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Case Report

Ear, Nose and Throat Section

Unusual Audiologic Profile in Unilateral Renal Agenesis: A Case Report

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ABSTRACT

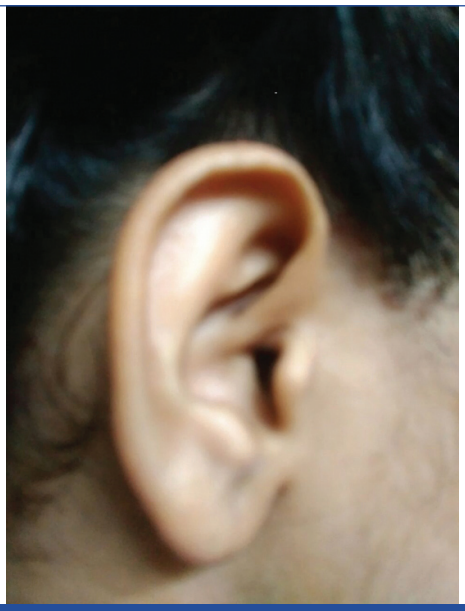
Unilateral Renal Agenesis (URA) is characterised by the absence of one kidney, and typically manifests asymptomatically due to compensatory hypertrophy of the remaining kidney. The condition is often detected incidentally because routine screening for renal anomalies is not universal globally. URA is generally associated with mixed or sensorineural hearing loss. A case involving contralateral conductive hearing loss is discussed in the present case report. The present case involves a eight-year-old female with isolated URA and no other concurrent abnormalities. The present report highlights an atypical audiological profile in URA, diverging (conductive type of hearing loss) from the commonly observed sensorineural or mixed hearing loss patterns. Understanding such variations contributes to the broader knowledge of the genetic and developmental links between the auditory and renal systems. This case underscores the importance of comprehensive audiological assessments in individuals with renal anomalies, potentially broadening diagnostic considerations and therapeutic approaches.

Keywords: Conductive hearing loss, Deafness, Hypertrophy, Organogenesis, Solitary kidney

CASE REPORT

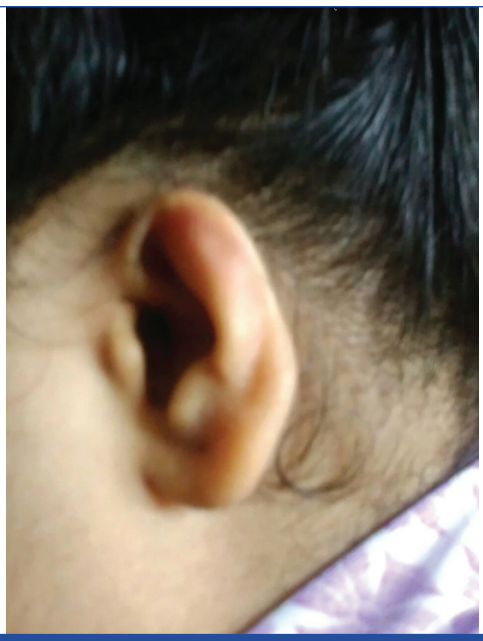
An eight-year-old female child presented to the Department of Audiology with the concern of reduced hearing in left ear for the past three years, as noticed by the parents. Medical history revealed non-visualised right kidney with compensatory hypertrophy of left kidney, representing absent/ectopic right kidney and normal uterus detected at one year of age through transabdominal ultrasound. There was no history of trauma, and the patient was otherwise in good health with no delay in speech and motor milestones. Her scholastic and extracurricular performances were observed to be at par.

On visual examination the dimensions of right ear were appropriate whereas the left ear appeared abnormal and was classified as microtia grade I [1] with possible stenosis of external acoustic meatus [Table/Fig-1,2].



[Table/Fig-1]: Right ear with normal pinna dimensions.

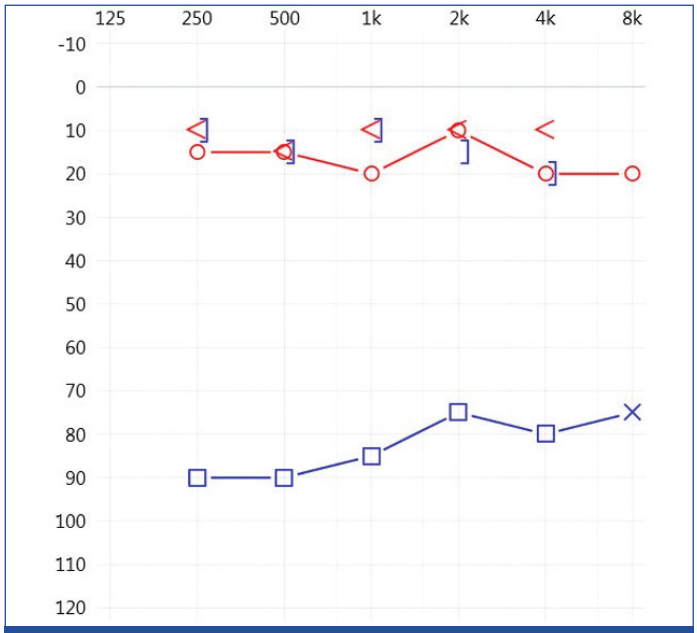
Pure Tone Audiometry (PTA) testing was performed using Modified Hughson-Westlake procedure [2] which revealed normal hearing sensitivity in the right ear and a score of 100% in speech discrimination testing, whereas a moderately severe hearing loss was observed



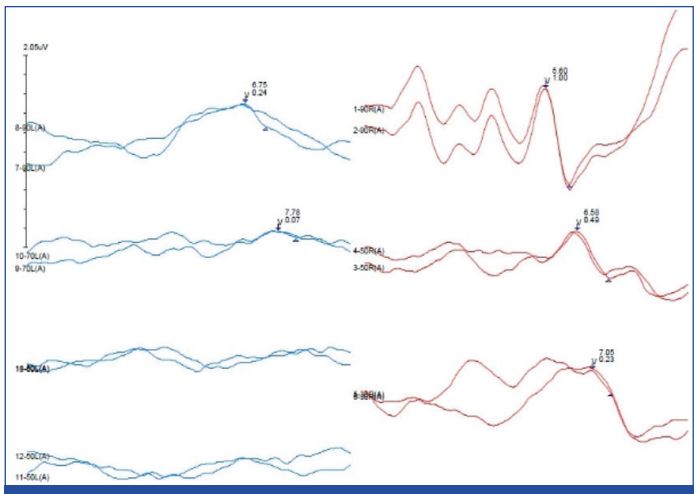
[Table/Fig-2]: Left ear with microtia grade I.

in left ear [Table/Fig-3]. Surprisingly, the masked bone conduction thresholds were well below 20 dBHL (deciBells Hearing Loss), giving rise to a large air-bone gap, which inadvertently led to labelling this type of hearing loss as conductive, despite the greater degree. Plateau method of masking was utilised to mask the right ear. She obtained extremely poor scores (12%) in speech discrimination testing using Phonetically Balanced (PB) words with contralateral masking. To verify the presence of middle ear pathology in left ear, Immitance Audiometry was performed. Normal tympanometric findings in right ear alongside abnormal findings in left ear were noted as follows: Although conventional 226 Hz tympanometry revealed A type tympanogram in both ears, multi-component tympanometry in the left ear at 678 Hz probe tone showed 5B3G pattern, which raised the suspicion of a probable mass dominant middle ear system. This inference was based on Vanhuyse model [3]. She had robust Distortion Product Otoacoustic Emissions (DPOAE) in right ear and absent emissions in the left ear. DPOAE was done using diagnostic protocol involving L1/L2: 65/55 dBSPL; F2/F1:1.2 and 2 points per octave. While threshold estimation of Auditory Brainstem

Response (ABR) via air conduction revealed normal hearing in right ear (V peak observed till 30 dBnHL), findings in left ear were suggestive of moderate to moderately severe degree of hearing loss (V peak observed till 70 dBnHL). Absolute latencies of peak V were prolonged (6.75, 7.78 at 90 and 70 dBnHL, respectively) with compromised wave morphology [Table/Fig-4]. As existence of conductive component was apparent through the large air-bone gap in PTA, 5B3G pattern at 678 Hz and prolonged absolute latencies in ABR in the left ear, Bone Conduction ABR (BC ABR) was opted to know the true degree of cochlear hearing loss, by excluding effects of external and middle ear systems. It was done for the left ear by introducing a contralateral masker. V peak was extant at 50 dBnHL (deciBells noise-induced Hearing Loss) with an absolute latency and amplitude of 6.30 and 0.54, respectively, validating the presence of conductive hearing loss rather than a cochlear hearing loss and thus endorsing findings and conclusions drawn from other audiological tests. To investigate the cause, a high field 1.5T Magnetic Resonance Imaging (MRI) of the brain and cochlea was performed which revealed no demonstrable abnormality in the brain and inner ear (bilateral vestibulocochlear nerve bundle appeared normal). An ultra-low dose multislice-Computed Tomography (CT) of temporal bone revealed small bony bar fixing in the incus to the lateral wall of left middle ear with suspicious partial fusion of left incudomalleolar joint. However, no genetic testing was done due to financial constraints, which may have helped associate the phenotype to a specific mutation, if any.



[Table/Fig-3]: Pure Tone Audiometry (PTA) results of right and left ear where the left ear threshold indicates severe conductive hearing loss.



[Table/Fig-4]: Waveform of Air Conduction Auditory Brainstem Response (AC ABR) where the V peak is obtained till 70 dBnHL in left ear.

Based on collective interpretation from the aforementioned battery of tests, a diagnosis of normal hearing sensitivity for the right ear and moderately severe conductive hearing loss for left ear was given. The treatment option suggested was a bone conduction amplification device due to preserved bone conduction threshold and large air-bone gap, after which the patient did not follow-up.

DISCUSSION

Since the discovery of Potter's sequence, renal agenesis has received great attention. The relationship of renal agenesis with craniofacial anomalies has been extensively studied over the years [4]. The consequences, although debilitating, is highly variable. One detrimental feature noted in these individuals is hearing loss. A substantial 47% of children with renal agenesis exhibited hearing loss out of 151 children studied in an earlier investigation [5]. This considerably high prevalence has led to numerous attempts in understanding the pathophysiologic link between renal agenesis and hearing loss. In the late years, embryogenic and molecular explanations have unraveled the grounds of this confounding association [6,7]. The presence of external ear abnormalities coexisting with Unilateral Renal Agenesis (URA) such as in the present patient has been previously reported in the literature [8]. However, the presence of conductive hearing loss contralateral to the side of renal agenesis in the patient as opposed to ipsilateral mixed [9] or sensorineural hearing loss [10] reported in the literature brings to the limelight, new characteristics of hearing loss associated with URA. The conductive loss in the left ear of the patient is due to the middle ear ossicular malformations confirmed through radiologic findings. The immittance, DPOAE and ABR findings were consistent with findings expected in conductive hearing loss. In conductive hearing loss, speech discrimination is generally unimpaired. In contrast to this putative notion, the present patient had poor scores. Few notable research supports this finding by affirming that even in conductive hearing loss, poor speech discrimination can be obtained, possibly due to prolonged periods of raised thresholds [11]. It is also noteworthy to mention that no other craniofacial or musculoskeletal anomalies except microtia and stenosis of external ear canal were found in the present case. Other typical features noted in females with URA is the presence of double uterus and even genital abnormalities which was again not observed in the present case, as confirmed by imaging [12]. Other findings in patients with URA include gastrointestinal, cardiac, musculoskeletal issues, intellectual deficits, vertebral malformations and even being associated with syndromes [12]. However, the patient's radiological and clinical findings did not show any of these previously described associations. Even the hearing loss which is said to occur alongside URA was reported to be mixed or sensorineural type with contralateral presentation [10], in contrast to the patient discussed who presented with conductive and ipsilateral hearing loss. Unilateral hearing loss usually remain undetected, especially in paediatric patients until a certain age. This is due to the covert nature of hearing loss itself and intact audition through the other ear. In some cases, it may not be first noticed by the child but rather by parents or teachers. This late detection can be avoided if all children with URA are periodically screened for hearing loss given the substantial literature evidence linking both. This may enable receiving early intervention, which may significantly contribute to appropriate development of higher auditory skills that require binaural hearing as an important pre-requisite. A bone conduction amplification device alongside subsequent auditory training is most suitable for the present patient. An in-depth investigation comprising of molecular or genetic analysis in the patient may have justified this presentation. Unfortunately, no genetic evaluation was done.

CONCLUSION(S)

This unreported group of manifestations is proposed to introduce and educate the fraternity of a new likelihood of signs. However, to

better understand the dynamics of damage to the auditory system in URA, vestibular evaluations and other electrophysiological assessments such as Vestibular Evoked Myogenic Potential (VEMP) and Video Head Impulse Test (VHIT) are recommended. Early intervention, hearing conservation practices and long-term follow-up is of prime importance in this population.

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