CASE REPORT

SIMULTANEOUS PRESENTATION OF MICHEAL AND MONDINI MALFORMATIONS IN A SINGLE PATIENT; REPORT OF A RARE CASE

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ABSTRACT

Autosomal-recessive genes account for over 80% of non-syndromic deafness patients, with a large proportion of those resulting in cochlear disease. In our case, on the left, there was a total lack of all inner ear structures, including the cochlea, semicircular canals, vestibular and cochlear aqueducts, resulting in Michael deformity/full labyrinthine aplasia. However, cochlea abnormally has 1.5 turns: a basal turn and a cystic apex with an expanded vestibule, resulting in the mondini deformity on the right. Pakistan has a high frequency of inherited pre-lingual deafness due to the strong cultural practice of consanguineous marriages and a lack of information regarding screening procedures.

Keywords: Ear Malformations, Micheal Malformation, Mondini Malformation, CT, MRI, Radiology.

Introduction

Malformations can affect the outer ear, middle ear, and inner ear, often in combination. Inner ear anomalies, resulting from disrupted embryological development, include aplasia, hypoplasia, and labyrinthine malformations. Vestibular aqueduct variations are common, while cochlear aqueduct malformations are rare. Vestibuloacoustic ganglion cell reduction is prevalent in inner ear malformations, and the internal auditory canal may also be affected, leading to displacement of arteries and nerves, particularly the facial nerve.[1]

The prevailing etiology of neurosensory hearing loss is congenital inner ear abnormalities. Michel aplasia, characterized by the failure in otic placode development preceding the third gestational week, results in the absence of the cochlea, vestibule, and semi-circular canals. Michel aplasia exhibits no substantial gender predilection and is observed with comparable frequency in both genders.[2]

Mondini dysplasia was initially characterized as cochlear malformation featuring a reduction to 1.5 cochlear turns, alongside intact semi-circular canals, an inclined vestibular aqueduct, and an expanded endolymphatic sac. Now Mondini has been broadly utilized as an encompassing descriptor for diverse congenital cochlear deformities. The subtypes of Mondini-like dysplasia encompass Type A, defined by a two-turn cochlea with a fully developed bone at the modiolar base. Type B, on the other hand, involves 1.5 to 2 cochlear turns, featuring either the absence or hypoplasia of the bony wall at the modiolar base, potentially with or without communication between the cochlea and internal auditory canal while maintaining a complete basal turn.[3]

Case Presentation

We present a case of 3 years old female child, born via SVD with immediate cry. She presented with complaint of deafness since birth. BERA (brainstem evoked response audiometry) showed bilateral

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profound degree of sensorineural hearing loss. ABR (auditory brainstem response) and ASSR (auditory steady state response) showed bilateral profound degree of sensorineural hearing loss. She had never used hearing aids. There was no history of ear discharge. No significant perinatal or neonatal history was reported. Physical examination showed bilateral intact tympanic membranes. CT ear petrous bone without contrast was performed which showed Mondini malformation on right with abnormal cochlea having 1.5 turns; basal turn and a cystic apex with enlarged vestibule. Horizontal semicircular canal was normal, however, superior and posterior semicircular canals were fused to form a single semicircular canal. Endolymphatic sac was dilated. There was Michael deformity/complete labyrinthine aplasia on left with complete absence of inner ear structures including cochlea, semicircular canals, vestibular and cochlear aqueducts. MRI ear/internal auditory meatus without contrast was performed which showed Mondini malformation on right with normal seventh eighth cranial nerve complex. There was Michael deformity on left with non-visualization of cochlea, vestibule and semicircular canals. Left internal auditory canal was small and hypoplastic. Vestibulocochlear nerve complex was also small. Bilateral cochlear implantation was done and patient was discharged. Thus, it is a very rare till date in which simultaneous presentation of both Mondini and Michael malformations were seen in a single patient (Fig.1,2 and 3).

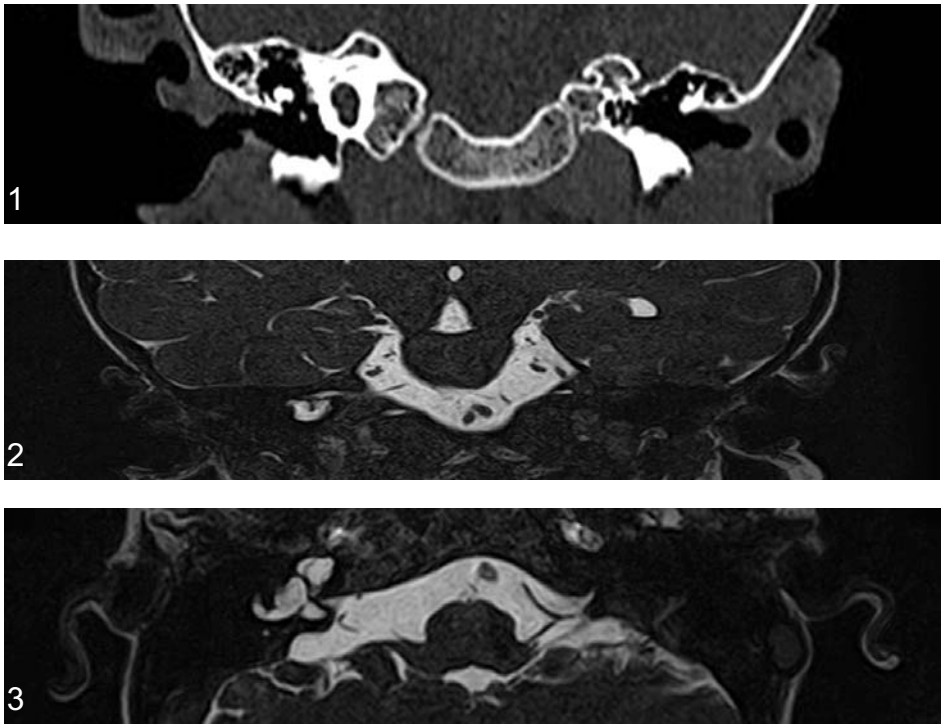

Figure 1,2,3: CT and MRI axial and coronal images showing Michael and Mondini malformations.

Discussion

Neurosensory hearing loss is most commonly caused by congenital inner ear abnormalities.[4] The cause of inner ear aplasia, also known as Michel aplasia, is the otic placode's inability to mature by the third week of pregnancy.[5] As the name implies, Michel initially discovered this aberration while reviewing the autopsy of a 12-year-old kid who had a history of congenital deafness.[6] For its diagnosis, temporal bone CT and MRI is helpful. Neural sensory hearing loss is most commonly caused by congenital anomalies of the inner ear. The term "Michel inner ear deformity" describes the complete aplasia of the inner ear, which is an uncommon developmental defect. It occurs on by an early developmental stop of the otic placode in

the third week of pregnancy.[2] Around the third week of pregnancy, otic placodes emerge from the surface of the ectoderm on each side of the rhombencephalon, signaling the development of otocysts. The cochlea and vestibule follow the diverticulum that buds from the otocysts in the fifth week to generate the endolymphatic sacs. By the end of the sixth week, the membranous cochlea has completed one to 1.5 turns, and by the end of the seventh week, it has completed 2.5 turns. During the seventh or eighth week of pregnancy, the utricle segments of the otocysts begin to grow into the semi-circular canals. Prior to the lateral canals forming, the posterior canals develop first. By the conclusion of the eighth week, the inner ear structures have fully developed into adult forms.[7]
The most severe and uncommon kind in these classifications mentioned by Sennaroglu et al. and Jackler et al., Michel aplasia, is caused by the absence of otic placodes at the onset of development.[7,8] There is no vestibule, cochlea, or semicircular canal. Michel aplasia does not significantly differ in appearance across genders and can affect both boys and girls. Predisposing variables include prenatal CMV infection, thalidomide exposure, and genetic disorders.
A short, flat cochlea, a big vestibule, broad, tiny, or absent semicircular canals, and immature sensorineural tissues are characteristics of Mondini dysplasia. It can happen alone or in combination with abnormalities in other organs. There are situations when the stapes footplate is faulty, which can result in meningitis and spontaneous perilymphatic fistula.[9] About 50% of instances with cochlear abnormalities are of this kind, making it the most prevalent. Following the postmortem examination of the 8-year-old child who had profound hearing loss in 1791, Mondini documented this anomaly. Instead of the typical 2‰ turns, the cochlea has 1‰ turns. The three parts of classic MD are the massive vestibular aqueduct, dilated vestibule, and cystic apex. There is development only in the base turn; the higher turns create a shared cavity. It is consistent with an incomplete cochlea division, according Jackler et al. Reduced spiral ganglion cell count can cause hearing loss that progresses from mild to extreme deafness.[10]
Patients with congenital neurosensory hearing loss benefit from CT because it ensures scanning of the otic capsule and bone labyrinth. Due to its excellent resolution, anatomical intricacy, and capacity to acquire extremely thin slices, multi-detector CT is a valuable diagnostic tool. In cases of Michel and mondini malformations, CT results are diagnostic. It shows up as flattening of the middle ear medial wall, constriction of the internal auditory canal, and lack of the vestibule, semicircular canals, and cochlea, which are inner ear structures.[11] Magnetic resonance imaging (MRI) is another radiaology based tool used to assess cochlear implant. The absence of ionizing radiation is a significant benefit, particularly for younger users. The diagnosis is made possible by the internal auditory canal's stenosis, the lack of the seventh and eighth cranial nerves, and the seventh cranial nerve's aberrant MRI path.[12]
Thus, simultaneous presentation of these deformities are rarely seen and are associated with many neurological deficits. Timely imaging based diagnosis can save patient from morbidity.

Conclusion

The most frequent cause of neurosensory hearing loss is a congenital inner ear defect, and Michel aplasia is defined by a whole lack of inner ear while Mondini deformity shows cochlear hypoplasia. The results of cross-sectional imaging are used to make the diagnosis. Concurrent these two abnormalities in a single patient are rarely reported till date.

Conflict of Interest: None

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