

CT and MRI findings in X-linked progressive deafness

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ABSTRACT

Congenital X-linked mixed deafness is a rare anomaly that has typical features and can be diagnosed on the basis of progressive mixed hearing loss and the typical imaging findings. Recognition of these findings may alter the course of treatment and perilymph gushing can be avoided. A 10-year-old male patient presented with a history of progressive hearing loss. Computed tomography of the temporal bones showed bulbous dilatation of the fundi of the internal auditory canals (IAC) and the absence of the bony plates separating the basal turn of the cochleas and IAC. Magnetic resonance imaging demonstrated obliteration of the labyrinthine spaces at the right side.

Key words: • computed tomography • magnetic resonance imaging • X-linked congenital progressive deafness

The association between mixed X-linked deafness and the abnormal communication between the cerebrospinal fluid and perilymph, resulting in leakage during stapes surgery (“stapes gusher”) is rare. Although stapes surgery is often performed to improve conductive hearing loss, such surgery is not recommended in this disorder, because there is a high likelihood of worsening the hearing loss. Therefore, recognition of mixed X-linked deafness is essential. We present the imaging findings of a male patient with congenital progressive mixed hearing loss with stapes gusher whose complaints became apparent soon after meningitis infection.

Case report

A 10-year-old male patient presented with bilateral hearing loss. His family reported that he had longstanding mild-to-moderate hearing loss; two months prior to evaluation, he had meningitis, followed by total bilateral hearing loss. Physical examination revealed bilateral negative Rinne tests by using the 512-Hz tuning fork. On evoked response audiometry (ERA) testing at our hospital, there was no response at 100 Db. Computed tomography (CT) of the temporal bones obtained by using 1-mm axial and coronal sections showed bulbous dilatation of the fundi of the internal auditory canals (IAC) (Fig. 1) and the absence of the bony plates separating the basal turn of the cochleas and IAC (Fig. 2). The labyrinthine segments of the facial canals were normal. The vestibular aqueducts had normal size. On the right side, the semicircular canals and the vestibule were partially obliterated. On the left side, the semicircular canals and the vestibule were normal. The middle ear cavity and the ossicles were completely normal. Lumbar puncture was performed, and opaque material (iohexol, 4 cc) was introduced into the subarachnoid space; progression of the opaque material from the IAC into the cochlea was observed (Fig. 3), leading us to conclude that there was passage of cerebrospinal fluid from IAC into the cochlea through bony defect. The three-dimensional Fourier transformation constructive interference in steady-state (3D FT-CISS) magnetic resonance imaging (MRI) sequence (TR, 12.25 ms; TE, 5.9 ms; slice thickness, 1 mm) successfully demonstrated obliteration of the labyrinthine spaces at the right side (Fig. 4). Obliteration of the semicircular canals on the right side was thought to be related to meningitis.

Discussion

The association between X-linked deafness and the flow of perilymph or cerebrospinal fluid during stapes surgery (stapes gusher) was first described by Nance et al. (1). Because of the X-linked recessive type of inheritance, male patients had severe mixed hearing loss at all frequencies, but female carriers were normal or had mild hearing loss. This is a rare

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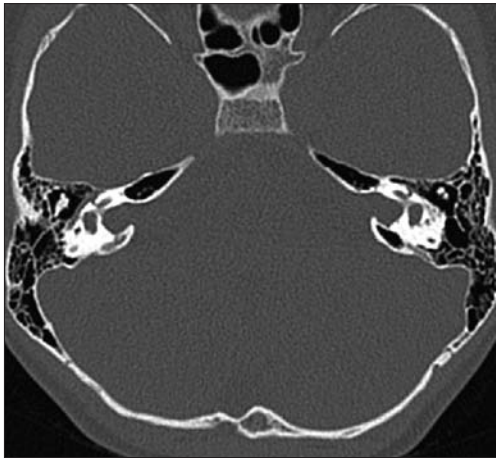


Figure 1. Axial temporal CT section (thickness, 1 mm) demonstrates bilateral bulbous dilatation of the fundi of the internal auditory canals.

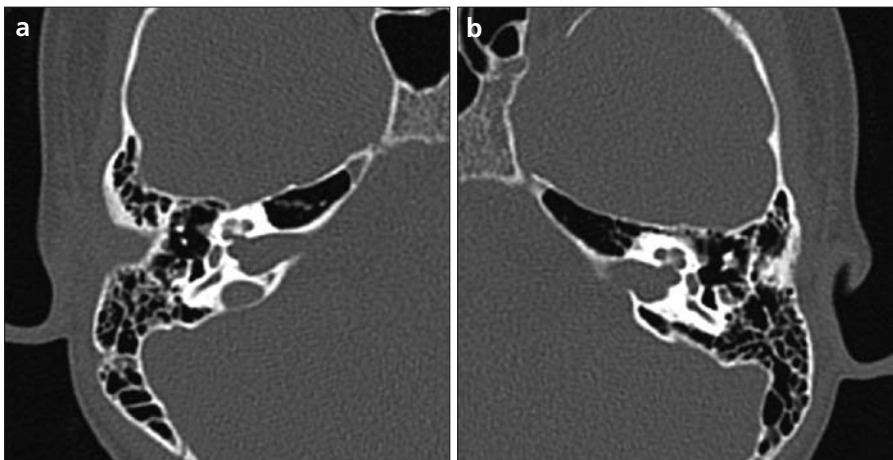


Figure 2. a, b. Axial temporal CT sections of the right (a) and left (b) temporal bones (thickness, 1 mm) show the bilateral absence of the bony plates separating the basal turn of the cochleas and internal auditory canals.

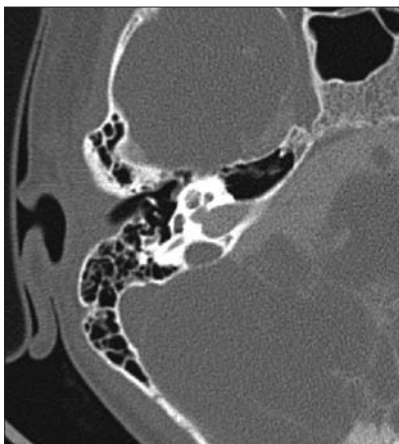


Figure 3. Axial plane CT section (thickness, 1 mm) of the right temporal bone show the progression of the opaque material from the internal auditory canal into the cochlea after opaque material (iohexol) was given into the subarachnoid space by lumbar puncture.

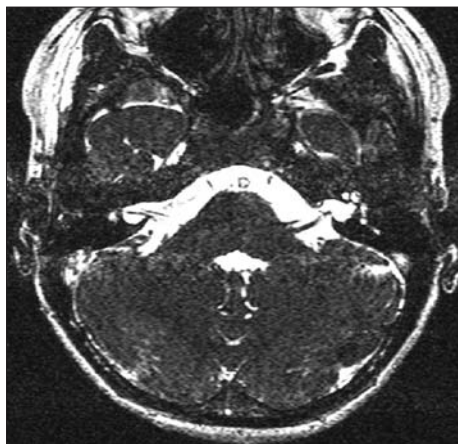


Figure 4. Axial 3D FT-CISS (three-dimensional Fourier transform constructive interference in steady state sequence) MR image (TR, 12.25 ms; TE, 5.9 ms; slice thickness, 1 mm) demonstrates obliteration of the labyrinthine spaces on the right side.

disorder, and preoperative recognition of this problem plays a crucial role in the identification of patients at risk for stapes gushing; surgery is avoided because of the risk of worsening of sensorineural hearing loss (2).

The association of X-linked mixed deafness with stapes gusher has been recognized for many years; initial imaging studies by polytomography showed dilatation of the lateral end of the IAC in some cases (3, 4). Later, using thin section high resolution CT in two planes, detailed assessment of the state of the middle and inner ears became possible. The first and the largest case series describing the CT features of X-linked mixed hearing loss was published by Phelps et al. (5). They reported imaging features of this syndrome across seven pedigrees. They found that 16 of 24 affected male patients had bulbous IAC and incomplete separation of basal turn of cochleas from the fundi of the IAC. They also demonstrated enlarged first and second parts of intratemporal facial nerve canals. They reported that some of the obligate female carriers had milder forms of the same anomaly with milder forms of hearing impairment. Later, Talbot and Wilson (6) described four patients with X-linked deafness. In addition to the typical findings described above, they described absence of bony modiolis and abnormal vestibular aqueducts in all patients. Tang and Parnes (7) described two patients with widening of the fundi of the IAC and absent bony plate between them and the basal turn of the cochlea, and Papadaki et al. (8) reported two female cases of this syndrome with normal male relatives (the only female patients in the literature).

Today, it is accepted that X-linked deafness with stapes gusher is associated with communication between the subarachnoid and perilymphatic spaces, probably caused by a deficiency or absence of the bony plate (the lamina cribrosa) separating the lateral end of the IAC from the basal turn of the cochlea (8). This communication provokes elevation of the perilymphatic pressure and stapes footplate fixation, with impairment in the conductive component of hearing. Even mild manipulation of the stapes during surgery can promote a stapes gusher in these patients. The increased perilymphatic pressure causes progressive cochlear nerve dysfunction,

leading to progressive sensorineural component hearing loss. The overall consequence is mixed progressive hearing loss with perilymphatic gusher during stapes surgery (7, 8).

In our case, bulbous dilatation of the fundi of the IAC and the absence of the bony plates separating the basal turn of the cochleas and IAC was demonstrated with CT. The labyrinthine segments of the facial canals appeared normal. By applying opaque material with lumbar puncture, the passage from subarachnoidal space into perilymphatic space was shown. Additionally, obliteration of the semicircular canals on the right side was demonstrated quite well with MRI. The obliteration of the semicircular canals at the right side was thought to be related to his recent meningitis. Although no family history was available, clinical history and imaging findings established the diagnosis of X-linked progressive deafness syndrome. With preoperative recognition of the syndrome, surgery which might precipitate perilymphatic stapes gushing was prevented, and the anatomic con-

figuration was decided to be inappropriate for cochlear implantation.

X-linked mixed hearing loss is a well-recognized but rare clinical syndrome. Most reported cases appear in the clinical literature, but radiologists may not be familiar with this entity. The recognition of the syndrome alters treatment and precludes stapedectomy (2). The radiologist should be aware of this entity and prevent harmful interventions that cause communication of the middle and external ear with the subarachnoid space and thus increase risk for meningitis. Also, the radiologist should be an essential part of the evaluation of candidates for cochlear implants; most individuals with this abnormality would be inappropriate candidates for this procedure.

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