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A Case Study

**BILATERAL POSTERIOR AND LATERAL SEMICIRCULAR CANALS
APLASIA ASSOCIATED WITH OTHER INNER EAR
MALFORMATION IN A 2-YEAR-OLD CHILD:
A RARE CASE REPORT**Fatimah Albazroun^{1*}, Zahrah Almoumen², Hazim Al-Eid³¹ King Fahad Specialist Hospital-Dammam, Saudi Arabia² Department of ENT, King Fahad Specialist Hospital-Dammam, Saudi Arabia³ Department of ENT, King Fahad Specialist Hospital-Dammam, Saudi Arabia**Abstract**

Inner ear malformation accounts for approximately 20% of reported cases of congenital sensorineural deafness. It results from developmental arrest in different stages of embryogenesis. Aplasia of one of the semicircular canals with or without cochlear malformation is well known and has been reported in journals. However; to our knowledge, aplasia of two semicircular canals have been rarely reported, and for this reason the clinical reflection of this anomaly is poorly understood. This case will be hopefully used to gain an in-depth understanding of illness and help in the diagnosis and proper management of future cases. In this paper we present a report concerns a 2-year-old boy with no family history of hearing loss, presenting with bilateral profound sensorineural hearing loss with no other congenital anomalies. CT scan and MRI shows dysplastic bilateral vestibules, bilateral hypoplastic cochlea and bilateral lateral and posterior semicircular canal (SCC) aplasia.

Keywords: Semicircular canal aplasia, sensorineural hearing loss, CT scan, cochlear hypoplasia**Corresponding author:****Fatimah Albazroun,**

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INTRODUCTION:

Inner ear in embryo starts to develop during 3rd week of gestation. A structure called ‘otic placodes’ which arise from the surface ectoderm on each side of the rhombencephalon represent the fundamental embryological part of inner air developmental. The otic placodes subsequently invaginate and form otocysts. Diverticulum buds from the otocysts form the endolymphatic sacs, which eventually by the 5th week; form the cochlea and vestibules, while utricle segments of the otocysts form the semicircular canals by the end of the 7th week. The Superior semicircular canal develops first while the lateral develops last [1]. Premature arrest in different parts of development leads to inner ear malformation. The earlier the arrest the more severe the deformity [2]. Morphologically inner ear malformation is classified into two categories; membranous malformations which involve the inner ear hair cells and therefore can't be diagnosed based on CT or MRI imaging of the temporal bone. And bony labyrinth malformation which can be radiologically demonstrated [3].

CASE REPORT:

We report the case of one of the non-identical twin brothers: a 2-year-old boy, born of a consanguineous marriage to medically free parents, was first seen and diagnosed in our institution when he was 1-year-old to have bilateral profound sensorineural hearing loss (SNHL). His hearing loss was first noticed by his mother when he was five months of age. She described him to be a ‘Hard of hearing’ that even loud sounds fail to startle him compared to his twin brother. Since the diagnosis, the patient has been in regular follow-up at our ENT outpatient department. His parents had not reported any active complaints about him up until he started walking, they stated that he had unsteady gait and poor balance which usually followed by Falls or in some occasions ‘a fainting episode’ which they have never

noticed with his twin brother. Other than speech delay; the patient had displayed normal developmental milestones appropriate to his age. He was born at term by lower segment caesarean section (LSCS) with birth weight of 2.4 kg. Apart from gestational induced hypertension, his mother had uneventful pregnancy. His parents reported no family history of hearing loss, he has 7 healthy siblings.

Examination of the face showed frontal bossing and depressed nasal bridge. Right ear, nose and throat examination were normal, while left ear examination revealed otitis media with effusion. Tympanogram was performed; it revealed type B with normal ear canal volume (ECV) bilaterally. ABR was done, it showed bilateral unidentifiable repeatable (I,iii,v) waveforms at 90 dBHL and low wave V was traced down to 20 dBHL. Radiological examination computerized tomography (CT) scan of temporal bone showed bilateral aplasia of the posterior semicircular canal and lateral semicircular canals, dysplastic bilateral vestibules (**figure 1**) and bilateral cystic fusion of the middle and apical turns of the cochlea (**figure 2**). Magnetic resonance imaging (MRI) confirmed CT scan finding, and additionally showed normal cochlear nerve bilaterally. In addition to that, patient completed vestibular evoked myogenic potential (CVEMP) in a seated position in response to tone burst stimuli 800 Hz frequency with a two cycle rise/fall and no plateau (black mam gated) it exhibited lowered threshold on left side compared to right side (80 dBnHL versus 95 dBnHL) with no significant intraaural asymmetry ratio 25% to the left. Molecular genetic analysis of the patient detected the heterozygous variants c.362T>A p(Val121Glu) and c.3467G>A p(Cys1156Tyr) in the OTOG and OTOGL genes, respectively. Additionally, potentially relevant variants were detected; they are reported in the table below (**table1**)

Table 1. Potentially Relevant Variants

Variant (HGVS)	Protein (HGVS)	Zyg	Reported (literature/DB)	GnomAD	Classification
NM_004525.2(LRP2):c.1973A>G	p.(Tyr658Cys)	Het.	ClinVar:332195	0.089%	VOUS
NM_001145026.1(PTPRQ):c.6568G>T	p.(Ala2190Ser)	Het.	DbNSP: rs199746225	0.035% 64 het.; 2hom	VOUS
NM_0033056.3(PCDH15):c.2990A>G	p.(Glu997Gly)	Het.	ClinVar: 46456	0.032%	VOUS
NM_000091.4(COL4A3):c.1863A>G	p.(Gln612=)	Het.	DbNSP: rs771390525	0.022%	VOUS

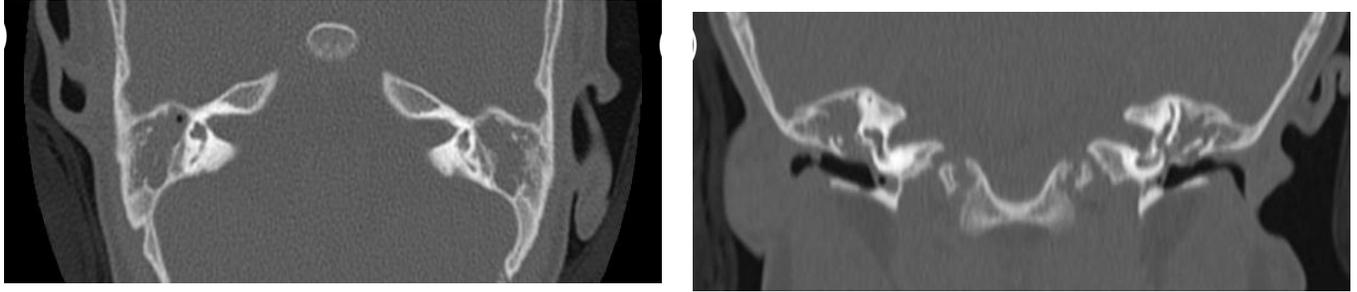


Figure 1. High resolution computed tomography (a) axial view, showing aplasia of the lateral semicircular canal and dysplastic vestibule bilaterally. (b) coronal view.

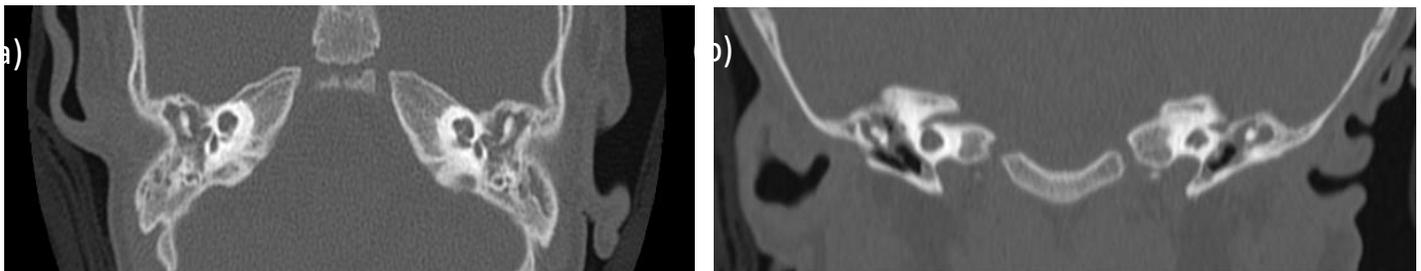


Figure 2. High resolution computed tomography (a) axial view, cystic fusion of the middle and apical turns of the cochlea bilaterally. (b) coronal view.

DISCUSSION:

Inner ear malformations accounts for 20% of cases that present with congenital hearing loss [9]. Jacker et al classified bony labyrinths malformation in 1987 [6], where he grouped inner ear malformation into two categories; category A which contains malformations associated with aplasia of the cochlea. category B; which contains malformation associated with normal cochlea [4]. The classification is illustrated in the table (**Table 2**).

Table 2. Classification of inner ear malformations according to Jackler [4]

Category A	Aplasia or malformation of cochlea	
	vestibule and semicircular system	1. Labyrinthine aplasia (Michel deformity) 2. Aplasia of cochlea, normal or deformed
	deformed vestibule and semicircular system	3. Hypoplasia of cochlea, normal or deformed
	vestibule and semicircular system	4. Incomplete cochlea, normal or deformed
	build a common space without internal architecture, normal or deformed semicircular system	5. Common cavity: cochlea and vestibule
	Note: enlarged vestibular aqueduct possible.	
Category B	Normal cochlea	
	1. Dysplasia of vestibule and lateral semicircular canal, normal anterior and posterior semicircular canal.	
	2. Enlarged vestibular aqueduct and normal or dilated vestibule, normal semicircular system.	

Cochlear Hypoplasia which accounts for 15% of cochlear malformations define as; ‘cochlea with 1.5 turns’ where the dimensions of the cochlea are less than those of a normal cochlea with other various architecture deformities [7]. There are Four types of cochlear hypoplasia:

- a- CH-I (Bud-like cochlea)
- b- CH-II (Cystic hypoplastic cochlea)
- c- CH-III (Cochlea with less than 2 turns)
- d- CH-IV (Cochlea with hypoplastic middle and apical turns)

Most patients with cochlear hypoplasia have severe to profound hearing loss, which make them candidates for cochlear implant if cochlear nerve was normal. In case of bilateral hypoplasia, it’s important to know which ear has less malformation and larger cochlear nerve. Intraoperatively, facial nerve malposition is anticipated finding as hypoplasia of the cochlea are usually associated with semicircular abnormalities. Surgeons are usually using thin and shorter electrodes to insert in the cochlea as the numbers of turns are smaller and narrower. Unfortunately, Patients with cochlear hypoplasia are at higher intraoperative risks, such as cerebrospinal fluid leakage, meningitis, and electrode displacement [6]. Other inner ear abnormalities such as semicircular canals aplasia is not that common compared to dysplasia. It’s usually associated with abnormal course of the facial nerve, atresia of the oval window, and abnormal ossicles [7]. Aplasia of all semicircular canals can be seen in patients with CHARGE syndrome [7,8] while isolated aplasia of the posterior semicircular canal is seen in patients with Waardenburg syndrome and Alagille syndrome [7]. CT scan is the modality of choice to diagnose semicircular canal aplasia [7].

CONCLUSION:

In conclusion, this is a case of a 2-year-old patient with bilateral posterior and lateral semicircular canals aplasia with cochlear hypoplasia and vestibular dysplasia, apart from the inner ear malformation, no

other congenital anomalies, patient underwent uneventful cochlear implantation surgery. From our side, we recommend that more patients with similar finding should be examined and investigated by genetic analysis for better delineation of this disorder and understanding of the cause.

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