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Case Report

Ear, Nose and Throat Section

Congenital Absence of Stapedius Muscle and Tendon: Case Report with Review of Literature

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ABSTRACT

Absence of stapedius muscle and its tendon is an extremely rare congenital anomaly, with only nine indexed reports in live patients. It presents with non progressive, occasionally bilateral conductive deafness, although this cannot be confirmed from Pure Tone Audiometry (PTA) and High Resolution Computed Tomography (HRCT). This makes it one of the least probable differential diagnosis of conductive deafness with unremarkable tympanic membrane. The definitive diagnosis is made only at time of surgery. The condition often co-exists with other genetic/acquired disorders of the middle/inner ear (otosclerosis, tympanosclerosis, etc.,). In absence of standardised, evidence based management protocol, diagnosis and appropriate treatment become challenging, and are predominantly situational. Its occurrence follows complex embryologic sequences where the internal portion of the interhyale- the mesenchymal condensation separating the cranial mesenchyme of the second pharyngeal arch (stapedial anlage) from Reichert's cartilage- remains either absent or undergoes regression. Here, authors presents a case of 19-year-old female with congenital absence of stapedius muscle and tendon. She presented with congenital, bilateral, non progressive, moderate conductive hearing loss, and was diagnosed with this condition at surgery. There was co-existent tympanosclerosis in the middle ear that resulted in ossicular chain fixation and absence of round window reflex. The tympanosclerotic plaques were meticulously removed, ossicular chain mobility was restored, and a type II ossiculoplasty was done, along with reconstruction of the posterosuperior bony canal wall with cartilage perichondrium complex. Apart from the clinical presentation and its management, the present report also emphasises the plausible theories on relevant embryology explaining such an unusual event.

Keywords: Congenital middle ear anomalies, Hearing reconstruction, Interhyale, Reichert's cartilage, Stapedial anlage

CASE REPORT

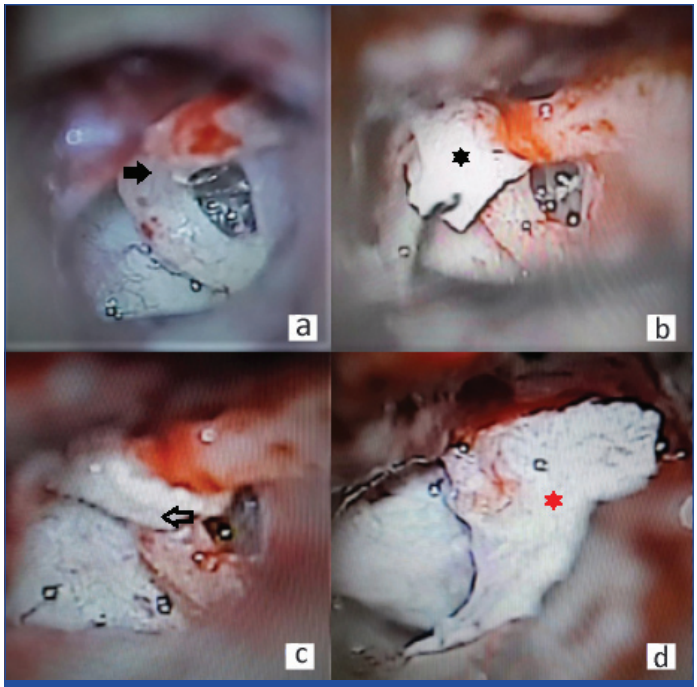
A 19-year-old healthy female presented to the Department of Otorhinolaryngology and Head-Neck Surgery with bilateral non progressive deafness since birth. There was no history of trauma or ear discharge, although she suffered from recurrent episodes of blocked ears in her childhood during cold seasons. She was otherwise healthy, without any co-morbidities and there was no family history of congenital or early onset hearing impairment. Presently, her tympanic membranes were normal on otoscopy. Pure Tone Audiometry (PTA) showed moderate conductive hearing impairment in both the ears with Pure Tone Average (PTAv) of 45 dB (decibel) in the right ear and 53.3 dB in the left; impedance audiometry revealed 'A' and 'As' curves respectively, with no acoustic reflex. HRCT of the temporal bones showed well aerated middle ears, intact ossicles and normal inner ear. The tympanomastoid cellular system on either side was poorly pneumatised but without any opacity.

The patient was planned for left sided exploratory tympanotomy under general anaesthesia. Her preanaesthetic evaluation was unremarkable. She was counselled about the realistic expectations regarding postoperative hearing outcome.

At surgery, following Rosen's endomeatal incision, the tympanomeatal flap was raised from 11 to 6 o'clock and was reflected anteriorly. Posterosuperior bony overhang was curetted until the posterior part of stapes footplate was visible. The middle ear mucosa at places had areas of tympanosclerosis; however there was no effusion or active disease process (cholesteatoma and/or granulations). At this stage, the stapedius tendon along with its muscle and the pyramid were found absent [Table/Fig-1a], and the stapes suprastructure was oriented obliquely towards the promontory. Ossicular integrity was maintained, although mobility of the malleus was restricted with fixation of the Incudo-Stapedial (IS) and Incudo-Malleal (IM) joints. Thin tympanosclerotic plaques were noted over footplate and the ossicular assembly; following atticotomy, they were found to extend within the attic. The plaques were meticulously removed, and mobility of the footplate and inter-ossicular joints was re-established with restoration of the round window reflex.

Interestingly, the IS joint was found to be replaced by loose fibrous tissue joining the long process of incus and stapes head. The fibrous tissue was excised and the IS joint was dislocated. Tragal cartilage autograft was harvested which was stripped off its perichondrium on one side. A small portion of the bare cartilage was sliced out from the tragal cartilage shield, and a type II ossiculoplasty was performed by interposing it between the long process of incus and stapes head [Table/Fig-1b]. Portion of the peeled off perichondrium was wrapped around the interposition for stabilisation [Table/Fig-1c]. Round window reflex with the cartilage-ossicle-perichondrium assembly in-situ was ensured. The middle ear was then loosely packed with blood soaked gel foam pieces. The attic and the posterosuperior bony External Auditory Canal (EAC) were reconstructed with the remaining cartilage-perichondrium complex which was thinned out before placement [Table/Fig-1d]. The tympanomeatal flap was replaced, and the EAC was packed with antibiotic soaked gel foam pieces.

The patient recuperated well after surgery. PTA done 12 weeks later showed an improvement of 15-20 dB in all frequencies with PTAv at 33.3 dB. At seven month, prior to writing up this report, she had an acceptable subjective hearing and was doing well in her social and academic life. Since she has comparable hearing impairment in the right side too, authors contemplate the presence of similar pathology there, and plan for exploratory tympanotomy after another few months at her convenience. Written informed consent was obtained from the patient.



[Table/Fig-1]: Following anterior tympanotomy and curettage of the posterosuperior bony overhang, the stapes suprastructure could be identified; however, there were no pyramid (containing the stapedius muscle) and its tendon (a). The stapes suprastructure could be seen obliquely oriented towards the promontory (a). There were diffuse areas of tympanosclerosis in the middle ear and over the ossicular chain (a), extending into the attic. Atticotomy was followed by meticulous removal of the tympanosclerotic plaques from the ossicular chain, and its mobility was established. The IS joint was found to be replaced with a loose fibrous tissue (a: solid arrow). The joint was dis-articulated and a small piece of tragal cartilage (black asterisk) was interposed between the stapes head and long process of incus (b). The assembly was wrapped with perichondrium (c; hollow arrow). The attic and the posterosuperior part of bony canal were reconstructed with thinned out tragal cartilage-perichondrium complex (d; red asterisk).

DISCUSSION

In this report, the clinical presentation of a teenage girl was illustrated with congenital absence of stapedius muscle/tendon unit, discuss the challenges at diagnosis and treatment, and analyse the probable aetiology from embryologic perspectives.

Congenital middle ear malformations include minor anomalies (fixation/disruption of the ossicular chain) and major anomalies (with additional aural atresia, poorly aerated or hypoplastic middle ear cleft, dysplastic ossicular chain, and alteration in facial nerve course) [1]. The individual anomalies and syndromic disorders within this broad family are well-established clinical entities, with overall incidence of ~1 in 10,000 births [2]. However, among these, absence of stapedius muscle and its tendon, that represents second pharyngeal arch dysmorphogenesis, is extremely rare. Review of English literature (indexed with PubMed/MEDLINE and Scopus) reveals its incidence of 0.5%, with only nine patients on record [Table/Fig-2] [3-6]. This however excludes the data from Hough JV that could not be retrieved [7]. Most of the published documents are single case reports; the largest series of five patients was reported by Hough JVD [6]. However, there have been several documentations of this entity at cadaver temporal bone dissections [8-10]. Patients usually present with congenital, non progressive conductive deafness, unlike generally progressive hearing impairment seen in acquired conductive defects [4]. Since, this condition is seldom encountered in routine otology practice it forms one of the least probable differential diagnosis of hearing impairment with intact, unremarkable tympanic membrane. Definite preoperative diagnosis is often not possible even with HRCT scan of the temporal bones [4], and attempts at hearing reconstruction pose a challenge.

Sl. No.	Studies	Presentation	Investigations and findings	Treatment
1.	Present case	Congenital, bilateral, non progressive hearing loss in a 19-year-old girl	• PTA: Moderate conductive hearing loss in both ears, with PTAv at 45 dB in right ear and 53.3 dB in left ear • 'A' and 'As' curves respectively in right and left ears with no acoustic reflex (impedance audiometry) • HRCT temporal bones: Unremarkable At surgery • Absent stapedius tendon, muscle and pyramid • Tympanosclerosis in the middle ear mucosa and over the ossicles • Fixation of incudo-malleal and incudo-stapedial joints • Incudo-stapedial joint replaced with loose fibrous tissue	Exploratory tympanotomy in the left ear. Meticulous removal of the tympanosclerotic plaques, re-inforced type II ossiculoplasty, reconstruction of the posterosuperior canal wall.
2.	Dalmia D and Behera SK, 2017 [3]	Bilateral hearing loss in a 47-year-old woman	• PTA left ear: Moderate-to-severe conductive hearing loss with mixed hearing loss at 2kHz • PTA right ear: Moderately severe-to-severe mixed hearing loss • Bilateral As curve (tympanometry) At surgery: • Stapedius tendon, muscle, pyramid absent • Fixed footplate	Exploratory tympanotomy in the right ear followed by stapedotomy and insertion of Teflon piston.
		Left sided hearing loss in a 45-year-old woman	• As curve in left ear (tympanogram) At surgery: • Stapedius tendon, muscle, pyramid absent • Fixed footplate	Exploratory tympanotomy in the left ear followed by stapedotomy, repair of floating footplate with vein graft, and insertion of Teflon piston.
3.	Kanona H et al.,2015 [4]	Left sided progressive hearing loss in a 13-year-old girl, since 2 years	• PTA: Conductive hearing loss in the left ear with PTAv at 68 dBHL. • HRCT temporal bones: Suggestive of the anomalies on retrospection At surgery: • No connection between stapes suprastructure and footplate • Fixed footplate • Absent stapedius tendon	Exploratory tympanotomy in the left ear followed by stapedotomy and insertion of piston.
4.	Magnuson T and Har EIG1994 [5]	Congenital, left sided hearing loss in a 13-year-old boy	• PTA left ear: Conductive hearing loss with PTAv at 60 dbHL • As type curve in left ear (tympanometry) At surgery: • Abnormal position of the fallopian canal (in the mesotympanum) • Anterior crus of the stapes smaller and attached to the fallopian canal • Posterior crus longer, with free-floating tip • Absent oval window/footplate • Absent stapedius muscle, rudimentary stapedius tendon, well-formed pyramidal process	Not described.
5.	Hough JVD. 1958 [6]	This is a thesis manuscript that describes five patients who were diagnosed with this disorder at surgery (the now-obsolete stapes mobilisation). Clinical details were not provided.		

[Table/Fig-2]: The present patient, and those with congenital absence of stapedius muscle and tendon documented in the PubMed/MEDLINE and Scopus databases: presentation, investigations, and treatment.
*excludes data from Hough JV [7] that could not be retrieved.
PTA: Pure tone audiometry; PTAv: Pure tone average; HRCT: High resolution computed tomogram; DHL: Decibel hearing level

Congenital middle ear malformations belong to the family of first and second pharyngeal arch dysmorphogenesis, which includes isolated syndromes within the broad category of oculo-auriculo-vertebral spectrum (hemifacial microsomia, branchio-otic/branchio-oto-renal spectrum disorders, Goldenhar syndrome, etc.,) [11], or might be associated with related syndromic disorders like Pierre-Robin, Treacher-Collins, Crouzon, Mobius, Fanconi, Vater and Klippel-Feil. In the embryonic life, craniofacial growth occurs following orderly composition of mesodermal and cranial neural crest cells through complicated signalling networks. Combination of genetic and environmental factors might lead to dysregulation of these cell-signalling pathways resulting in hypoplasia/aplasia of the structures formed from the first two pharyngeal arches [3].

The second pharyngeal (hyoid) arch enlarges by the sixth intra-uterine week to overlap the third, fourth and sixth arches [12]. The Reichert's cartilage- part of the second arch mesenchyme- forms the styloid ligament, manubrium of the malleus, long process of incus, the stapes suprastructure, and the body and lesser horn of the hyoid bone. Muscular component of this arch gives rise to the muscles of facial expressions, the stapedius, stylohyoid, and posterior belly of digastric- all innervated by the facial nerve.

Recent observations in the study of related to embryology reveal conflicting reports regarding development of the stapes footplate, suprastructure, and the stapedius muscle/tendon unit. The posterior belly of digastric near the mastoid process supposedly gives off fascicles that pass through the stylomastoid foramen and form the stapedius muscle [3]. From the traditional embryologic perspectives, stapes originates from two sources- its footplate from the otic capsule (derived from neuroectoderm), and the suprastructure from the Reichert's cartilage mesenchyme [4].

However, recent studies contradict this hypothesis [10,13]. Both the footplate and suprastructure are now said to originate from stapes anlage, the cluster of signalled embryonic cells derived from the cranial mesenchyme of the second pharyngeal arch. The stapedius muscle/tendon unit originates from the internal part of interhyale, a localised mesenchymal condensation separating cranial mesenchyme of second arch with Reichert's cartilage [13]. According to this theory, the stapes anlage, interhyale, and the Reichert's cartilage are compartmentalised components of the second pharyngeal arch mesenchyme arranged cranio-caudally, which are anatomically linked but with distinct derivatives. The present concept can suitably explain the occurrence of congenital absence of the stapedius muscle/tendon unit by the fact that the internal part of the interhyale might be either absent or have undergone regression, independent of the stapes anlage [10].

The stapedius muscle responds primarily to high intensity and low frequency sound. When the incident sound wave reaches 20 dB above the acoustic reflex threshold (normal: 70-100 dB sound pressure level), the attenuation reflex reduces the intensity of sound transmitted to the organ of Corti by ~15 dB [14]. Therefore, the stapedius muscle/tendon unit is pivotal in maintaining the physiology of hearing and vocalisation. One of the challenges in managing congenital absence of stapedius muscle/tendon unit lies in establishing the diagnosis prior to surgical intervention. As mentioned earlier, it is one of the least probable differential diagnosis of conductive hearing impairment with an intact, unremarkable tympanic membrane. Given the rarity, it is seldom suspected so as to warrant an HRCT of temporal bones as routine. In present case HRCT was performed; however, it could not establish the anomaly. Truly, Multi Detector Computed Tomography (MDCT) scan and cone beam CT with 3-dimensional reconstruction remain gold standards in defining the finer bony components of craniofacial anatomy, including malformations of the middle ear ossicles [15]. Within the temporal bone, the stapes suprastructure, footplate, pyramidal eminence and the stapedius muscle/tendon can be viewed with fine axial sections on an MDCT scan, assisted by axial and coronal reformatted images along the planes of the lateral and superior semicircular canals, respectively [16]. These tests were not performed in index patient, and as evident from our present experience, HRCT might not have enough precision to detect such intricate details.

Without specialised imaging facilities and with low index of suspicion, definitive diagnosis for such rare congenital anomaly can be reached only at surgery. Interestingly, this condition might co-exist with other middle/inner ear diseases, like tympanosclerosis (as seen in index patient), or otosclerosis, as noted by Dalmia D and Behera SK [3]. There may also be other co-existing congenital disorders that may contribute to the conductive deafness. For example, absent stapedius tendon might be associated with lack of communication between the footplate and suprastructure [4]. Management in such situations should aim at addressing the associated disease(s) besides the primary anomaly. However, review of the PubMed/MEDLINE and Scopus databases did not reveal unanimous, evidence based management protocol. This was due to poor reported case strength of the grossly heterogeneous family of congenital middle ear disorders which demands individualised decision making.

Jahrsdoerfer RA et al., described a 10-point rating criteria based on the findings of preoperative HRCT of temporal bones that could determine the candidature for suitable hearing reconstruction depending upon the various congenital middle ear defects, and help predict postoperative improvement in speech reception threshold [17]. There have been more extensive classification systems, like that of Siegert R et al., who devised a 0-28 point prognostic scoring system [18], and some specifically related to ossicular malformations (Teunissen EB and Cremers WR [19]), with recommendations for hearing reconstructive techniques based on the ossicular discontinuity status (Batti JS and Bluestone CD [20]). However, authors' present experience shows that those might not be exhaustive, because none of them specifically cites the absence of stapedius muscle/tendon unit as a possible or probable congenital anomaly. This could be because, most of them were based on HRCT findings which might not be precise enough to detect intricate details of middle ear malformations.

Evidently, decision making on hearing restoration in congenital disorders like absence of the stapedius muscle/tendon unit remains a challenge and is mostly situational, especially when associated with other middle/inner ear diseases. In index patient, exploratory tympanotomy revealed intact ossicles but without true synovial IS joint, absence of stapedius muscle/tendon unit, and fixed ossicular chain and footplate due to tympanosclerosis. Following atticotomy and meticulous removal of tympanosclerotic plaques from the ossicular chain and footplate, a technically type II tympanoplasty was performed with an inter-ossicular cartilage autograft wrapped with perichondrium, along with reconstruction of attic and posterosuperior bony EAC with cartilage perichondrium complex.

CONCLUSION(S)

Congenital absence of stapedius muscle/tendon unit is a rare second pharyngeal arch anomaly that usually presents as non progressive hearing impairment. It is often associated with other middle/inner ear pathologies, and even with multiple congenital disorders affecting the hearing. These factors contribute to the severity of deafness, and play a decisive role during surgical attempts at restoring hearing mechanism. Understanding the embryology of this anomaly is crucial in determining the pathogenesis and planning the management, and more importantly, considering it as a potential differential diagnosis of non progressive conductive deafness with intact tympanic membrane. The present clinical illustration revisits this unusual clinical entity through presentation of a new patient, and discussion on its embryologic perspectives and management aspects in terms of diagnosis and surgical reconstruction of hearing mechanism.

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