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Hearing, Speech, and Language Characteristics in a Case with Hemoglobinopathy Secondary to Beta Thalassemia Intermedia

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Abstract

Background: *Thalassemia is an inherited disorder characterized by a reduced amount or absence of hemoglobin, the oxygen-carrying protein inside the red blood cell. Thalassemia has its types called alpha and beta-thalassemia. Beta thalassemia is a condition in which there is a reduction or deficit in the synthesis of the beta-globin chain of hemoglobin molecules caused by a mutation in chromosome eleven. Beta thalassemia can be broadly categorized into three main categories: beta thalassemia minor, intermedia, and major. Hearing loss is a predominant trait in cases with beta-thalassemia intermedia and major. Understanding the pathophysiology of thalassemia is important for the management of these individuals. Hence there is a strong need for audiological and speech-language evaluation in them. Thus, this case report highlights the audiological, speech, and language findings in beta thalassemia intermedia.* ***Case Description:*** *Routine audiological and speech and language evaluation was carried out. Pure Tone Audiometry showed moderate mixed hearing loss in the right and severe sensorineural hearing loss in the left ear. Speech and language evaluation revealed delayed receptive and expressive abilities due to Hearing impairment.* ***Conclusion:*** *The present case report highlights the effect of Non-Transfusion Dependent treatment received by beta thalassemia intermedia patients on speech, language, and hearing abilities. The present case report also suggests that ongoing treatment of thalassemia may be a cause of hearing loss progression. Hence, periodic monitoring of hearing, speech and language skills is essential in individuals with thalassemia.*

Keywords: Beta-thalassemia, hemoglobinopathy, hearing loss, thalassemia intermedia, non-transfusion dependent treatment

INTRODUCTION

Thalassemia is an inherited genetic blood disorder that affects the human body's ability to produce less oxygen-carrying protein (haemoglobin) in the blood and healthy red blood cells. It is usually of either alpha or beta type of Thalassemia. Symptoms depend on the type of thalassemia and can vary from none too severe.

Beta thalassemia is caused by mutation in chromosome number 11 and reduced synthesis of beta-globin chains. This type can be broadly divided into three main subtypes, i.e., beta thalassemia minor, intermediate, and major. Beta thalassemia minor involves having a deficit in one beta-globin gene, leading to mild symptoms or no symptoms at all [1]. Beta thalassemia involves missing or deficit in two beta-globin genes and might cause mild to moderate anemia. Individuals with beta-thalassemia intermedia might not need regular blood transfusion [1]. Beta thalassemia major is termed as Cooley's anemia, and it is a more

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severe form caused by missing or deficit of two beta globin genes. Individuals with this condition need regular blood transfusions for their living [2]. The major characteristics of beta thalassemia are hemolytic anemia, paleness, hearing loss, jaundice, gallstones and enlarged spleen.

In recent times, a lot of focus has emerged in establishing a relationship between thalassemia and hearing loss. Approximately 17.6 % (minor), 45.45% (intermedia), and 57.4% (major) prevalence of hearing loss in India were reported among patients with beta-Thalassemia based on the subtypes [1–3]. Though the prevalence rates are high reported in Indian population too, there were not many studies reporting details of the hearing loss. Some studies have reported that individuals with beta thalassemia encounter conductive hearing loss and or sensorineural hearing loss [4, 5]. The intake of folic acid supplements which treat low blood level and folate deficiency, hydroxyurea, Chelating therapy, routine blood transfusions and bone marrow transplants are some lifesaving successful treatment options available for these individuals, they may also have some sort of effect on hearing mechanisms. The regular blood transfusion may lead to iron deposition and iron chelators (Deferasirox, Deferiprone) would be recommended, which can be ototoxic and lead to hearing loss [6]. Understanding of the pathophysiology of thalassemia is important and helps in planning intervention. But as medical intervention is a must for disease progression and life saving measures, the hearing loss needs to be regularly monitored. Various studies reported about hearing impairment in beta thalassemia patients who received Transfusion Dependent Treatment (TDT), but sparsely any studies on beta thalassemia intermedia receiving Non-Transfusion Dependent treatment (NTDT). Though there are few attempts to report the effect of beta thalassemia on hearing abilities, the combined clinical findings on the same are limited. Hence the present case report highlights the effect of NTDT received by beta thalassemia intermedia patient on speech, language and hearing abilities.

CASE DESCRIPTION

A 7-year-old male child was brought to the department with the complaint of reduced hearing sensitivity in both ears for over 3 years. Detailed history reveals that the nature of hearing loss was progressive and acquired. Medical history revealed that a known case of thalassemia intermedia subtype and the child was under medications like Hydroxyurea, and folic acid supplements for the last 3 years. Family history was significant with second degree consanguineous marriage among parents and both parents detected with thalassemia-minor and two children including the present child was diagnosed as beta thalassemia Intermedia. The Thalassemia was the reported cause for the death of the first child. Apparently, parents were provided with genetic counseling and knew about the consequences of thalassemia condition. Detailed audio, speech, and language evaluation were conducted, and the findings were tabulated below, as shown in Tables 1, 2, and 3.

Table 1. Hematology laboratory findings.

Investigations	Biological Reference Value	Observed Value
Fetal Hemoglobin	0.0–20%	93%
Hemoglobin in A0 (HbA0)	94.3–98.5%	5.2%
Hemoglobin A2 (HbA2)	1.5–3.7%	1.8%
**Impression:* Suggestive of Beta Thalassemia Intermedia.		

Note:
#Laboratory test was done and verified by MD Pathologist.
 # Laboratory Photoelectric colorimeter used as Instrument to measure Hemoglobin level.
 # Variables controlled between Fetal Hemoglobin & Hemoglobin A0 to identify the presence of Beta Thalassemia Intermedia.

Hematological findings revealed that the type of thalassemia was Beta Thalassemia Intermedia.

The audiological findings revealed bilateral tympanic membrane visible, and cone of light seen. Immittance evaluation revealed “As” type of tympanogram in right ear which would indicate (?) middle ear pathology, while in left ear “A” type tympanogram depicting normal middle ear functioning, DPOAEs in both ears were absent indicating outer hair cells dysfunction. Pure tone audiometry

possesses moderate mixed hearing loss in the right ear and severe sensorineural hearing loss in the left ear with speech recognition scores of 50% in right and 30% in left ear, respectively. To correlate the behavioral responses Auditory brainstem response was carried out which gave an impression of moderate hearing loss in right ear followed by severe hearing loss in left ear.

Table 2. Audiological findings.

Investigations	Findings	Impression
Otoscopic examination	Tympanic membrane is visible & presence of a cone of light in both ears.	Indication of Normal external auditory canal and tympanic membrane
Pure Tone Audiometry (PTA)	*Right ear:* 53 dBHL. *Left ear:* 77 dBHL.	*Right ear:* Moderate Mixed hearing loss. *Left ear:* Severe sensorineural hearing loss
Speech Recognition Score (SRS)	*Right ear:* 50%. *Left ear:* 30%.	–
Immittance Audiometry	*Right ear:* "As" type tympanogram with absent acoustic reflexes. *Left ear:* "A" type tympanogram with absent acoustic reflexes	*Right ear:* (?) indication of middle ear pathology. *Left ear:* indication of normal middle ear functioning.
Oto acoustic emission (OAE)	Absence of DPOAE in both ears	Bilateral indication of outer hair cells dysfunction
Auditory Brainstem Response (ABR)	*Right ear:* clear & replicable V peak obtained up to 70 dBnHL (Clicks) and absolute latencies were delayed which might be due to middle ear pathology (Figure 1). *Left ear:* No clear & replicable V peak obtained at 90dBnHL (Clicks) and 80 dBnHL (Tone burst) (Figure 1).	*Right ear:* Moderate Hearing loss *Left ear:* severe hearing loss
**Provisional diagnosis:* Moderate mixed hearing loss in right ear and Severe sensorineural hearing loss in left ear.		

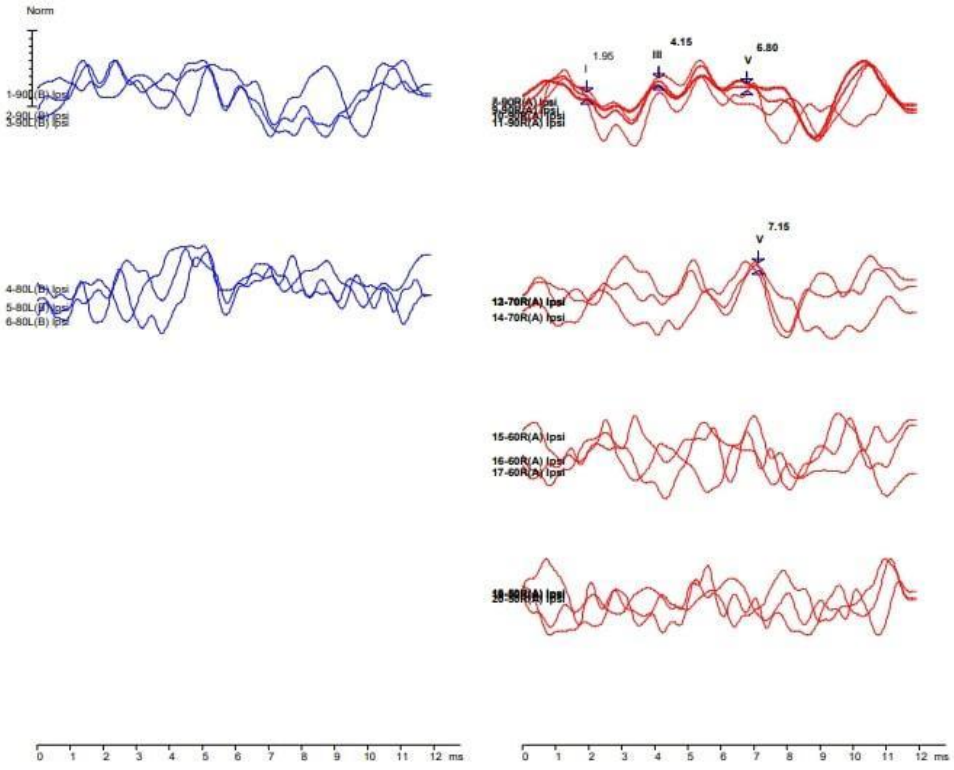

Figure 1. Waveform of Brainstem Auditory Evoked Potentials (BAEP).

Table 3. Speech and language evaluation.

Investigations	Findings
Oral Peripheral Mechanism Examination (OPME)	All articulators were structurally and functionally normal for speech production.
Receptive Expressive Emergent Language Scale (REELS)	*Receptive Language Age (RLA):* Scattered between 5–7 years. *Expressive Language Age (ELA):* Scattered between 27–36 months.
Communication Development Eclectic Approach to Language Learning (Cognitive domain) (COMDEALL)	Cognitive domain is achieved up to 66–72 months.
**Provisional Diagnosis:* Spoken Language Disorder secondary to hearing impairment.	

Speech and language evaluation revealed that delay in receptive and expressive language abilities due to hearing loss associated with thalassemia. Parents were asked to take regular follow-up evaluations as the hearing loss is progressive in nature and were asked to attend speech and language intervention for the child.

DISCUSSION

Thalassemia causes a deficiency in the production of hemoglobin, resulting in anemia with reduced oxygen supply to cochlea, which might lead to hearing loss. Bone deformities, and less blood supply to cochlea might be the cause for mixed hearing loss [4]. Thalassemia might lead to severe bone deformities, which may include stiffness in the middle ear transmission system, resulting in conductive hearing loss [5]. Intake of hydroxy urea may cause blood vessel damage, which may interrupt the blood supply to the ear too [6, 7]. Excessive folic acid intake may lead to mutation in the Methylenetetrahydrofolate reductase (MTHFR) gene [Chromosome 1] mutation may have the risk of hearing impairment [8, 9]. Above all, the medications used to cure thalassemia might exacerbate hearing loss. The present case entails Non-Transmission Dependent Transfusion, treatment side effects and genetical predisposition which led to hearing loss.

The outcome of present study is quite different to that of study by S. De Virgiliis et al. (1979). The results of their study showed presence of conductive and mixed hearing loss because of chelating therapy in 75 children with beta thalassemia major [10]. Another study by Jimmy Passat, Bulan Ginting Munthe (2001) where Brainstem Auditory evoked potentials (BAEP) in 65 thalassemic patients between 3 to 18 years, revealed hearing loss ranging from moderate to severe degree [11].

The present case report focuses on hearing, speech, and language characteristics of beta thalassemia intermedia patient which is distinctive from other studies and is an additional case report to those reported in literature and suggests hearing loss due to the combined effect of genetic predisposition and treatment side effects for individuals with Thalassemia. The current work supports evidence-based practice in this area.

CONCLUSIONS

Since the outset of hearing loss is not predictable in cases with thalassemia audiological testing is suggested before start of treatment to detect the hearing impairment and regular monitoring need to be done to know the progressiveness of the problem. To manage the hearing issues with appropriate amplification devices can be prescribed like behind the ear and receiver in the canal and it should be made mandatory for one to attend regular speech and language therapy along with alternative augmentative communication strategies to improve overall quality of life.

Declarations

We confirm that the manuscript has been read and approved by all named authors. We further confirm that all of us have approved the order of authors listed in the manuscript.

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