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### Original Article

# Audiological test outcomes in Thrombocytopenia Absent Radius (TAR) Syndrome- A case study.

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#### ABSTRACT

**Abstract:** - Aim:-The aim of present study in case with Thrombocytopenia absent radius (TAR) syndrome was to evaluate the hearing sensitivity. Methods: TAR syndrome was diagnosed based on the pedigree, laboratory findings (hemogram, platelet count, peripheral smear), bone marrow biopsy, radiological examination. Audiological test includes behavioural observation audiometry (BOA), immittance audiometry otoacoustic emission, and auditory brainstem response audiometry. Results: Four consecutive audiological evaluations were carried out over time. Results of behavioural observation audiometry revealed normal hearing sensitivity. However auditory brain stem response, otoacoustic emission and immittance audiometry results showed normal hearing sensitivity in the right ear and profound hearing loss in the left ear. Conclusion: To conclude on these case reports pediatrician, otologist and audiologist need to be aware of this relevant information and take immediate steps to provide services to patients. Regular follow-up of the clients is an important aspect as it is highlighted in present study.

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### 1. Introduction

Permanent childhood hearing loss occurs in 1 to 2 births per 1,000. It is believed that 50–60% of these cases are the result of mutations in the genetic code contained in the chromosomes, and it appears that several hundred genes are linked to hereditary hearing loss.<sup>[1]</sup> Of these genetic hearing loss causes, approximately one-third are syndromic and two-thirds are non-syndromic. Among the syndromic disorder one of the syndrome is Thrombocytopenia absent radius (TAR) syndrome. Thrombocytopenia absent radius (TAR) syndrome is a hereditary autosomal recessive inheritance genetic disorder.<sup>[2,3,4]</sup> The finding based on the genetic evaluation suggested that the <sup>[5,6,7,8,9,10,11,12]</sup> TAR syndrome is tied with microdeletion in 1q21.1, but it is not sufficient to cause TAR syndrome.

Thrombocytopenia-absent radius (TAR) syndrome is characterised by hypomegakaryocytic thrombocytopenia and bilateral radial aplasia. Additional skeletal features associated with TAR syndrome include shortening and, less commonly, aplasia of the ulna and/or humerus. Sensorineural hearing loss was also seen in 3% of total TAR syndrome cases.<sup>[5]</sup>

### Case report:-

**Case History:** A patient was a 9.1 months old male child, hospitalized due binocular white reflex and upper limb anomalies. Laboratory findings: red blood cell count was  $4.78 \times 10^6/\mu\text{L}$ , haemoglobin value was 7.2 g/dL, white blood cell count indicated  $10.1 \times 10^3/\mu\text{L}$ , platelets count value was  $512 \times 10^3/\mu\text{L}$ . Megacaryocytes appeared in the bone marrow aspiration in the decreased number, or did not appear at all. At the radiological examination of the upper limbs, radius was absent in both shoulders. Report of TORCH test by fully automated cobas e411 system using electrochemiluminescence (ECLIA) showed that abnormally high toxoplasmosis gondii antibodies IgG<sub>I</sub>e 3.15 IU/ml.

**Instrumentation:-** (i) BOA screener:- A duly AC-40 interacoustic, diagnostic type II audiometer with loudspeaker was used to assess the behavioural responses. (ii) Otoacoustic Emissions:- A duly calibrated DPOAE instrument of MIACO Corporation was used for screening purpose. (iii) Impedance audiometry :- A calibrated GSI-Tymp Star Middle Ear Analyzer (Version 2) to evaluate the middle ear pathology. (iv) ABR (click):- A duly calibrated ABR instrument, INTELLIGENT HEARING SYSTEM, Smart EP Version 3.8 with insert earphones ER-3 Etymotic Research with pediatric tip was used.

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**Procedure:-***Behavioural observation audiometry (BOA):-*

Child was taken for behavioural observation for auditory stimuli where he was presented with different warble tone (500, 1000, 2000, 4000Hz) with loudspeaker placed at 45°. Along with warble tone live speech and noise like BBN and narrow band noise (500Hz to 4000Hz) at different intensity level from 40 to 110 dBHL. Observable response like head turning, stilling, sound searching, localization or any behavioural changes were noted. Gradually intensity was lowered in 10dB step till 30dBHL.

**Immittance evaluation:**

Tympanometry and acoustics reflex thresholds were obtained using a calibrated GSI Tymptstar diagnostic immittance Middle Ear Analyzer. Tympanometry was performed bilaterally using 1000Hz probe tones and measurements of ipsilateral and contralateral acoustic reflex thresholds up to 110 dB were attempted at 500; 1,000; 2,000; and 4,000 Hz.

**Otoacoustic Emission:**

Distortion Product evoked Otoacoustic Emissions elicited using an 80 dB SPL peak equivalent (peak) level click, present in 5 successive trials, where P1-65 dB and P2-55dB was set and a signal-to-noise ratio of at least 6 dB with a reproducibility score of at least 70% in using ECHO Scan screening MAICO OAE instrument was measured, in frequency band of 2KHz to 5KHz.

*Auditory Brainstem Evoked Response Audiometry (ABER) recording:*

For ABRE measurement, a single channel recording was used with vertex as non-inverting electrode. The earlobe or mastoid ipsilateral to the stimulus was used as inverting electrode site while the earlobe or mastoid contralateral to the stimulus was used as ground electrode. All impedances were less than 5 kohms. A high level ABR was obtained by presenting 90 dB nHL, 100 microsec rarefaction clicks through an insert earphone using a rate of 5.1 clicks per second. Two recordings were obtained to ensure repeatability. The recording parameters consisted of a 25msec time window. The bioelectrical activity was amplified 100,000 times and filtered 300–3000 Hz. Approximately 1500 averages were obtained for each recording.

**Results:**

Case came to the audiology department for hearing screening. Routine audiological evaluation was carried out. First audiological test administered was Screening OAE revealed right ear pass and left ear refer which was suggestive of right ear normal outer hair cell integrity in the right ear and abnormal outer hair cell functioning in the left ear since the hearing loss is more than 40dB (figure 1).

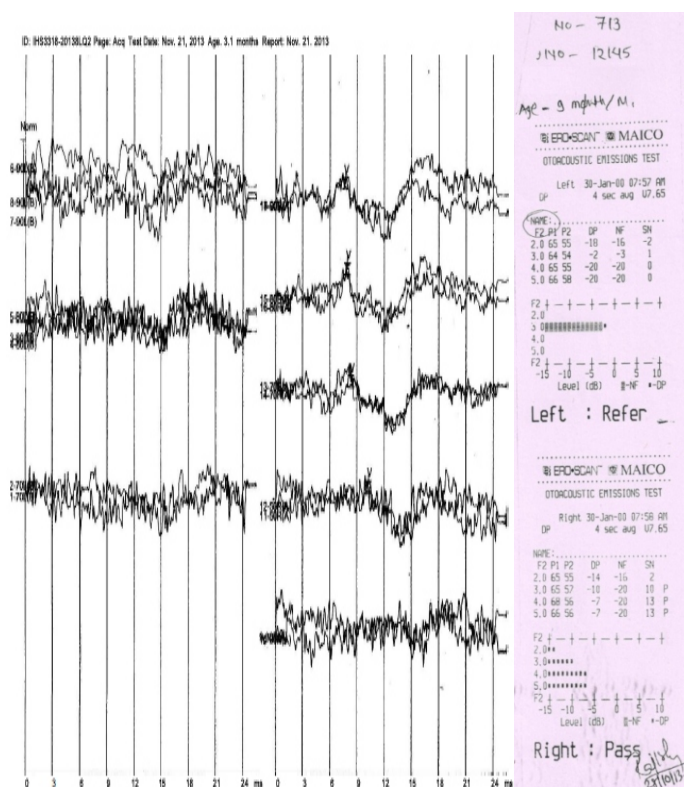
Results of impedance audiometry revealed that bilateral “A” type tympanogram along with presence of acoustic reflexes in all frequencies (right ipsilateral and left contralateral) and absence of acoustic reflexes in all frequencies (left ipsilateral and right contralateral) with normal ear canal volume and middle ear pressure which was objectively suggestive of middle ear pathology in both ear (indicated in table-1)

**Table 1: Details of test administered, instrumentation and findings.**

Test Administered	Instrument used	Right Ear	Left Ear	Impression
OAE	ECO-Scan (Screening)	pass	Refer	Left ear indication of hearing loss (>40 dB). Right ear indication of normal OHC functioning.
Immittance audiometer: Tympanogram and reflexometry	GSI- Tymptstar	A type, RE- ipsi present contra absent	A type, LE- ipsi absent, contra present	Unilateral hearing loss, no indication of middle ear pathology.
Behavioural Observation audiometry	AC-40 interacoustic, diagnostic type II audiometer	Head turning response till 40 dB. (no ear specific information)	-----	? normal hearing sensitivity.
BERA	Intelligent hearing system	Clear observable peaks till 40 dB nHL	No peaks observed till 90 dB nHL	Left ear severe degree of hearing loss. Right ear normal hearing sensitivity.

Next evaluation administered was BOA evaluation revealed observable response like head turning, stilling, sound searching, localization were noted till 30dBHL which is suggestive of normal hearing sensitivity. Auditory Brainstem Evoked Response audiometry which revealed left ear severe degree of hearing loss, as no peaks observed [figure. 1] even after repeated trials at high intensity 90dBnHL and clear observable peak was found for till 50nHL but not at 30 dB nHL suggestive of normal hearing sensitivity in the right ear.

Figure 1. ABER wave and otoacoustic emission for both ears (Right hand side: RE, Left hand side: LE)



### Discussion:-

The present report outlines profound sensorineural hearing loss in the left ear and normal hearing sensitivity in the right. Though sensorineural hearing loss has been reported in a very few literature but unilateral hearing loss have not been reported till yet. All test battery gave valuable information regarding symmetry and degree of hearing loss. Impression in Behavioural observation audiometry showed normal hearing sensitivity which was further highlighted in auditory brainstem evoked response audiometry were the results revealed that unilateral hearing loss. Though in right ear wave form were present only till 50dBnHL but not at 30dBnHL, that could be due to developmental or maturation factor. The cause of sensorineural involvement is unknown.

The TAR syndrome have varying degrees of severity. Finally, it is important to remember the genetic nature of disease and possibility of prenatal diagnosis in subsequent pregnancies. Although study is done only in one child of the family there might be condition to occur among siblings, this insists the importance of prenatal genetic counselling for all pregnant mothers. The condition rarely runs in families, unless it is a symptom of a genetic disease.

The data concerning the frequency of hearing loss in the TAR syndrome are not reported till yet; it could be because of other associated factors, lack of cooperation, and early death, is the most important cause of neurological morbidity and mortality in preterm and full-term neonates. Documentation is required for regular follow up and awareness towards occurrence of range and variety of outcomes mainly while audiological evaluation due to delayed diagnosis because of infrequent and rare symptoms of spasm in early stage. Although most cases of TAR syndromes are detected early by other medical specialist, an increased awareness of the condition involvement of audiologist in the diagnosis as early as possible and providing immediate management in terms of hearing and speech language development.

This report reinforces the importance of objective and subjective audiological test in the clinical and prognostic evaluation.

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