ABSTRACT

Mondini dysplasia is a type of congenital cochlear anomaly. It is described as incomplete partition type II with large vestibular aqueduct. We report two cases of a young females presenting with unilateral sensory neural hearing loss, diagnosed by imaging with HRCT temporal bone and MRI of inner ear as Mondini dysplasia before the occurrence of meningitis. This study briefly discusses Mondini dysplasia a subtype of cochlear anomalies, its imaging features and complications.

KEYWORDS
inner ear anomalies, cochlear anomalies, enlarged vestibular aqueduct, cochlear incomplete partition type II

INTRODUCTION

Mondini dysplasia is a type of congenital cochlear anomaly which is also known as incomplete partition type II with large vestibular aqueduct. It was first described by Carlo mondini. Patients present with sensorineural hearing loss. With the improvements in imaging technology, congenital bony inner ear malformation, originally considered as a rare disease entity, now proven to be a common cause of pediatric deafness. Abnormalities of the temporal bone can be identified by use of high-resolution computed tomography (HRCT), in 20% to 30% of children with sensorineural hearing loss (SNHL).

Cases are now detected by HRCT temporal bone and MRI inner ear. We discuss two case of unilateral Mondini dysplasia and its imaging features.

CASE REPORT

Case 1

A 18 year old female presented with left sensorineural hearing loss and otorrhea. Evaluation in ENT department showed normal otoscopic findings. Weber test showed localization to contralateral right ear. Patient was referred to the radiology department for HRCT temporal bone. MRI inner ear was also done for the patient.

HRCT temporal bone showed unilateral left sided dysplastic cochlea with reduced turns, prominent vestibule along with left oto-mastoiditis. Absence of long process of incus, foot plate of stapes noted. Short process of incus was deformed and irregular and mildly enlarged. Left incomaleal joint was not visualized. [FIG: 1b]. Left semicircular canal showed dilated anterior limb, posterior limb was not visualized, lateral and posterior limb form common cavity with dilated vestibule. [FIG: 2] cystic appearance of geniculate (labyrinthine) part of left facial nerve region was seen suggestive of dehiscence. Right temporal bone was unremarkable, normal middle and inner ear.[FIG 1a]

MRI inner ear was also done with Philips Multiva 1.5T also confirmed presence of dysplastic cochlea and left semicircular canal [FIG: 3]. MRI showed cystic enlarged left cochlear vestibule with absent modiolus. [FIG: 4].

A diagnosis highly suspecting Mondini dysplasia was made. Patient was advised multichannel cochlear implantation. Both patients were not willing for surgery due to financial causes.

Case 2

A 21 year old female presented with left sensorineural hearing loss more specifically low frequency hearing loss. Evaluation in ENT department showed normal otoscopic findings. Weber test showed localization to contralateral right ear. Patient was referred to the radiology department for MRI inner ear. HRCT temporal bone was also done for the patient.

MRI inner ear was done with Philips Multiva 1.5T showed prominent basal turn, cystic conglomeration of middle and apical turns [FIG: 5], prominent vestibule and end olymphatic sac. [FIG: 6]. It also showed reduced number of cochlear turns on left as compared to right side. [FIG: 7].

HRCT temporal bone confirmed the findings of MRI and showed bilateral normal semicircular canals. [FIG: 8].

A diagnosis highly suspecting Mondini dysplasia was made. Patient was advised multichannel cochlear implantation.

Both patients were not willing for surgery due to financial causes.
DISCUSSION

The incidence of congenital hearing loss is 1 in 1000 newborns. Cochlear anomalies are a variety of congenital anomalies which may have different manifestations depending on the exact time at which an insult occurs during embryogenesis.

This entity was first classified in 1987 by Jackler et al. It has become widely accepted (with various modification), which divides congenital cochlear anomalies according to the timing of the developmental arrest. Starting from 3rd week till the end of 7th week of gestation, an insult during each week results in a distinct inner ear abnormality. Insult in 3rd week results in complete labyrinthine aplasia or Michel deformity, 4th week in cochlear aplasia, early 5th week in common cavity malformation to the cochlea and vestibule, late 5th week in cochlear incomplete partition type I including cystic cochleovestibular anomaly, 6th week in cochlear hypoplasia, 7th week in cochlear incomplete partition type II including Mondini dysplasia.

Patients have sensorineural hearing loss, which is usually bilateral; in both our cases it was unilateral. High-frequency hearing is generally preserved as the basal turn of the cochlea is intact.

Most affected individuals have profound sensorineural hearing loss, but some may have residual hearing. There have also been reports of affected individuals having normal hearing. Mondini dysplasia can also predispose to recurrent meningitis because the defect can act as a "port of entry" to cerebrospinal fluid. Many individuals are not diagnosed before several episodes of recurrent meningitis occur.

The pathogenesis is thought to result from a relatively late insult during the 7th week of embryological development when most of the inner ear has already formed. Some suggest retinoids (vitamin A) or other factors a fetus may be exposed to early in pregnancy have contributed to some cases of isolated MD (occurring with no other abnormalities). Mutations in the SLC26A4 gene cause Pendred syndrome which is associated with Md7.

Instead of the expected 2.5 there are only 1.5 turns to the cochlea. Sac-like cochlea seen due to failure of formation of the interscalar septum between the middle and apical segments.

The Mondini abnormality consists of a triad -abnormal cochlea with only 1.5 turns (instead of the normal 2.5 turns) and normal basal turn with a cystic apex in place of the distal 1.5 turns, enlarged vestibule with usually normal semicircular canals, enlarged vestibular aqueduct containing a dilated end olympic sac.

Genetic testing are available for Mondini dysplasia if it is associated with a specific syndrome for which genetic testing is available, or if a
mutation has previously been identified in an affected individual in the family. Unfortunately, there is no clinical genetic testing available for many cases of isolated Mondini dysplasia.

Role of radiologist is in diagnosis and pre-operative assessment for cochlear implantation and for prompt identification of complications. HRCT temporal bone makes diagnosis possible by its high resolution, visualization of the cochlear anatomy which in Mondini dysplasia includes identifying number of turns, presence of basal turn and vestibular anatomy which in Mondini dysplasia includes enlarged vestibule and separation from cochlea. CT also helps in visualization of semicircular canal anatomy which are usually normal. In one of the two cases we had unilateral dysplastic semicircular canals. Associated narrowing or atresia of oval and round window can be visualized by CT. Facial canal can be assessed by CT.

MRI plays a role in confirming the CT findings along with visualization of vestibular aqueduct and endolympathic sac dilation, cochlear nerve and facial nerve course, facial nerve dehiscence. Better visualization of peri-lymphatic fistula.

Complete or partial absence of the normal interscalar septum is also present, which can now be demonstrated on MRI.

Associations include thalidomide and rubella embryopathies as well as a number of syndromes such as Pendred syndrome which is bilateral sensorineural hearing loss and goiter, CHARGE syndrome, Klippel-Feil syndrome, DiGeorge syndrome and Wildervanck syndrome.

Complications include meningitis and perilymphatic fistula. Both our patients did not develop complications.

A perilymph fistula communicating between the subarachnoid space and the middle ear is common in patients with Mondini dysplasia. CSF otorrhea and rhinorrhea may occur in such patients, but there is no CSF otorrhea in those with an intact tympanic membrane.

One patient presented with otorrhea but MRI showed no perilymphatic fistula, otorrhea was due to concurrent otomastoiditis.

If an individual has residual hearing, hearing amplification aids may be useful. Hearing can be improved by implantation of a multichannel cochlear implant. Treatment options include surgical repair of the defect to prevent recurrent meningitis. Prophylactic antimicrobial therapy to prevent infection and conjugate pneumococcal vaccination are also useful.

CONCLUSION

Mondini dysplasia is a rare cause of hearing impairment, and delay in diagnosis may lead to recurrent meningitis and neurologic sequelae. For children with unilateral sensorineural or mixed-type hearing impairments, temporal bone computed tomography should be arranged before the occurrence of CSF rhinorrhea, CSF otorrhea, or recurrent meningitis. Early detection and treatment are valuable, since these can prevent the occurrence of meningitis. Cochlear anomalies are being detected more frequently at high-resolution computed tomography and MRI inner ear. Recognizing the imaging features of these abnormalities is necessary for diagnosis and surgical planning for multichannel cochlear implantation.

REFERENCES