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TREACHER-COLLINS SYNDROME PRESENTS IN A PRIVATE OUTPATIENT AUDIOLOGY CLINIC TO EVALUATE HEARING THRESHOLDS: A CASE REPORT

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Abstract

Background: *Treacher-Collins syndrome (TCS), additionally known as Franceschetti syndrome or mandibulofacial dysostosis, is an unusual craniofacial development defect that occurs in approximately one out of every 50,000 childbirths. Deafness, bilateral auricular anomalies and facial bone hypoplasia are its distinguishing features. Four genes control its inheritance: TCOF1, POLR1C, POLR1D, and POLR1B. Clinical symptoms are the primary determinant of diagnosis; however, genetic studies can help confirm it.*

Objective: *The aim of this case report was to evaluate the hearing thresholds in a pediatric patient with Treacher Collins syndrome who presented to a private outpatient audiology clinic.*

Methodology: *Our case report was conducted at the Audiology Centre in Lahore in late 2024. A detailed history-taking of the patient was obtained. A patient with Treacher-Collins syndrome underwent various tests, including otoscopy, immittance audiometry, transient evoked otoacoustic emissions (TEOAEs), and auditory brainstem response audiometry (ABR). Consent was obtained from parents.*

Results & Findings: *A patient, a 5-month-old female infant, underwent immittance audiometry, which showed bilateral flat (B low) tympanograms with reduced ear canal volume. Transient evoked otoacoustic emissions (TEOAEs) were referred bilaterally. ABR displayed Wave V with clear responses that were reliable and reproducible at 60 dBnHL via air conduction in both ears and at an intensity of 30 dBnHL in BC-ABR, giving us a diagnosis of bilateral moderate conductive hearing loss. CT and MRI imaging revealed that ossicles in the right middle ear were difficult to appreciate, non-aerated mastoids bilaterally, and asymmetrical canals with significant stenosis in the right ear. The cochlea and internal auditory canals were well-formed, but anomalies in the external ear were noted.*

Conclusion: *This case report presents a significant bilateral conductive hearing loss in a 5- month -old female diagnosed with Treacher-Collins syndrome (TCS). Management involves bone conduction hearing aids, supportive therapies, and regular follow-ups. This case highlights that early intervention and appropriate support can significantly enhance the quality of life for TCS patients.*

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INTRODUCTION

Treacher-Collins syndrome (TCS), additionally known as Franceschetti syndrome or mandibulofacial dysostosis is an uncommon, craniofacial development abnormality that affects around one in every 50,000 live births [1].George Andreas Berry first identified Treacher-Collins syndrome (TCS) in 1889. It is an inherited malformation see in neonatal period with craniofacial structural malformations and other systemic ramifications [2]. The syndrome got its name from Edward Treacher Collins, an English physician who originally documented its symptoms in 1900 [3]. It influences the formation of structures from the first and second brachial arches during early embryonic development. It is distinguished by deafness, hypoplasia of facial bones (mandible, maxilla, and cheekbone), and antimongoloid slant of palpebral fissures, lower lid coloboma, and bilateral auricular abnormalities. It is a disorder characterized by underdeveloped cheek and jaw bones [4]. Specific to audiology, it presents with anomalies of the outer ears, atresia of the external auditory canals, and deformities of the ossicles, which all predispose the patient to conductive hearing loss [5]. It is primarily transmitted through a de novo mutation, however, it can also be passed down through families. Its inheritance is determined by four genes: TCOF1 (autosomal dominant), POLR1C (an autosomal recessive), POLR1D (an autosomal dominant or recessive), and POLR1B. TCOF1 is the cause in 95% of the cases. The clinical symptoms are the primary determinant of the diagnosis; however, genetic tests can provide additional confirmation. Patients with less serious TCS are asymptomatic and rarely suffer from problems. They are diagnosed after the birth of a child with more serious TCS and are symptomatic [6].

A small percentage of TCS patients may have cleft lips and/or palates [7]. Congenital atresia of the aural canal is diagnosed early, and rehabilitation with a bone-conducting hearing prosthesis, or BAHA, is preferred over surgical repair [8]. One of the most noticeable features of TCS is bilateral microtia, which is present in around 85% of patients. This deformity might be nonexistent or minor, and the patient may have total anotia with no auricular remnant [2]. TCS is distinguished by a severe counterclockwise motion of the plane of occlusal motion as well as microretrognathia, with bone deficits in both the mandible's body and ramus [9]. Treacher-Collins syndrome abnormalities affect several craniofacial regions, and the optimal outcome can only be achieved by developing complete serial therapy plans tailored to the malformation characteristics of individual patients [10]. Genetic testing is essential for recognizing clinically recognized disorders such as Treacher-Collins syndrome since it properly predicts the chance of recurrence based on the family genotype [11]. Oral anomalies in TCS lead to an increased occurrence of caries and the development of calculus because of the inadequate oral hygiene caused by the abnormalities [12]. Treacher-Collins syndrome is frequently inherited in an autosomal- dominant manner, with high clinical variability and no phenotype-genotype relationship [13]. Next-generation sequencing (NGS) is required for TCS diagnosis, and genetic consultation is useful in guiding treatment [14]. TCS-related craniofacial abnormalities can result in airway blockage, speech difficulty, hearing loss, and problems with feeding [15]. Therapy for individuals with TCS must be adapted to each individual's specific needs, as it is a highly intricate condition [16]. Regarding the several disorders that afflict the area, our awareness of the biology that drives Treacher-Collins syndrome has developed in recent years [17].

The aim of our case report was to evaluate the hearing thresholds in a pediatric patient with Treacher Collins syndrome who presented to a private outpatient audiology clinic. The Treacher Collins Syndrome study advances our understanding of the genetic and developmental causes of facial deformities. It promotes early detection, informs treatment options such as surgery and hearing aids, and enhances the quality of life for those affected and their family.

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Case Report:

A 5-month-old female, born to non-consanguineous parents, presented at the Audiology Centre in Lahore, with concerns regarding possible hearing loss. The pediatric patient was clinically examined and revealed characteristic features of Treacher Collins syndrome, including downward-slanting palpebral fissures, a retruded chin, a bird-like facial appearance, and sunken cheekbones. Additional findings included a broad nasal bridge, bilateral microtia, and malformed auricles with external auditory canal stenosis (Figure 1).

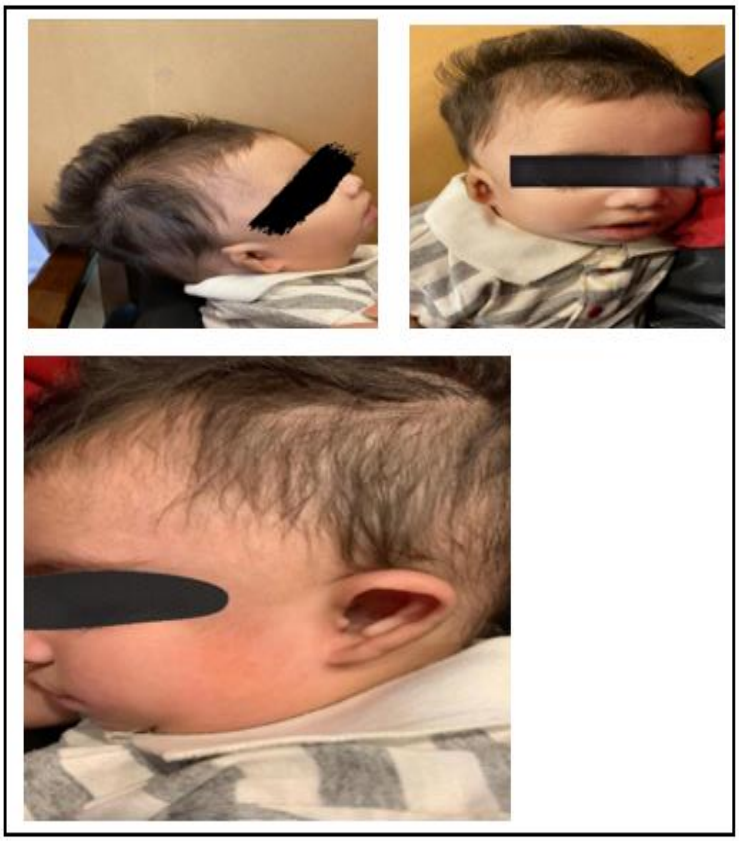

Fig 1: Pediatric Patient with Treacher-Collins syndrome

However, there were no skeletal deformities or cleft palates. A CT imaging showed bilateral middle ear cavities with fluid. Small ossicles in the right middle ear were difficult to appreciate. The ossicles were normal on the left side along with non-aerated mastoids bilaterally. External auditory canals were asymmetrical and funnel-shaped. There was significant stenosis of the cartilaginous and osseous segments of the right external ear than the left. The stenosis was prominent at the junction of the two segments of the canal in the left ear canal (the junction of cartilaginous and bony portions). The MRI was unremarkable, with well-formed cochlea(s), particularly the basal turns. The cochlear canal and the adjoining internal auditory meati/canals were also normal. The vestibulocochlear nerve root complexes were normal, with preserved root entry zones bilaterally. External ear asymmetric anomalies are noted. A genetic investigation was requested, but the results were pending when we saw the patient for evaluation.

Material and Methods:

The pediatric patient, diagnosed with Treacher-Collins syndrome, was evaluated at the Audiology

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Centre in Lahore in late 2024. Audiological and physical examinations were conducted. A detailed history-taking of the patient was obtained. Otoscopy, Immittance audiometry, transient evoked otoacoustic emissions (TEOAEs) and auditory brainstem response audiometry (ABR). Consent was obtained from parents. The Auditory Brainstem Response (ABR) is an objective assessment of auditory system function. ABR testing for children takes place while the child is sleeping or sedated. During ABR testing, a positive electrode is positioned at the high forehead (Cz). Two electrodes are inserted on the right and left earlobes, or mastoids (A1 and A2). A ground electrode is applied to the lower forehead. The most significant measurements obtained from an ABR are the absolute wave latencies, amplitudes, and interwave durations from waves I to III, III to V, and I to V. Waves I, III, and V are the most important of the seven, with wave V being clinically robust and capable of detecting the threshold. ABR recording parameters are attached in Table 1.

Table 1
The ABR recording parameters used

ABR recording parameters	
Electrode Location	**Click, NBchirp & 2kHz / 4kHz tone pip** **0.5kHz / 1kHz tone pip** Positive : High forehead (as close to vertex as possible but avoiding fontanelle) Negative : Ipsilateral mastoid Common : Contralateral mastoid
Stimulus type	Alternating polarity
Stimulus timing	Click: 100µs Tone pip: 2-1-2 cycles (linear rise–plateau–fall) or 5-cycle Blackman
Stimulus rate	45.1 - 49.1/s 35.1 - 39.1/s
Calibration values for 0dBnHL	Refer to NHSP calibration data
Amplifier reject levels	±3 to ±10µVg peak-to-peak. Start at ≤ ±5µV peak-to-peak
Amplifier filters	Low frequency: 30Hz High frequency: 1500Hz
Window length	20ms 25ms
Number of sweeps averaged per replication	If the artefact rejection level is ±5 µV: Typically: 4000 click & CE chirp
Display scale	Within range 25-100nV ≡ 1ms
Display	Wave V up

Results & Findings:

Otoscopic findings revealed bilateral microtia with stenosis of external auditory canals. Immittance audiometry showed bilateral flat (B low) tympanograms with reduced ear canal volume. Transient evoked otoacoustic emissions (TEOAEs) were referred bilaterally. ABR displayed Wave V with clear responses that were reliable and reproducible at 60 dBnHL via air conduction in both ears and at an intensity of 30 dBnHL in bone conduction (BC-ABR), giving us a diagnosis of bilateral moderate conductive hearing loss.

Treatment and Management plan:

Supportive care was recommended for this 5-month-old female infant diagnosed with Treacher Collins syndrome, which is included the use of appropriate bone conduction hearing aids, along with speech rehabilitation and regular follow-ups appointments. Management of TCS patients require specialist, multidisciplinary care from

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pediatrics, clinical inheritance, an olaryngologist, an orthodontist, an audiologist, a speech therapist, and a psychologist, with significant input from several types of surgeons.

Discussion:

We evaluated the case of a 5-month-old female infant who was diagnosed with TCS after a clinical examination when her parents approached us for an audiological assessment. Treacher Collins syndrome (TCS) is a genetically and phenotypically diverse condition with autosomal dominant and recessive inheritance. It corresponds to the human chromosome 5q32 location [15].

Bożena Anna Marszałek-Kruk (2021) reported a case of Treacher-Collins syndrome, which showed that TCS patients had varying degrees of malformed auricles, and the majority of TCS patients experienced hearing loss due to conductive causes. Conductive hearing impairment is primarily caused by middle ear abnormalities, which occur in people with hearing impairments or a lack of auditory ossicles, while our case showed the same results as conductive hearing loss [18].

Francisco Rosa (2015) found that TCS has a high prevalence of ear abnormalities such as microtia, hypoplastic middle ear cavity, atresia, and dysmorphic ossicular chain, which can result in moderate to severe hearing loss, while our case report presents moderate conductive hearing loss (19). According to Zhaoyu Pan (2021), the patient with TCS had significant craniofacial anomalies and conductive hearing loss, while our report showed the same results [14, 27].

According to Xinmiao Fan (2019), bone conduction hearing rehabilitation may be the best option for TCS patients with bilateral conductive hearing loss, while our 5-month-old infant had bilateral conductive hearing loss, and we recommend bone conduction hearing aids due to canal stenosis [20]. Patients with TCS showed (Jacqueline Kloos et al., 2023) an increased rate of otological problems, including conductive hearing loss, while our report showed similar results [21].

W. Rooijers found that in 2022, TCS patients had hearing loss, which included conductive, mixed, and sensorineural loss, while our case reported bilateral conductive hearing loss [22]. In 2024, Haojie Sun reported a case with TCS with clinical signs such as conductive hearing loss, mandible hypoplasia, and downward slanting palpebral fissures, while our case report presents the same features and conductive hearing loss [23]. According to Linda D. Vallino-Napoli, all patients had hearing loss, with 93% conductive and 7% mixed, while our case reported that bilateral conductive hearing loss [24].

TCS is vulnerable to numerous problems and requires multidisciplinary medical treatment. However, they all require psychiatric care in order to combat social rejection [25].

Early detection and repair of deafness with hearing aids or surgery is crucial for the development of these patients, especially because the majority have normal intelligence (D.L. Mittman et al.,). A child's capacity to hear during their first three years of life is crucial for their speech and language development [26, 27].

Conclusion:

This case report emphasizes the importance of early detection and management of bilateral conductive hearing loss in a 5-month-old female infant with Treacher Collins syndrome (TCS), a rare genetic disorder characterized by craniofacial anomalies and auricular malformations. The timely identification of hearing impairment in this patient facilitated appropriate audiological and medical interventions. Early use of hearing amplification, along with rehabilitative care, is essential for optimizing auditory development and overall quality of life. This case highlights the value of early screening and coordinated multidisciplinary management in improving long-term outcomes for individuals with TCS.

Conflicts of Interest:

The authors declare no conflicts of interest regarding this manuscript.

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None.

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