CASE REPORT

IMAGING APPEARANCES IN A CASE OF INNER EAR APLASIA

Awais Ansari , Imtiaz Ali, Raisa Altaf, Ramsha Fatima, Rabia Ahmed , Bushra Shamim

Department of Radiology, Liaquat National Hospital and Medical College, Karachi, Pakistan.

PJR April - June 2023; 33(2): 97-100

ABSTRACT

Congenital inner ear abnormality is a common cause of sensory neural hearing loss. Michel anomaly is also known as inner ear aplasia and defined as congenital absence of inner ear. This congenital anomaly is secondary to failure of development of the otic placode during gestational period before 3rd week of pregnancy. We present the radiological imaging findings of Michel anomaly in a 04-year-old male patient who came in the ENT outdoor patient department with history of bilateral hearing loss.

Key words: Inner ear aplasia, Michel anomaly, Hearing loss, Sensory neural.

Introduction

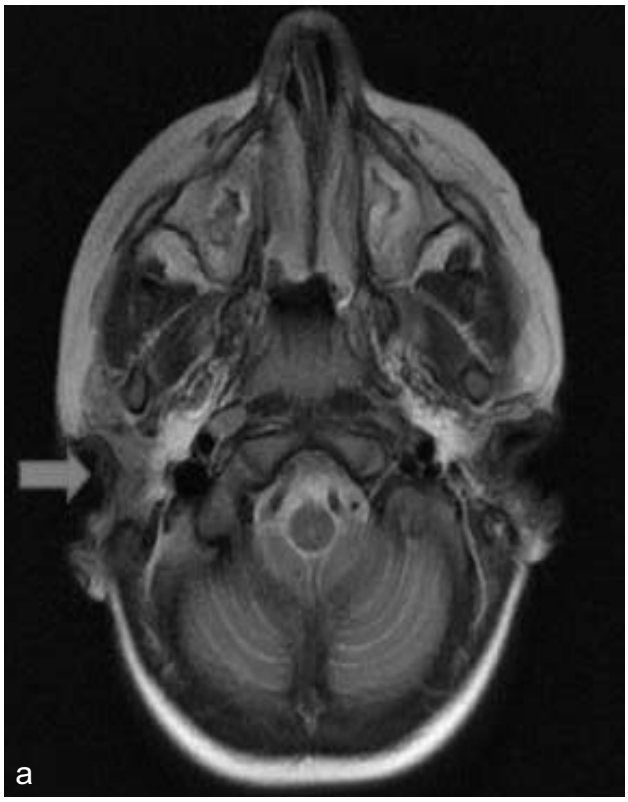
Michel anomaly is also called complete labyrinthine aplasia (CLA).[1,2] It is a severe congenital abnormality of the inner ear defined as total absence of inner ear structures.[1,3] This uncommon congenital abnormality of inner ear was first reported by Michel in 1863 and only few cases have been reported in literature.[1,2,3] Complete labyrinthine aplasia (CLA) or Michel anomaly occurs due to failure of development of otic placode (ectoderm) before 3rd week of gestation.[3,4,5] Other skull abnormalities can be associated with this anomaly including platybasia, hypoplasia of petrous bone, and aberrant course of jugular vein, facial nerve.[2] Multi-detector CT scan with thin slices is a very effective diagnostic radiological imaging investigation for inner ear bony structures.[4] Temporal bone MRI can also be useful and diagnostic imaging investigation for soft tissue of inner ear structure.[4] The management of such cases is by hearing aids, counselling, implants.

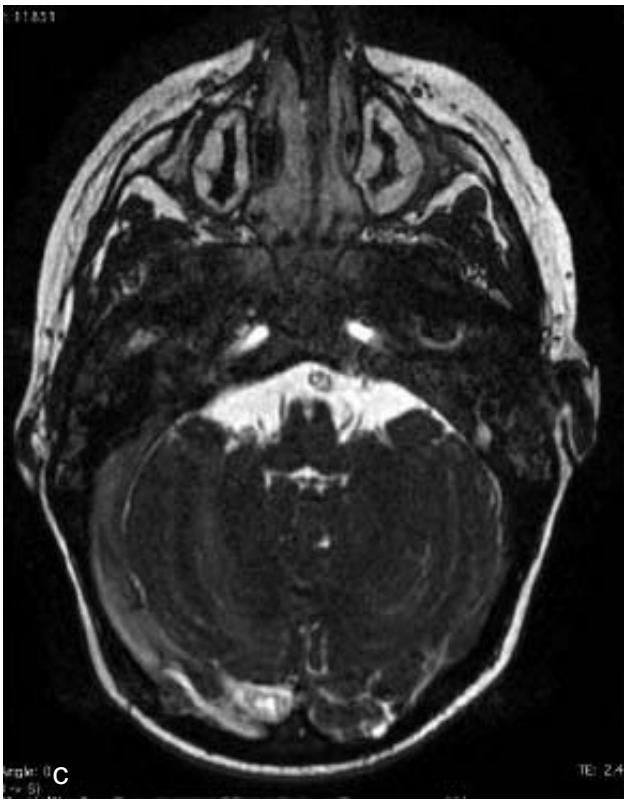
Case Report

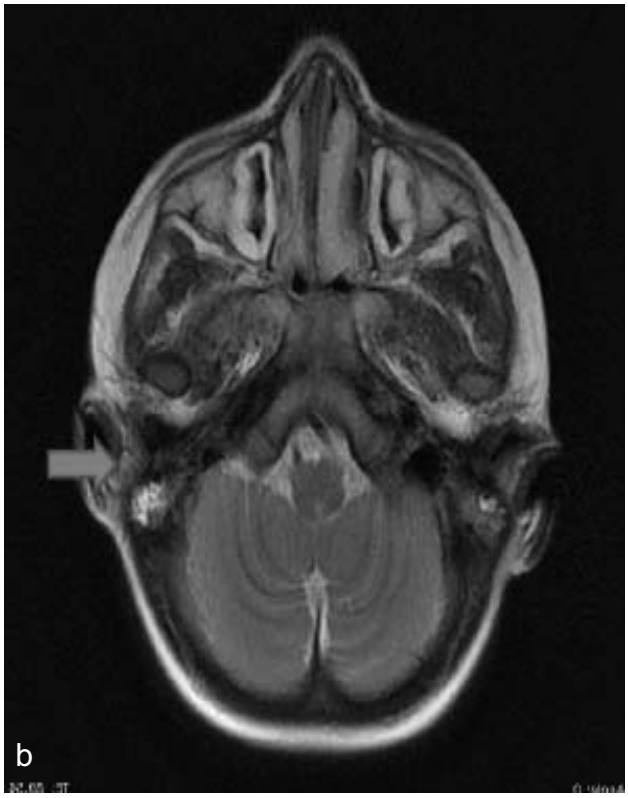
Our patient was a 4-year-old boy born at term via a normal delivery with no significant complications during the prenatal or postnatal period. The parents had no history of consanguinity or hearing loss in their family. The child had no significant medical or surgical history. On examination, the child appeared healthy and had normal growth and development. However, there was no response to any sounds or verbal stimuli. There was no sign of any external ear abnormalities or craniofacial anomalies. MRI scan of the head and temporal bones was performed, which revealed the absence of bilateral semicircular canals, vestibules, and facial vestibulocochlear nerve complexes, consistent with inner ear aplasia (Fig.1a,b, c,d,e). The family was counseled regarding the disease and treatment options were explained however due to financial constraints they were lost to follow up. (Fig.1a,b) shows axial T2 images with non visualization of inner ear structures including cochlea, vestibule, semicircular canals and vestibular and cochlear aqueducts bilaterally. (Fig.1,c,d) show similar finding in FEISTA sequeance (Fig.e) coronal image showing absence of inner ear bony structures.

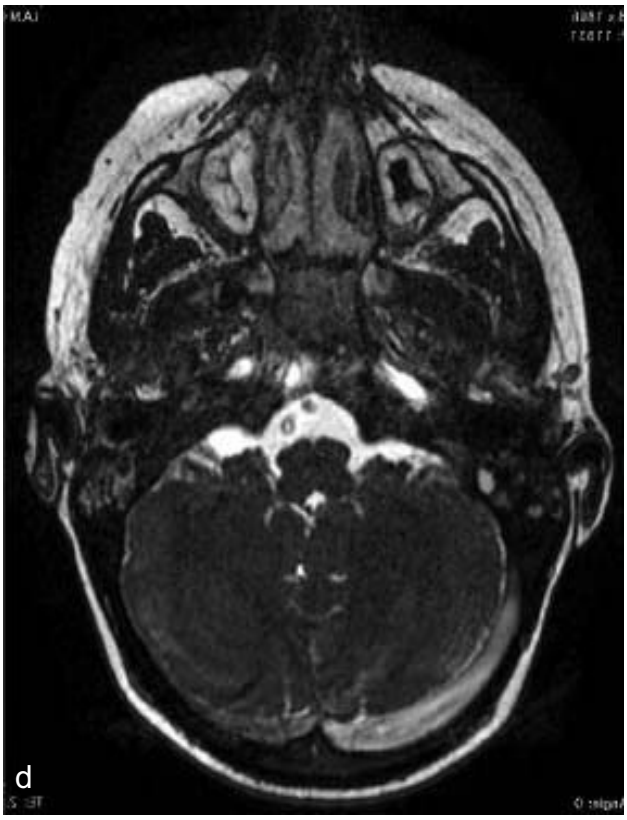
Correspondence : Dr. Awais Ansari
Department of Radiology,
Liaquat National Hospital and Medical College,
Karachi, Pakistan.
Email: ansarimawais791@gmail.com

Submitted 19 May 2023, Accepted 29 May 2023


a


c


b


d

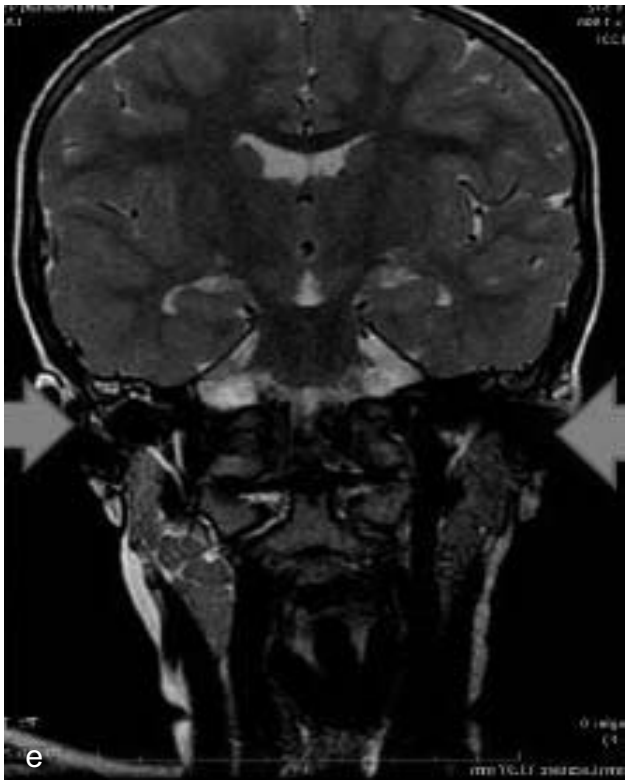


Figure 1abcde: Axial T2 **(ab)**, axial fiesta **(cd)**, coronal T2 **(e)** images show absence of bilateral semicircular canals, vestibules, and facial vestibulocochlear nerve complexes, consistent with inner ear aplasia

Discussion

Inner ear aplasia was first described in the medical literature in 1867 by the German physician.[1,2] Since then, numerous studies have been conducted to better understand the condition and its underlying causes. In 1978, Michel, Keeney. Marsot-Dupuch, and Daneshi all reported the same finding. While not explicitly stated in the manuscript, Hersh and Gross. also presented figures in their case reports that illustrated this finding.[1] This classification system has been widely used in subsequent studies. In the 1990s and 2000s, advances in imaging technology, such as high-resolution CT and MRI, enabled more detailed and accurate diagnosis of inner ear aplasia. These studies revealed that inner ear aplasia is often associated with other malformations, such as anomalies of the temporal bone and facial nerve, and that there is significant variability in the severity and presentation of the condition. Recent studies have focused on the genetic basis of inner ear aplasia. In 2012, a study published in the American Journal of Human Genetics identified mutations in the GATA3 gene as a cause of inner ear aplasia and associated hearing loss.[6] This discovery has helped to advance our understanding of the molecular mechanisms underlying inner ear development and has potential implications for genetic counseling and treatment strategies.

Inner ear aplasia is a rare congenital condition in which the inner ear structures fail to develop properly during fetal development.[1,2,3] This results in a lack of sensory cells and nerve fibers in the inner ear, leading to profound hearing loss or deafness.[2,3,4] Inner ear aplasia can occur in isolation or as part of a syndrome that affects other organs and tissues.[1]

Patients with inner ear aplasia typically have severe hearing loss or deafness and may experience balance problems due to the absence of the vestibular system, which is responsible for maintaining balance and orientation.[1,2,3,4] As discussed previously discussed in our case there is non-visualization of bilateral semicircular canals, vestibules and facial vestibulocochlear nerve complexes representing inner ear aplasia

Specifically, the CT and MRI images will show a complete lack of the bony labyrinth of the inner ear, with no visible cochlear capsule, modiolus, or internal auditory canal.[1] The vestibule and semicircular canals are also absent, with no visible structures or air-filled spaces in their expected locations. In addition to these imaging findings, patients with Michel's anomaly may also present with other symptoms, such as profound sensorineural hearing loss, vestibular dysfunction, and a high risk of developing meningitis. These symptoms can be used to support the diagnosis of Michel's anomaly in conjunction with the CT findings. MRI can also be used to evaluate the integrity of the eighth cranial nerve,[1] which is responsible for transmitting sound and balance information from the inner ear to the brain. In patients with Michel's anomaly, the eighth cranial nerve may be absent or severely hypo plastic or absent, reflecting the absence of the inner ear structures.[1,2]

It is important to note that MRI may not always be necessary for the diagnosis of Michel's anomaly, as CT imaging is often sufficient to identify the characteristic findings.[2,3,4,5] However, MRI may be useful in certain cases where additional information is needed or when other congenital malformations of the inner ear are suspected.

Unfortunately, there is no cure for Michel's anomaly, and the condition typically results in profound sensorineural hearing loss and vestibular dysfunction.[2,3,4,5] Treatment options for Michel's anomaly are focused on managing the symptoms of the condition and may include:

1. Hearing aids: Individuals with Michel's anomaly may benefit from the use of hearing aids to amplify sound and improve communication abilities.

2. Cochlear implants: In severe cases of Michel's anomaly, cochlear implants may be recommended. Cochlear implants are surgically implanted devices that bypass the damaged inner ear structures and directly stimulate the auditory nerve.[3,4]

3. Vestibular rehabilitation: Individuals with Michel's anomaly may experience balance issues due to the absence of the vestibular system. Vestibular rehabilitation exercises can help improve balance and reduce the risk of falls.[1,2,3,4]

4. Speech therapy: Speech therapy can be beneficial for individuals with Michel's anomaly who have difficulty with speech and communication due to hearing loss.

5. Management of associated conditions: Individuals with Michel's anomaly may be at an increased risk of developing other conditions, such as meningitis. Therefore, appropriate vaccinations and regular monitoring for any signs of infection are important.

It is important to note that the management of Michel's anomaly should be individualized based on each patient's unique needs and circumstances. A multidisciplinary approach involving an audiologist, otologist, and other healthcare professionals may be necessary to provide comprehensive care.[1,2,3,4]

Conclusion

Michel anomaly is very rare congenital abnormality of ear and most common cause of neurosensory hearing deafness and characterized by total absence of inner ear structures, diagnosed on radiological imaging findings. Radiologist should aware of imaging findings and its congenital other associations and convey the diagnosis to clinician so that he can counsel the patient s parents regarding treatment options and outcome.

Conflict of Interest: Declared none.

References

1. Ozgen B, Oguz KK, Atas A, Sennaroglu L. Complete labyrinthine aplasia: clinical and radiologic findings with review of the literature. AJNR Am J Neuroradiol. 2009; **30(4):** 774-80.

2. Marsot-Dupuch K, Dominguez-Brito A, Ghasli K, Chouard CH. CT and MR findings of Michel anomaly: inner ear aplasia. AJNR Am J Neuroradiol. 1999; **20(2):** 281-4.

3. Umul A, Demirtas H, Celik AO. Radiological Findings of Michel Aplasia. Acta Inform Med. 2016; **24(3):** 215-7.

4. Daneshi A, Farhadi M, Asghari A, Emamjomeh H, Abbasalipour P, Hasanzadeh S. Three familial cases of Michel's aplasia. Otol Neurotol. 2002; **23(3):** 346-8.

5. Giesemann AM, Goetz F, Neuburger J, Lenarz T, Lanfermann H. From labyrinthine aplasia to otocyst deformity. Neuroradiology. 2010; **52(2):** 147-54.

6. van der Wees J, van Looij MA, de Ruiter MM. Hearing loss following Gata3 haploinsufficiency is caused by cochlear disorder. Neurobiol Dis. 2004; **16(1):** 169-78.