI findings in X-linked progressive deafness. *Diagn Interv Radiol*. 2008;14:117-9.