Isolated Congenital Round Window Atresia: Report of 2 Cases

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Abstract

Round window atresia (RWA) is an uncommon condition and can result in a conductive hearing loss. Two cases of nonsyndromal bilateral RWA in 2 members of the same family are reported. Both cases presented with a conductive hearing loss of 20 to 30 dB. High-resolution computed tomography scanning was used to diagnose the condition. The patients were rehabilitated with hearing aids. Review of the literature has shown disappointing results in hearing improvement with cochlear fenestration in an attempt to address this condition. Patients presenting with unexplained conductive hearing loss should be offered computed tomography scanning. The cases we report add to the literature to benefit future patients in preoperative counseling and better inform management.

Keywords

round window atresia, round window aplasia, middle ear malformation, surgery, round window drillout, computed tomography

Introduction

Round window atresia (RWA) is a rare condition that can occur as a manifestation of an underlying syndromic disorder or as an isolated anomaly. In the former, it is associated with Treacher Collins syndrome or Branchio-oto-renal syndrome.1,2 Round window atresia can also arise from otosclerosis.3,4 Round window atresia was first described by Scarpa and concurrent abnormalities of the ossicle are often noted.1,2 Non-syndromal, isolated RWA anomalies are exceedingly rare, and to date, less than 15 cases have been reported in the literature.2 Reports seem to point toward a familial association with this anomaly.1,2

Clinically, RWA can mimic otosclerosis, and concurrent stapes footplate ankylosis is sometimes seen. Nevertheless, hearing results following stapedectomy have remained unimpressive.5 Some authors have attempted to recreate the round window by performing cochlear fenestration, but results have so far been disappointing.5-8

The 2 patients presented in this case study are mother and son. Apart from detailing this familial association, we sought to highlight the role of computed tomography (CT) in the diagnosis of this condition. The different management options are reviewed.

Case Reports

Case 1

A 33-year-old woman presented in December 2016 for consultation regarding a long-standing hearing loss. There was no tinnitus or vestibular symptoms nor other otological history. On physical examination, the vestibular system and cranial nerves were normal. Otoscopy was unremarkable. Tuning fork test (512 Hz) did not demonstrate any reversal (positive Rinné).

Pure tone audiometry showed a bilateral conductive loss from 250 to 2000 Hz with an air-bone gap averaging 20 dB. Her speech discrimination scores were good at elevated thresholds. Type A tympanograms were obtained. Ipsilateral and contralateral acoustic reflexes were absent bilaterally.

High-resolution computed tomography (HRCT) scan (Figure 1) revealed bilateral RWA. The ossicular chain was intact with no other abnormality seen. There was no radiological sign of otosclerosis.

She was counseled on the unpredictability of outcome and possible limited benefit of surgery. The patient opted to proceed with hearing aid fitting.

Case 2

A 6-year-old boy with a normal prenatal and birth history was first seen in the clinic in April 2014 with recurrent otitis media...
with effusion (OME). Audiogram showed bilateral conductive hearing loss across all frequencies with an air-bone gap of 20 to 30dB. He had bilateral type C tympanograms. Ipsilateral reflexes and distortion product otoacoustic emissions were absent bilaterally.

The OME was treated conservatively and resolved, but the hearing loss remained unchanged on serial audiological testing. He was given hearing aids and was coping well at school. No deterioration in balance was reported.

After his mother (presented above) was diagnosed with bilateral RWA, an HRCT scan was requested and showed absence of the round window niche bilaterally (Figure 2). His parents have opted for nonsurgical management of his pathology and he is to continue with hearing aids and serial audiometry.

Discussion

Conductive hearing loss has a broad range of causes—otosclerosis, chronic otitis media, and congenital middle ear anomalies are some of the differential diagnoses to consider. Nonsyndromal, isolated, congenital RWA causing conductive hearing loss is exceedingly rare and only a handful of case reports are described in the literature (Table 1).1,2,5-10 The 2 patients in our series are related (mother and son). Reports in the literature have highlighted a familial association related to this anomaly, thereby supporting the hypothesis that this condition is genetically inherited.1,2,9 It is postulated that it may be inherited in an autosomal dominant fashion with variable penetrance, although no specific genetic mutation responsible for RWA has been identified to date.2

The audiometric findings in patients with RWA are interesting and challenge the traditional notions of hearing physiology. As can be seen in our 2 patients and patients listed in Table 1, the baseline air-bone gap ranges from 10 to 40 dB. Remarkably, no patient presented with maximal conductive hearing loss. It is traditionally accepted that the presence of the 2 mobile middle ear windows (round and oval windows) allow bulk motion of the functionally incompressible inner ear fluid.2 In RWA, sound pressure arriving at the stapes footplate cannot result in an outward displacement of equal volume of inner ear fluid at the nonexistent round window. Interestingly, this did not result in maximal conductive hearing loss. Previous authors have hypothesized that in this situation, the cochlea must be also sensitive to an alternative method of stimulation,9 or there exists an alternative pressure outlet from the cochlea (a third window).11

Figure 1. Case 1: Axial CT serial 1-mm sections showed absence of round window (right). The appearances in the left inner ear were identical. CT indicates computed tomography.

Figure 2. Case 2: Axial CT serial 1-mm sections demonstrating round window atresia (left). The appearances in the right inner ear were similar. CT indicates computed tomography.
Our case series, as well as reports from the literature, have indicated that HRCT is a sensitive tool in the diagnosis of this condition. There were instances where the RWA was not diagnosed during the initial middle ear exploration, only for the condition to be subsequently diagnosed radiologically. It supports the notion that this condition may be missed unless the round window niche is specifically looked for during tympanotomy.

In the literature, only a few reports on the outcome after surgery can be found and no definitive recommendation can be made given the relative paucity of data. Shea et al found that 1% of patients with otosclerosis have obliteration of the round window. In this subset of patients, the majority does not have significant hearing improvement following stapedectomy and would still need hearing aids. Also, no significant improvement in hearing was noted after stapedectomy in RWA unrelated to otosclerosis. Promontory fenestration at the anticipated area of the round window was performed in a few cases, with marginal improvement in hearing. In some cases, the authors have noted a membrane following drilling away of promontory bone with the membrane noted to be moving in conjunction with stapedial motion.

There are many reports of various congenital anomalies of the temporal bone in a variety of different settings and with today's advanced radiology, these lesions can often be diagnosed more easily. Although this small series is not a significant new addition to the literature on this topic, the management of this condition remains controversial. Our patients were rehabilitated with hearing aids. We are cognizant of the fact that promontory fenestration runs the risk of causing sensorineural hearing loss and debilitating vestibular symptoms. No long-term deterioration in air-bone gap was seen in our patients, or patients in the literature, suggesting that this is a nonprogressive condition.

In conclusion, isolated, nonsyndromal RWA is an uncommon malformation. It can mimic otosclerosis. Modest surgical benefits reported in the literature favor hearing aid rehabilitation. This condition can be diagnosed with HRCT. Patients presenting with unexplained conductive hearing loss should be investigated with CT scan.

**Authors’ Note**
All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study.

**Declaration of Conflicting Interests**
The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

**Table 1. Data of Reported Cases of Nonsyndromal Round Window Atresia.**

| Paper (Year)        | Gender | Laterality | Other Pathology                  | Degree of Hearing Loss (dB) | Treatment                                      | Postoperative Air-Bone Gap (dB) | Other Affected Family Members |
|---------------------|--------|------------|----------------------------------|-----------------------------|------------------------------------------------|--------------------------------|-------------------------------|
| Harrison et al (1964) | F      | Bilateral | Nil                               | 10-40                       | Cochlear fenestration                            | Up to 20                       | –                             |
| Ombredanne et al (1968) | M      | Bilateral | Deformity of external ear          | 15-25                       | Cochlear fenestration                            | Up to 20                       | –                             |
| Richards et al (1981) | M      | Bilateral | Nil                               | 10-40                       | Cochlear fenestration                            | Up to 20                       | –                             |
| Clifford et al (1990) | F      | Bilateral | Stapes ankylosis                  | 30                          | Cochlear fenestration (previous stapes surgery)  | –                              | –                             |
| Pappas et al (1998)  | M      | Bilateral | Stapes ankylosis                  | 40                          | Stapedectomy and bucket handle prosthesis        | 40                             | –                             |
|                    | F      | Bilateral | Stapes ankylosis                  | 30-40                       | Stapedectomy and bucket handle prosthesis        | 30-40                         | Twin brother has same pathology |
|                    | M      | Unilateral | –                                | 30                          | Nil                                             | NA                             | Twin sister has same pathology |
| Martin et al (2002)  | F      | Bilateral | Nil                               | 20-50                       | Middle ear exploration                          | 20-50                         | –                             |
| Linder et al (2003)  | F      | Unilateral | Absence of stapes tendon, malleus fixation, long and bent incus | 5-25                       | Incus interposition                             | Slight improvement             | Daughter has same pathology |
|                    | F      | Unilateral | Normal                            | 10-30                       | –                                               | NA                             | Mother has same pathology    |
| Bormann et al (2007) | F      | Bilateral | Stapes ankylosis                  | 20                          | Revision stapes surgery                          | Up to 20                       | Son has same pathology        |
|                    | M      | Bilateral | –                                 | 10-20                       | Nil                                             | NA                             | Mother has same pathology     |

Abbreviations: –, not reported; NA, not applicable.
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References
1. Pappas DG Jr, Pappas DG Sr, Hedlin G. Round window atresia in association with congenital stapes fixation. Laryngoscope. 1998;108(8 Pt 1):1115-1118.
2. Borrmann A, Arnold W. Non-syndromal round window atresia: an autosomal dominant genetic disorder with variable penetrance? Eur Arch Otorhinolaryngol. 2007;264(9):1103-1108.
3. Adkins WY Jr., Gussen R. Oval window absence, bony closure of round window, and inner ear anomaly. Laryngoscope. 1974;84(7):1210-1224.
4. Shea JJ, Farrior JB. Stapedectomy and round window closure. Laryngoscope. 1987;97(1):10-12.
5. Clifford AR, Fagan PA, Doust BD. Isolated congenital round window absence. J Laryngol Otol. 1990;104(12):980-981.
6. Harrison WH, Shambaugh GE Jr, Derlacki EL. Congenital absence of the round window: case report with surgical reconstruction by cochlear fenestration. Laryngoscope. 1964;74:967-978.
7. Ombredanne M. Congenital absence of the round window in certain minor aplasias. Further cases. Ann Otolaryngol Chir Cervicofac. 1968;85(5):369-378.
8. Richards SH. Congenital absence of the round window treated by cochlear fenestration. Clin Otolaryngol Allied Sci. 1981;6(4):265-269.
9. Linder TE, Ma F, Huber A. Round window atresia and its effect on sound transmission. Otol Neurotol. 2003;24(2):259-263.
10. Martin C, Tringali S, Bertholon P, Pouget JF, Prades JM. Isolated congenital round window absence. Ann Otol Rhinol Laryngol. 2002;111(9):799-801.
11. Tonndorf J, Tabor JR. Closure of the cochlear windows: its effect upon air- and bone-conduction. Ann Otol Rhinol Laryngol. 1962;71:5-29.
12. Thomeer H, Kunst H, Verbist B, et al. Congenital oval or round window anomaly with or without abnormal facial nerve course: surgical results for 15 ears. Otol Neurotol. 2012;33(5):779-784.