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CASE REPORT

A case of congenital bilateral stapes agenesis

Doğuştan iki taraflı stapes agenezisi: Olgu sunumu

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Congenital absence of the stapes and the oval window is an anomaly reported in only sporadic cases. We present a 17-year-old male patient with congenital bilateral conductive hearing loss. The external auditory canal and the tympanic membrane appeared normal on both sides. An exploratory tympanotomy in the right ear revealed dehiscence of the the horizontal segment of the facial nerve, which was displaced inferiorly, occupying the area of the absent oval window. The stapes was totally absent and a malformed long process of the incus was attached by a fibrous band to the promontory. Manipulation of the facial nerve in association with stapedectomy or vestibulotomy was avoided in order not to injure the nerve. Instead, amplification with hearing aids was recommended to the patient.

Key Words: Branchial region/anatomy & histology; ear canal/abnormalities; ear ossicles/abnormalities; ear, middle; facial nerve/abnormalities/embryology; hearing loss, conductive/diagnosis/etiology; stapes/abnormalities/embryology.

Doğuştan oval pencere ve stapes yokluğu çok nadir bildirilmiş bir anomalidir. Bu makalede doğumdan itibaren iki taraflı iletim tipi işitme kaybı olan 17 yaşındaki bir hasta sunuldu. Hastanın dış kulak yolu ve timpanik membranın görüntüsü iki tarafta da normaldi. Sağ eksploratif timpanotomi yapıldığında fasyal sinirin horizontal segmentinin açık olduğu ve aşağıya deplase olarak oval pencere alanını tamamen kapladığı gözlendi. Stapesin olmadığı ve malforme olan inkus uzun kolunun fibröz bir bant ile promontoryuma bağlandığı görüldü. Fasyal sinire zarar vermemek düşüncesiyle, sinirin düzeltilmesi ve stapedektomi veya vestibülotomi yapılmasından kaçınıldı ve hastaya işitme cihazı ile amplifikasyon önerildi.

Anahtar Sözcükler: Brankiyal bölge/anatomi ve histoloji; kulak kanalı/anormallik; kulak osikülü/anormallik; kulak, orta; fasyal sinir/anormallik/embriyoloji; işitme kaybı, iletim tipi/tanı/etyoloji; stapes/anormallik/embriyoloji.

Congenital malformations of the middle ear have been described in association with various head and neck anomalies and syndromes. Isolated middle ear anomalies may become appreciable only with conductive hearing loss and can be identified only during surgical exploration. In order to treat these conditions safely and effectively, the otologic surgeon must be able to recognize any ossicular malformation and understand the embryologic background leading to these conditions. One type of ossicular malformation, stapes agenesis with absence of the oval window, has been rarely reported.[1-5] A patient with this anomaly is presented together with a review on the embryology and management of this condition.

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CASE REPORT

A seventeen-year-old male patient presented with bilateral hearing loss since his birth. He reported no fluctuation of hearing and no history of otitis media, trauma, vestibular dysfunction, tinnitus, or otorrhea. Nor did he have a family history of an otologic disease. His craniofacial development was normal. On physical examination, both tympanic membranes were intact and mobile. The external auditory canals were completely normal. Pure-tone audiometry demonstrated bilateral 50 dB conductive hearing loss with 88% discrimination. Tympanometry showed normal middle ear pressure with a steep curve consistent with a possible ossicular discontinuity (type A_d). Ipsilateral and contralateral acoustic reflexes were absent (Fig. 1). Under local anesthesia, a right exploratory tympanotomy was performed. The stapes was completely absent and dehiscence of the tympanic segment of the facial nerve was noted, being displaced inferiorly wherein it occupied the entire oval window niche obscuring the stapes footplate. The malleus handle appeared normal. The long process of the incus was short and thick, at the tip of which there was a cleft 1 mm in size attached by a thin fibrous band to the promontory. The round window was readily discernible (Fig. 2). Surgery was completed without any attempt of reconstruction because of the absence of surgical landmarks on the medial wall of the middle ear. To avoid further risks to the facial nerve, amplification with hearing aids was recommended to the patient. Postoperative temporal bone computed tomography (CT) showed malformed ossicles with the absence of the stapes, normal pneumatization of mastoid air cells and normal inner ear morphology (Fig. 3a, b).

DISCUSSION

Congenital absence of the oval window and the stapes in the presence of a normally appearing external auditory canal and tympanic membrane is rare. A review of the embryology of the middle ear may help explain the anomalies found in this case. The stapes develops from the second branchial arch (Reichert's cartilage). By five to six weeks of gestation, the embryologic mass of the stapes becomes recognizable and begins its growth toward the otic capsule. At seven weeks, the stapes assumes an annular shape and attaches to the lateral mass of the otic capsule, producing an invagination. This invagination will be the site of the oval window and the developing stapes footplate will be of dual origin, namely, the tympanic part from the Reichert's cartilage, and the vestibular part from the otic capsule.[1,4] According to Hough,[1] variation in the amount of remaining tissue and the place of separation as well as its point of attachment might explain many variations encountered.

The most common anomaly in the middle ear is aplasia of the long process of the incus and the head of the stapes, which is occasionally associated with aplasia of the manubrium of the malleus.[6,7] The cause of an abnormal stapes development with the absence of the oval window is uncertain. It has been

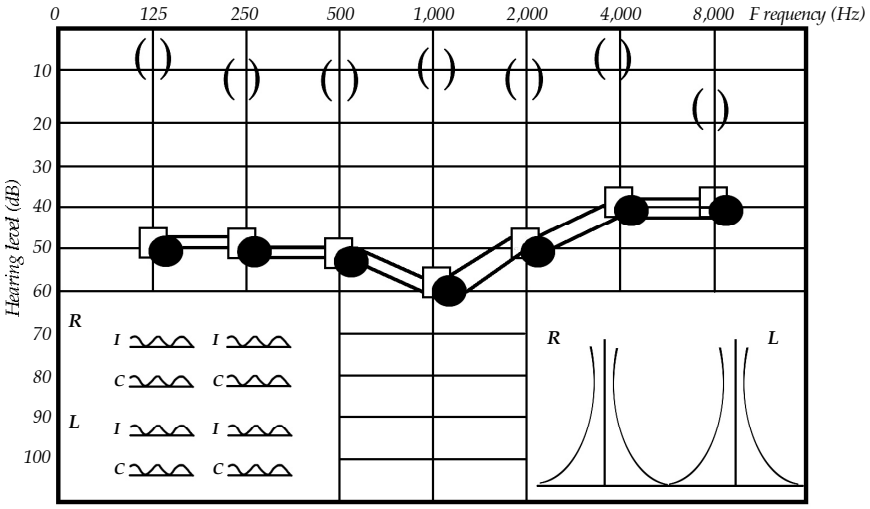

Fig. 1 - *Pure-tone audiometry, tympanometry, and acoustic reflex tests of the patient.*

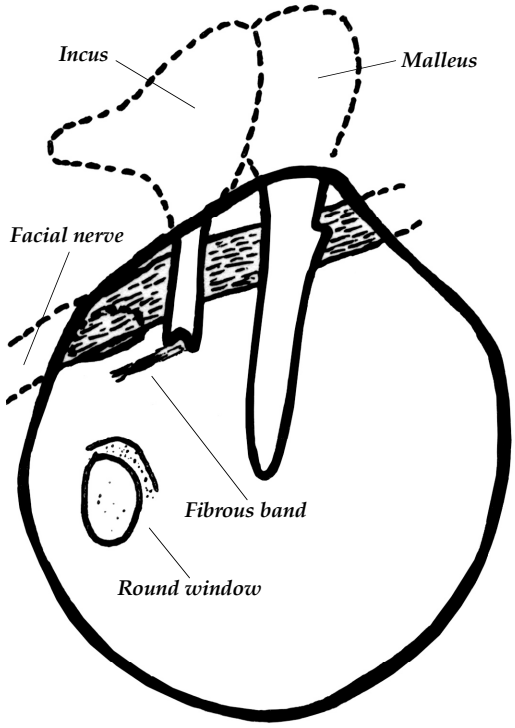

Fig. 2 - *Schematic drawing of the middle ear as seen in the operation.*

suggested that this condition may be linked to the abnormal development of the facial nerve.[8,9] If anterior displacement of the facial nerve occurs during five to six weeks of gestation, the nerve will become interposed between the otic capsule and the stapes blastema. Displacement of the facial nerve over the oval window region could prevent the stapes blastema from contacting the otic capsule, resulting in the absence of the footplate. Other structures will continue to develop from the second branchial arch.[3,9,10]

It is hypothesized that displacement of the facial nerve occurs because of the delay in the development of the first branchial arch.[8] This results in a compensatory overshifting of the second branchial arch, with its nerve assuming a more anterior position. We agree with Jahrsdoerfer,[9] who concluded that the development of the facial nerve may strongly influence the stapes development.

Controversy exists as to the proper management of a patient in whom the absence of the oval window is detected at surgery. Reconstruction may be highly complicated by two concomitant abnormalities, one of which is the displacement of the facial nerve. Failure to recognize a dehiscent and displaced facial nerve at the time of exploratory tympanotomy may be hazardous. It is suggested that the facial nerve usually overlies the oval window area in cases with absent or atretic stapes.[8] It is possible to roll the facial nerve inferiorly or superiorly;[11] however, this may increase the risk for facial paresis and manipulation of the facial nerve may further interfere with the movement of the prosthesis. The other complicating factor pertains to the abnormal development of the long process of the incus, which may render the placement of a wire or wire piston prosthesis extremely difficult.

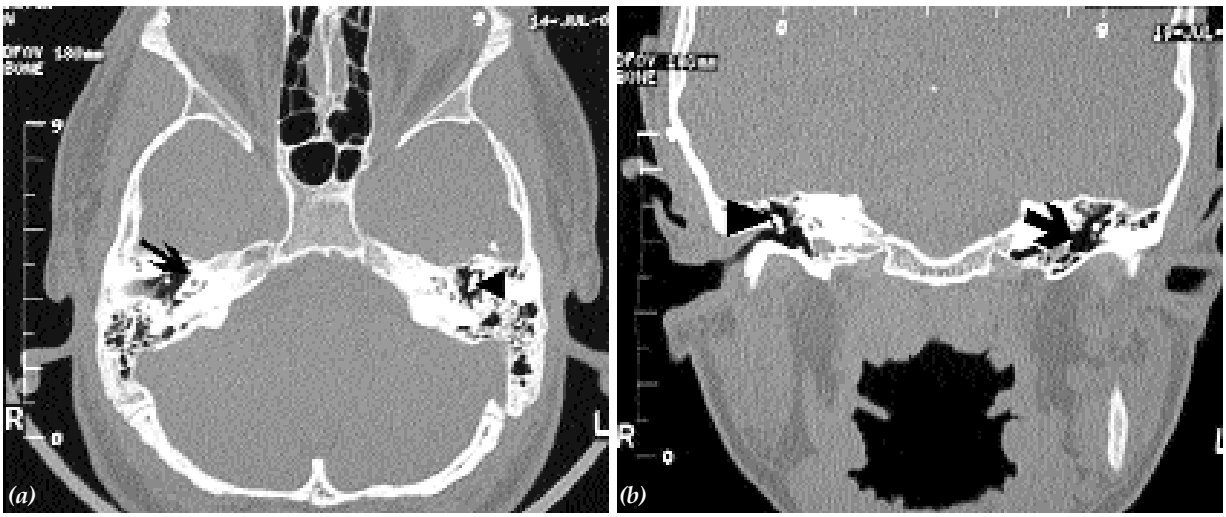

Fig. 3 - ***(a)*** *Axial and* ***(b)*** *coronal computed tomography scans of the temporal bone showing the absence of the stapes (arrow) and malformation of the ossicles (arrow head).*

Several ways have been recommended for the restoration of hearing, including fenestration of the semicircular canal,[12,13] vestibulotomy,[11] or the use of a hearing aid.[14] Lambert[10] performed vestibulotomy and reconstruction with House wires or ossicular prosthesis in six patients with absence of the oval window. Hearing initially improved in a range of 25 to 45 dB in four patients; however, much of this improvement was lost over time and the cause of the eventual loss of the early hearing gains was unclear. It is likely that a secure placement of the prosthesis in vestibulotomy is largely compromised in such patients.[5] Reiber and Schwaber[15] preferred hearing amplification in a patient with congenital absence of the stapes and facial nerve dehiscence.

In conclusion, craniofacial anomalies associated with malformations of the second branchial arch may be related to middle ear pathology. It is of note that, in a total of 15 cases hitherto been reported in the literature, there are no coexisting external abnormalities. In patients with congenital conductive hearing loss and with a normally appearing tympanic membrane, high-resolution CT examination is recommended prior to surgical exploration. Because of the high likelihood of injury to the facial nerve, reconstruction should not be attempted in the presence of the overlying facial nerve obscuring the oval window. As achievements in hearing may not be preserved over time, air conduction or implantable bone conduction hearing aids may be preferred in patients with stapes agenesis and absence of the oval window to minimize the risk for facial nerve injuries.

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