Bilateral Congenital Agenesis of Stapes and Oval Window in Two Members of a Family (Brother and Sister)

Case Report

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Abstract

Congenital agenesis of the stapes and the oval window is rare. Congenital stapedial agenesis (CSA) may be recognized preoperatively in the presence of conductive hearing loss. The principal radiological imaging approach of the temporal bone, computed tomography (CT), can be used to diagnose CSA. Our 17-year-old male patient (case A) had long-term hearing loss which was getting worse. A temporal bone CT scan revealed the absence of the stapes and the oval window on both sides and an abnormal position of the facial nerve. No anomalies were detected in the external ear structures. Explorative right ear tympanotomy revealed an abnormal inferior course and dehiscence of the facial nerve. The oval window and stapedial structures were absent. Patients were evaluated for continued hearing aid use or bone-anchored hearing aid implantation. Similar CT imaging and clinical abnormalities were seen in his 16-year-old sister (case B). They did not have any other siblings and neither of their parents nor any of their relatives had hearing loss. This report presents the CT scans of the two siblings with mixed hearing loss (mainly conductive) and the perioperative image of the first case. A genetic study may help explain the etiopathogenesis since both cases had similar clinical and imaging findings.

Keywords: Hearing loss, congenital anomaly, stapes, oval window, siblings, case report

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Introduction

Congenital absence of the stapes (CSA) together with the oval window is a very rare clinical entity. CSA can be recognized preoperatively in patients with isolated conductive hearing loss (CHL). Computed tomography (CT) is the main imaging method for temporal bone diagnosis. Understanding the anatomy and the embryological origins of the malformation is essential for accurate diagnosis and treatment. CSA may be associated with head and neck anomalies or syndromes (1).

The treatment options for CSA include the use of an air conduction hearing aid, or an implantable bone conductive hearing aid, or surgery. Surgical approaches are explorative tympanotomy with facial nerve rerouting, vestibulotomy, and ossicular reconstruction, or fenestration of the horizontal semicircular canal (2).

Case Presentation

In this report, we present the cases of two siblings with long-term and progressive hearing loss. Case A was a 17-year-old boy and case B was a 16-year-old girl. Their mother had no history of infection, disease, trauma, or drug use during pregnancy. The siblings had no history of disease, otitis media, tinnitus, trauma, or vestibular dysfunction. No pathologies were found on examination. Audiological tests of case A showed bilateral moderate-to-severe mixed hearing loss with a component of bilateral mild sensorineural hearing loss at low frequencies (Figure 1, discrimination 76%). The tests of case B showed bilateral moderate to severe mixed hearing loss with a component of bilateral slight sensorineural hearing loss (Figure 2, discrimination 80%). The mean (0.5 kHz, 1 kHz, 2 kHz, and 4 kHz) air-bone gaps were 36 dB in case A and 45 dB in case B. Weber's tests were in the middle and Rinne's tests were negative bilaterally. Ipsilateral and contralateral acoustic reflexes were negative. There was no nystagmus. Non-contrast axial and coronal plane CT scan (1-mm slice thickness) of the temporal bone revealed no stapes in the tympanic cavities and closed oval windows (Figures 3, 4). The tympanic segment of the facial nerve showed inferomedial course in both of the cases. There were no pathological findings in other ear structures or the mastoid cavity. In case A, exploratory tympanotomy verified the absence of the oval window and the stapes, dehiscence of the tympanic segment of the facial nerve, and the abnormal inferiomedial position of the facial nerve where the oval window should be on the lateral surface of the otic capsule (Figure 5). The malleus and the incus were mobile and intact. Genetic analysis was planned to determine the etiopathogenesis of the patients who had no comorbidities or clinical symptoms.

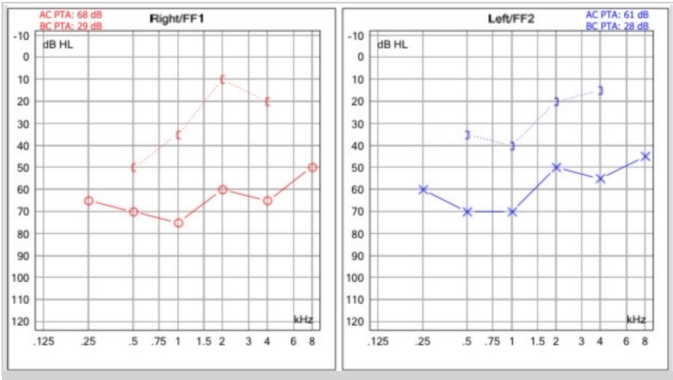

Figure 1. Audiological test of case A: bilateral moderate-to-severe mixed hearing loss with a component of bilateral mild sensorineural hearing loss at low frequencies

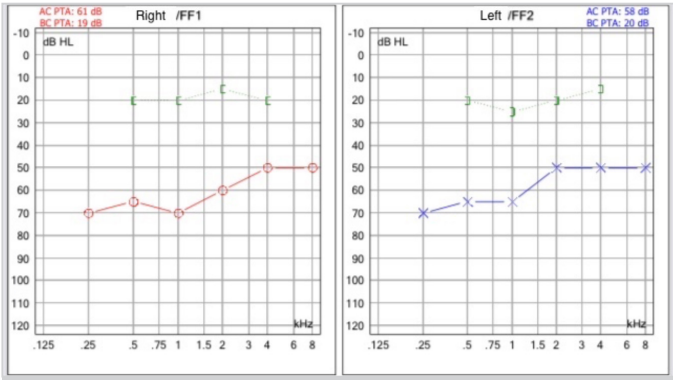

Figure 2. Audiological test of case B: bilateral moderate-to-severe mixed hearing loss with a component of bilateral slight sensorineural hearing loss

Written informed consent was obtained from the patients and the parents.

Discussion

CSA was first described by McAskile and Sullivan in 1955 (2). It is a rare entity of unknown etiology, with stapes and oval window agenesis linked to the abnormal development and course of facial nerve (1-4). A high-resolution temporal CT scan is recommended in cases suspected of congenital middle ear malformations (3). A CT scan can detect the

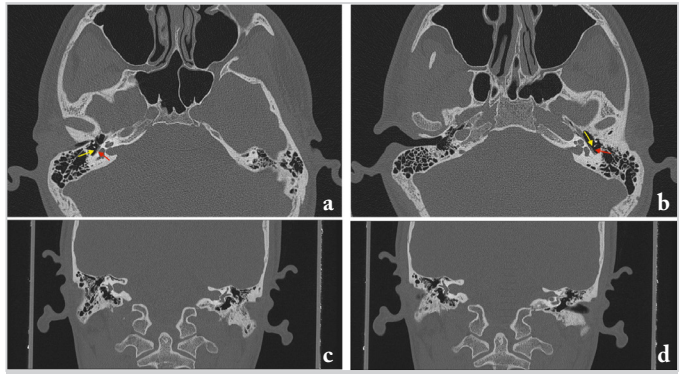

Figure 3. Case A: Right (a–c) and left (b–d) axial and coronal CT scans of the temporal bone showing the bilateral agenesis of the stapes (yellow arrow), the absence of the oval window, and the abnormal position of the facial nerve (red arrow); other ear structures appear normal

CT: Computed tomography

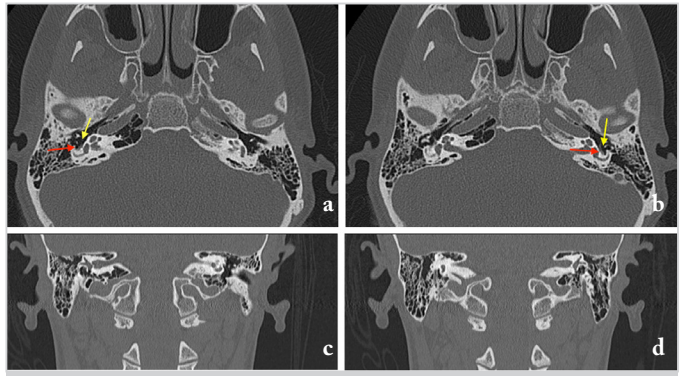

Figure 4. Case B: Right (a–c) and left (b–d) axial and coronal CT scans of the temporal bone showing the bilateral agenesis of the stapes (yellow arrow), the absence of the oval window, and the abnormal position of the facial nerve (red arrow); other ear structures appear normal

CT: Computed tomography

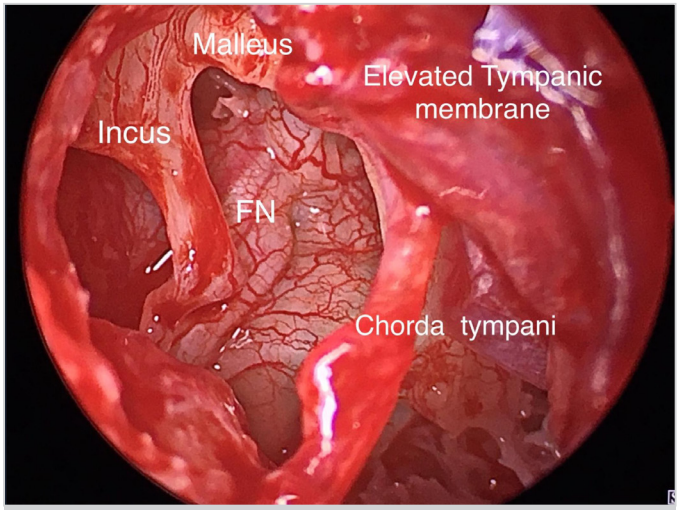

Figure 5. Exploratory tympanotomy image of case A verified the absence of the oval window and the stapes, the dehiscence of the tympanic segment of the facial nerve, and the abnormal inferomedial position where the oval window should be on the lateral surface of the otic capsule. The malleus and the incus are intact and mobile

FN: facial nerve

isolated absence of stapes, or stapes agenesis with oval window aplasia and abnormally located facial nerve in young patients with CHL (3). The stapes, the manubrium of the malleus, and the long process of the incus develop from the second branchial arch or Reichert's cartilage. The horizontal and vertical parts of the facial nerve become recognizable as the stapes develops toward the otic capsule at five to six weeks of pregnancy (1, 2). The stapes invaginates the otic capsule at seven weeks, determining the location of the future window. The tympanic part of the stapes footplate originates from Reichert's cartilage and the vestibular part from the otic capsule (5). If the facial nerve is displaced anteriorly, it prevents contact between the otic capsule and the stapes blastema. Contact is essential for stapes development (1, 2). Differences in this process could explain the observed variations (5). Delayed development of the first branchial arch can lead to the displacement of the facial nerve, and the development of the facial nerve strongly impacts the development of the stapes (1). However, the role of genetic abnormalities in etiopathogenesis is unknown.

CHL in the absence of a trauma, illness, or chronic medication use should be assessed for congenital middle ear abnormalities. Studies in the literature report that congenital hearing loss is usually more severe than in acquired cases (2, 3).

The typical CSA audiograms are flat, pure tone at speech frequencies, but not pathognomonic. It may not always be present, as in our cases (2). Interestingly, the observation of mixed hearing loss due to mild sensorineural hearing loss at low frequencies without dizziness in Case A could be due to a genetic etiopathogenesis such as enlarged vestibular aqueduct syndrome causing progressive sensorineural hearing loss (6).

Treatment in CSA is controversial due to a lack of publications (1). Treatment options include vestibulotomy, ossicular reconstruction, fenestration of the horizontal semicircular canal, and hearing aids (2). If possible, ossicular chain reconstruction is preferred. Displacement of the facial nerve between the remaining ossicles and the oval window is a problem. Because of the potential danger of abnormal course of the facial nerve, preoperative CT scanning is recommended to detect congenital CHL (1).

After the cases reported by Yi et al. (7), our cases are the second two siblings with bilateral CSA and oval window agenesis in the literature. The abnormally located facial nerve and the high risk of nerve damage due to the malformation seen in our cases suggested that the implantation of a hearing aid or bone anchored hearing aid (BAHA) instead of reconstruction would provide better functional results. It was decided to continue with the treatment with hearing aids, as the patients did not prefer BAHA for aesthetic reasons.

In conclusion, a collaboration between radiologists and clinicians in CSA cases contributes significantly to the diagnosis and planning of appropriate treatment, which should be individualized. A family history of CSA cases with mixed hearing loss suggests the necessity of genetic analysis.

Informed Consent: Written informed consent was obtained from the patients and the parents.

Peer-review: Externally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: H.G.D., R.Y., Concept: H.G.D., R.Y., Design: H.G.D., Data Collection and/or Processing: H.G.D., R.Y., Analysis and/or Interpretation: H.G.D., Literature Search: H.G.D., R.Y., Writing: H.G.D., R.Y.

Conflict of Interest: There is no conflict of interest to disclose.

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Main Points

- Our cases are the second two siblings in the literature with bilateral congenital stapedial agenesis and oval window agenesis.
- In the diagnosis of such cases, it is important to recognize the presence of an abnormally located facial nerve due to the high risk of nerve damage in the operation related to the malformation.
- The familial history suggests that genetic basis should be investigated.

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