Bilateral stapes agenesis: What can we find?

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ABSTRACT
We describe the youngest case of bilateral stapes agenesis and systematically reviewed the literature. Among those cases, 8/12 (66.7%) showed a bilateral agenesis. There is a male predominance 8/12 (66.7%). The most common associated anomaly reported being either a dehiscent or displaced facial nerve (7/12 – 58%). Furthermore, 5/12 patients (42%) had a narrow or an absent oval window. In our case, a 5-year-old male patient was referred for bilateral conductive hearing loss (air-bone gap 65 dB, bilateral). Operating findings on the left side included an inferiorly displaced facial nerve over the promontory, a narrow ossified oval window and an absent stapes. A stapedotomy with insertion of a modified stapes prosthesis allowed us to close the conductive gap. Even with possible good outcomes, surgical management should not be taken lightly, considering amplification is almost always a safe and effective option.

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Introduction
Among all possible middle ear anomalies, stapes agenesis is rarely part of the differential diagnostic for conductive hearing loss. First described in 1955 by McAskle and Sullivan [1], less than a dozen cases have been described so far. Even if uncommon, some key points have to be understood by the surgical team to ensure a safe and effective management. Mainly unrecognized, congenital stapes agenesis (CSA) may occur in patients with normal external auditory canal and tympanic membrane. So far, all cases have been reported separately and no common trend could have been extrapolated. Management varies from simple follow-up to vestibular fenestration. Thus, we report the youngest case of bilateral stapes agenesis, including his surgical management and follow-up. We also systematically reviewed all previous cases in the literature and analyzed their characteristics.

The case
A 5-year-old male patient was referred to our tertiary care center for bilateral conductive hearing loss. His past medical and surgical histories were negative, including perinatal complications or episodes of chronic middle ear effusion. The patient did not undergo newborn hearing screening test. His family history was negative for any otologic or genetic problem. He was initially referred to an otolaryngologist in another center for speech-delayed at 2 years old. Investigations for congenital deafness were performed in that center and revealed bilateral symmetric moderate conductive hearing loss (Figure 1(a) – threshold at 65 dB bilaterally, discrimination 100%). The patient successfully received bilateral hearing aids at that point. He had a positive evolution until our consultation, where his parents were concerned by school problems and a non-optimal audition. A middle ear computed tomography scan (CT-scan) revealed the absence of stapes suprastructures bilaterally, ossified oval window and an abnormal facial nerve location (Figures 2 and 3). Tympanic membranes and facial nerves function were both normal. After discussion, a left-sided exploratory tympanotomy with possible stapedotomy was planned.

Operative findings included, after drilling the tympanic rim and freeing the chorda tympani, an inferiorly displaced facial nerve without bony covering over the promontory, as seen on the CT-scan. The identification of the facial nerve was confirmed by electric stimulation (Medtronic Nerve Integrity
Monitor [NIM] Response). Then we could see that the long process of the incus was abnormally sloped, and its tip was fused with the posterior mesotympanic wall (anterior to the sinus tympani). The oval window was narrow and shallow, without any footplate. Moreover, there was not any evidence of stapes suprastructures (Figure 4). The attachment of the long process of the incus had to be drilled to allow the visualization of the abnormal footplate, and to allow normal mobility of the ossicular chain. A stapedotomy was done using first a 0.8 mm diamond burr for blue-lining. Then, we used the 0.6 mm microdrill. The stapedotomy prosthesis (Medtronic Big Easy™ Offset 5 mm) was inserted and crimped. The prosthesis had to be modified to fit the complex anatomy and OtoMimix® cement was then applied to secure it on the long process of the incus. The rest of the operation was uneventful.

His 12-month postoperative audiogram (Figure 1(b)) confirmed a significant gap closure (PTA gain of 42 dB). The patient reported a better understanding without his hearing aids. Other major benefits included the ability to enjoy aquatic activities without hearing aids, to hear potential hazard such as smoke...
alarm and to prevent any learning deprivation from broken hearing aids. Right-sided surgery will be scheduled when the patient reach adulthood when he could consent.

**Literature review results**

Among the 12 reported cases of stapes agenesis in literature (Table 1) [1–8], 8/12 (66.7%) showed a bilateral agenesis. There is a male predominance 8/12 (66.7%). Interestingly, two pairs of siblings were reported [5,8]. The most common associated anomalies reported being either a dehiscent facial canal or displaced facial nerve (7/12, 58%). Furthermore, 5/12 patients (42%) had a narrow or absent oval window with 3/12 patients (25%) with normal oval window and unreported findings for the remaining patients (4/12, 33%). There was no predominant management for these patients. No predominant syndrome was reported.

**Discussion**

The CSA is obviously a rare entity with only 12 cases reported in the literature, including ours. In all cases, CSA presents as a conductive hearing loss with normal external auditory canal and intact tympanic membrane. Deafness severity could vary according to associate anomalies, but mainly represents a maximum conductive gap.

Various managements have been described through time. This includes a broad range of options, from exploratory surgery and reconstruction prosthesis to amplification. The choice should be a shared decision between the patient and the surgeon. Nonetheless, we
have to keep in mind that most of these cases represent a conductive hearing loss and patients can easily and safely use hearing aids. In this present case, the patient could benefit from a stapedotomy and prosthesis. One key point is the ability of the surgeon to safely perform the surgery with direct visualization.

Table 1. Congenital stapes agenesis reported in the literature.

| Study/year            | Sex/age deafness | Footplate, oval or round window | Associated malformation                                      | Management/results                      |
|-----------------------|------------------|--------------------------------|-------------------------------------------------------------|-----------------------------------------|
| McAskile and Sullivan (1955) [1] | M/7-year-old unilateral NA | Oval window remnant            | Exploration only                                           |
|                       | M/24-year-old unilateral NA | Malformed, fixed incus and malleus Anomalous course facial nerve Microtia Fixed incus | Fenestration                             |
| Fisher et al. (1982) [2] | M/6-year-old bilateral Mobile footplate | Cervical spine anomaly Paddle-shaped + broad long process of the incus Narrow oval window Displaced facial nerve | Amplification NOS                          |
| Reiber et al. (1997) [3] | M/38-year-old bilateral Normal round window | Dehiscent facial nerve Malformed long process incus + fibrous band to promontory Displaced, dehiscent facial nerve | NA Amplification (hearing aid)          |
| Keskin et al. (2003) [4] | M/17-year-old bilateral Hidden by facial nerve | Absent oval window | Lempert’s fenestration horizontal semicircular canal (1 ear) Fenestration |
| Yi et al. (2003) [5]    | M/17-year-old bilateral No oval window Normal round window | Absent oval window | BAHA                                                      |
| Rodriguez et al. (2005) [6] | F/20-year-old bilateral No oval window Normal round window | F/20-year-old bilateral Mobile footplate | House prosthesis ABG 20 dB Goldenberg prosthesis PTA 45 dB ABG 40 dB BAHA |
| Casqueiro et al. (2009) [7] | M/23-year-old unilateral Mobile footplate | Absence of long process incus Fixed incus body | BAHA                                                      |
| Undabeitia et al. (2013) [8] | F/19-year-old bilateral Hypoplastic oval window | Displaced facial nerve Hypoplastic oval window | BAHA                                                      |
| F/22-year-old bilateral Hypoplastic oval window | Displaced facial nerve Hypoplastic oval window | BAHA                                                      |
| Bergeron and Côté (2017) | M/5-year-old bilateral Narrow footplate window | Displaced facial nerve | Stapedotomy with prosthesis                                |

Figure 4. Intraoperative images. (A) Facial nerve (*) and long incus process (+). (B) Long incus process curved toward sinus tympani. Chorda tympani (arrow). (C) Drilled incus, facial nerve, chorda tympani with the oval window without stapes (#). (D) Stapedotomy first step with a 0.8 mm diamond burr then with a 0.6 mm cutting burr (E) Medtronic Big Easy™ offset 5 mm placed over the stapedotomy. (F) Prosthesis and OtoMimix® in place (G) OtoMimix® over prosthesis and fat graft on oval window. (H) Facial nerve over the promontory above the round window niche with OtoMimix® on prosthesis.
of present landmarks. The footplate – even if abnormal – could be easily visualized in our case. Furthermore, the facial nerve was also under direct visualization, preserving its integrity during key points of the procedure. We also used NIM for the entire procedure to confirm its location.

Like the majority of the reported cases, our patient had an abnormal trajectory of the facial nerve. CT-scan should be ordered before any exploratory surgery. This might facilitate the surgical planning and the selection of the ear as other anomalies are commonly encountered. Facial nerve monitoring should also be considered. NIM could be of great help during that kind of case.

Interestingly, 2/3 of patients in the literature had a bilateral agenesis. Like most ear surgeries, sequential surgeries should be done to prevent potential – even if rare – catastrophic complications, such as bilateral sensorineural hearing loss. We should not forget that hearing aids is almost always a good option – both safe and effective – especially for pediatric patients. Any surgical intervention should be carefully evaluated and discussed with the patient and/or the parents.

**Conclusions**

CSA presents as conductive hearing loss, which may be bilateral in most cases. It is also more prevalent for male patients. Furthermore, more than half of the patients have an abnormal facial nerve, which may complicate the surgery. Round or oval window anomalies are also present in the majority of these patients. All those factors have to be considered to ensure a safe and effective management. Surgical management should not be taken lightly, considering amplification is almost always a safe and effective option.

**Disclosure statement**

Authors have no conflicts of interest to declare.

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