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HRCT and MRI findings in X-linked non-syndromic deafness patients with a POU3F4 mutation

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ABSTRACT

Objective: The aim of this study was to analyze HRCT and MRI findings in patients with X-linked non-syndromic deafness and a POU3f4 mutation.
Methods: HRCT and MRI data of four patients (males, 2–19 years old) with a POU3f4 mutation were collected and a retrospective review was performed. Cochlea, internal auditory canal (IAC), vestibule, semicircular canals, vestibular aqueduct, nerve canals in the IAC fundus, stapes and cochlear nerve were evaluated on 2D images (multi-planner reformation, MPR) and cochlear foramen on 3D images (CT virtual endoscopy, CTVE). Ten cases with normal hearing subjected to CT and MR exams served as controls.
Results: Inner ear malformations were bilateral and symmetrical. Cochlear malformation was shown to consist of as a relatively normal outer coat shape, absence of a cochlear modilous, and a direct intercommunication between the IAC and cochlear inner cavity. The lateral portion of the IAC was dilated. A spiral cochlear inner cavity was observed with CTVE images versus a helical cochlear nerve foramen as seen in controls. The labyrinthine facial nerve canal and superior vestibular nerve canal were enlarged. The Bill's bar was hypertrophic and partially pneumatized. A thickened stapes footplate was present and a fissura ante fenestram was absent in seven ears examined. A column shaped stapes was observed in one ear.
Conclusions: The absence of a cochlear modilous with a dilated lateral IAC and thickened stapes footplate were the remarkable features observed with imaging these in X-linked non-syndromic deafness patients with a POU3F4 mutation. Preoperative recognition of the image features in these patients is important because it precludes stapedectomy and indicates the risks in the surgery of cochlear implantation including CSF gusher and electrode insertion into IAC.

1. Introduction

Currently, high resolution computed tomography (HRCT) remains the modality of choice for assessing congenital inner ear malformations. Helical multi-detector scanners not only provide a means for rapid imaging but also have the capacity to reconstruct inner and middle ear structures in any 2D plane or 3D images. Although bony structures of the inner and middle ear can be effectively displayed on HRCT images, the membranous labyrinth and the nerves of the internal auditory canal (IAC) can only be evaluated with magnetic resonance imaging (MRI). Accordingly, HRCT and MRI are often used as complementary approaches in the study of inner ear malformations [1].

It has been established that X-linked deafness is clinically and genetically a heterogeneous disease accounting for less than 2% of non-syndromic hearing loss. Two genes, PRPS1 (responsible for DFNX1) and POU3F4 (responsible for DFNX2), have been identified as contributing to this form of hearing impairment. Deafness caused by POU3F4 mutation accounts for 50% of all cases of X-linked non-syndromic hearing loss [2,3]. Inner ear malformations as specifically caused by POU3F4 mutation have only been reported in a few sporadic cases [2,4,5]. Most studies on non-syndromic hearing loss have focused on gene analysis, while data from imaging have rarely been incorporated within these studies. The purpose of this retrospective study is to present the HRCT and MRI findings of four patients with X-linked non-syndromic deafness possessing a POU3F4 mutation.

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2. Materials and methods

2.1. Study population

The institutional research ethics review board approved this retrospective study. HRCT and MRI data from four patients with a POU3F4 mutation were collected at our institute from July 2004 to September 2013. All patients were male and ranged in age from 2 to 19 years. Two of the cases with congenital sensorineural hearing impairment were from the same family, while the other two cases with congenital mixed hearing impairment were from different families. Gene diagnoses of all patients were performed at the medical genetics institute of Shandong University. The control group consisted of 10 subjects without hearing impairment subjected to identical temporal bone CT or MRI protocols for causes of temporal bone trauma or vertigo.

2.2. CT and MRI scan protocols

All subjects were subjected to multi-detector CT examination following routine procedures designed for patients suspected of having ear diseases. A 64-section multi-detector CT scanner (Somatom Sensation Cardiac 64, Siemens Medical Solutions, Germany) was used with these patients. The subject's head was placed in a neutral position, with no chin tilt, to approximate the Reid base line. Bilateral temporal bones were imaged in the original scan. Image acquisition and reconstruction parameters for the individual CT scanners were as follows: 120 kV, 150 mAs, 0.6 mm section thickness, 0.6 mm detector collimation, 0.5 beam pitch, 50% reconstruction interval and B70 reconstruction kernels, 250 mm field of view. MR scanning of all subjects was performed in a 1.5 Tesla MR unit (Siemens NOVUS Medical Systems, Germany). The parameters were as follows: 3D-FSE sequence, dual 3-in. surface coil, TR = 5000 ms, TE = 300 ms, FOV = 12 cm, matrix = 256 × 256, slice thickness = 0.5 mm.

2.3. Post-processing of data and image analysis

The images in DICOM format were transferred from a PACS system to a separate workstation (Siemens Wizard, Germany). The oblique planes of the cochlea (showing cochlear turns, the modilous, cochlear nerve canal and the IAC in the same image), semicircular canals (showing the entire course of the semicircular canal in one image), cochlear nerve (showing the cochlear nerve as it traversed the IAC and cochlear nerve canal) and stapes (showing the oval window and entire structure of the stapes) were reformed using the multi-planner reformation (MPR) technique. Vestibule, facial nerve canal, vestibular nerve canal and vestibular aqueduct were evaluated on axial and coronal plane images. 3D imaging of the cochlear nerve canal was reformed using the CT virtual endoscopy (CTVE) technique (threshold: low values of 850–1150 and high values of 3071). Three experienced clinicians evaluated these image findings and provided a conclusion of their assessment.

3. Results

All inner ear malformations were bilateral and symmetrical within each of the four cases. Stapes abnormalities in two patients were not symmetrical.

3.1. Cochlea

Similar types of cochlear malformations were present in the four patients (eight ears). CT images revealed that the outer coat of the cochlea had a relatively normal shape. Both interscalar septa between the basal/middle turn and middle/apical turn were present. Cochlear modiolus and bony spiral lamina were absent, which resulted in a cochlear spiral canal opening (Fig. 1). MR images revealed that a fluid filled cavity replaced the cochlear modiolus and spiral lamina (Fig. 2). No bony or soft septum were present between the cochlear inner cavity and IAC, as observed with both CT and MR. Such a finding indicated that a direct intercommunication existed between the cochlea and IAC.

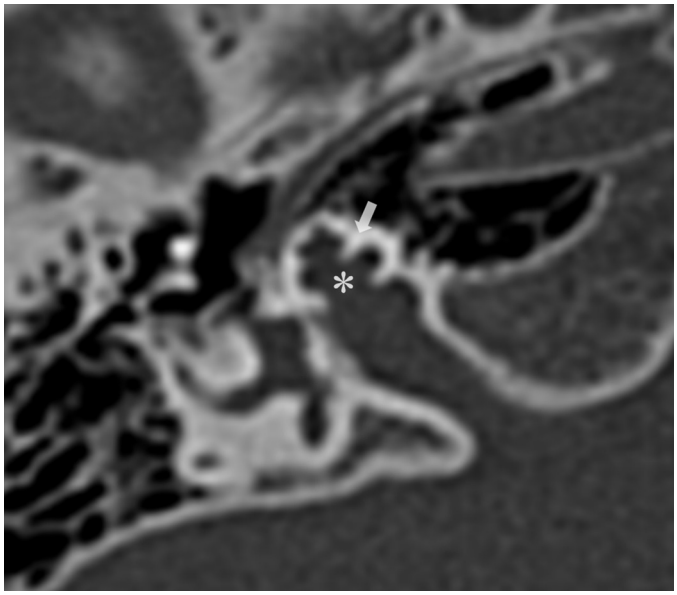

Fig. 1. A MPR CT image of right ear in one patient showing cochlea having a relative normal shape of outer coat (arrow), cochlear modiolus and bony spiral lamina being absent (*), a direct intercommunication existing between cochlea and IAC.

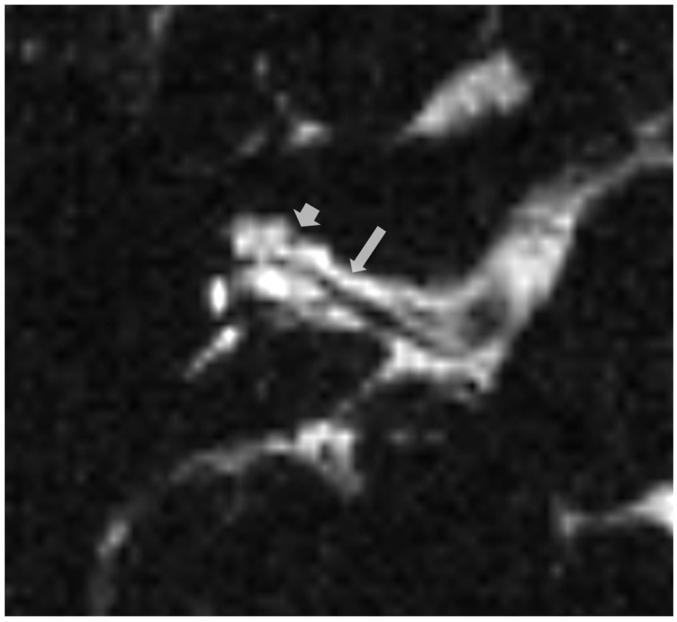

Fig. 2. A 3D-FSE MR image of right ear in one patient showing a fluid filled cavity replacing cochlear modiolus and spiral lamina (short arrow), cochlear nerve entering the inner cochlear cavity directly from IAC (long arrow).