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Inner ear anomalies in children with isolated unilateral congenital aural atresia

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ABSTRACT

Objectives/Hypothesis: We aim to define the frequencies of anomalies of the inner ear, oval window, and round window ipsilateral to isolated non-syndromic unilateral aural atresia.
Methods and materials: Retrospective case series. We reviewed high resolution computed tomography scans of the temporal bones of 70 children with isolated non-syndromic unilateral congenital aural atresia. Scans were reviewed according to the Jahrsdoerfer criteria and further evaluated for anomalies of the vestibule, semi-circular canals, cochlea, internal auditory canal and vestibulocochlear nerve.
Results: Inner ear dysplasia was seen in two of 70 atretic ears: one with a dysmorphic lateral semicircular canal and another with a large vestibule assimilating the lateral semicircular canal. Abnormalities of the oval window and round window ipsilateral to the atresia were identified in 21% (15) and 7% (5), respectively, of the atretic ears. Oval window and round window abnormalities were associated with disproportionately lower Jahrsdoerfer scores compared to aural atresia patients without these abnormalities ($P < 0.001$ and $P = 0.04$, respectively).
Conclusion: Compared to studies that included syndromic or bilateral atresia cases, we found inner ear and oval window abnormalities less common in children with isolated non-syndromic unilateral aural atresia. However, round window anomalies seem to occur at about the same frequency.

1. Introduction

Craniofacial microsomia (CFM) is a syndrome that includes a wide spectrum of phenotypes and severity related to malformations of the first and second pharyngeal arch derivatives [1]. Congenital aural atresia (CAA) is commonly seen in CFM, and isolated cases of CAA have been proposed to represent a variant of this disorder.

The first and second pharyngeal arches, which are influenced by the neural crest, give rise to the middle ear structures while the inner ear is derived separately from thickenings of surface ectoderm called the otic placodes that ultimately develop into the membranous labyrinthine structures, the oval window, and the round window niche [2,3].

Abnormalities of the oval window, round window and inner ear are documented in the setting of congenital aural atresia (CAA) [4–7] and craniofacial microsomia [1,8]. However, these studies have included patients with bilateral and/or syndromic cases of CAA. It is unclear whether the prevalence of these abnormalities is the same in children with isolated unilateral non-syndromic congenital aural atresia.

The aim of this study is to determine the frequencies with which radiologically apparent abnormalities of the inner ear, oval window, and round window occur in children with isolated non-syndromic unilateral congenital aural atresia.

2. Materials & methods

The University Institutional Review Board approved this retrospective self-controlled case series study of previously-acquired computed tomography (CT) scans of the temporal bone in children with unilateral CAA.

2.1. Subjects

Study subjects were selected by identifying patients, age birth

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through 18 years of age, who underwent CT of the temporal bones for congenital aural atresia. The patients were identified through the ISYS database query search of radiology reports at Children's Healthcare of Atlanta for the years 2002 through 2012. Children with bilateral aural atresia and those with a syndrome or other congenital structural abnormality or malformation of the head and neck in conjunction with aural atresia were excluded from the study. Cases with bilateral atresia were identified by viewing the CT scans. Cases with syndromic associations or concomitant head and neck anomalies were identified by the available diagnosis and problem lists in the medical record and/or during review of the CT scans. For CAA patients at this institution, individual practitioner and patient idiosyncrasies may prompt additional evaluations (e.g., renal ultrasound and consultations with medical genetics, cardiology and ophthalmology).

2.2. Identification of middle & inner ear abnormalities

A subspecialty-certified neuroradiologist with experience in pediatric neuroradiology (BPS) at an academic children's hospital reviewed the CT scans in the axial, coronal and sagittal planes using the General Electric Centricity™ Universal Viewer software. For the atretic ear, the Jahrsdoerfer score (Table 1) [9] was determined. In addition, the following inner ear structures were also assessed for the atretic ear: (1) vestibule, (2) semi-circular canals, (3) cochlea, (4) internal auditory canal and (5) vestibulocochlear nerve. The non-atretic ear was also evaluated for the presence or absence of abnormalities.

2.3. Data analysis

Continuous variables were summarized as mean ± standard deviation. Jahrsdoerfer scores were summarized with median and range. The Wilcoxon Rank Sum test for independent samples was used to compare the Jahrsdoerfer scores of children with and without abnormalities of the oval window and round window. All tests used a *P*-value of 0.05 as the threshold for significance. Analysis was performed using SAS JMP 12 software (2015; SAS Institute, Cary, NC).

3. Results

A total of 108 CT scans of children with CAA were identified; 70 were included in our study after exclusions (Fig. 1). The mean age of the study population at the time of CT scan was 61.8 ± 43.6 months and ages ranged from under one month to 206 months. Males comprised 54% of the group, and the atretic ear was on the right side in 70% of cases. The median Jahrsdoerfer score was 9 and ranged from 0 to 10. No correlation of Jahrsdoerfer score with age at CT scan was found (Spearman $r = 0.01$). The child with imaging obtained at age 1 month had a Jahrsdoerfer score of 8.

Table 1
Jahrsdoerfer criteria as used in the study.

Points	Parameter
2	Stapes present
1	Oval window open
1	Middle ear space large and favorable
1	Malleus-incus complex well-formed
1	Mastoid well-pneumatized
1	Incus and stapes connected
1	Round window present and open
1	Appearance of external ear is normal (by CT)
1	Facial nerve position
10	Total available points

Adapted from Jahrsdoerfer et al. [9].
CT = computed tomography.

Inner ear dysplasia was seen in just two cases (3%). Ipsilateral to the atresia, one female (age 59 months at the time of CT) had a dysmorphic lateral semicircular canal (SCC) composed of a laterally projecting outpouching of the vestibule without anterior or posterior crura (Fig. 2a); in the contralateral (non-atretic) ear, the lateral SCC was mildly hypoplastic (Fig. 2b). The second patient, a male (age 55 months at the time of CT), exhibited a large vestibule assimilating the lateral semicircular canal on the side of the atresia (Fig. 3). The Jahrsdoerfer scores of the patients were 2 and 5, respectively, and in both cases, there were concomitant abnormalities of the oval window, facial nerve and middle ear space on the side of the atresia.

Ipsilateral to the atretic ear, the oval window was abnormal in 15 (21%) cases. The median Jahrsdoerfer score in these patients (5, range 0–9) was lower than in children without oval window anomalies (9, range 2–10; $Z = -4.54$, $P < 0.001$). The most common abnormalities accompanying an abnormal oval window were of the middle ear space (12 of 15 ears) and the facial nerve (10 of 15 ears).

Round window abnormalities on the side of the atresia were apparent in 5 (7%) cases. The median Jahrsdoerfer score in these patients (4; range 0–8) was lower than in ears without round window anomalies (9, range 1–10; $Z = -2.73$, $P = 0.006$). Co-existing abnormalities of the facial nerve, oval window and middle ear space were each noted in 4 of 5 ears. No abnormality of the oval window or round window was noted for the contralateral non-atretic ear.

4. Discussion

Inner ear anomalies were uncommon in this study of children with isolated unilateral congenital aural atresia (CAA). They were seen in two children with more severe manifestations of aural atresia, and both involved the lateral semicircular canal (SCC) and vestibule. In the atretic ear, 7% patients had abnormal round windows, whereas abnormalities of the oval window were more common (21%). Based on review of the radiology reports, the contralateral, non-atretic ear was normal in all but one case in which it co-existed with an inner ear abnormality on the atretic side.

Vrabec and Lin [4] found abnormalities of the inner ear in 23 (22%) of 105 children with aural atresia, 19 of whom had bilateral atresia. The most common of these abnormalities was vestibular dysplasia involving the lateral SCC. In a study of 66 patients with complete unilateral or bilateral atresia, Naunton and Valvassori [5] found radiologic evidence of inner ear abnormalities in 7 (11%). Among 113 microtic or atretic ears in 92 patients, Mayer et al. [6] found abnormalities of the inner ear in 38 (34%). The most common of these were dysplasia of the lateral SCC or vestibule. This study also reported an absent oval window and closed round window in 36% and 6% of microtic ears, respectively. Phelps [7] found inner ear abnormalities in 34 (22%) of 157 children with congenital microtia and/or aural atresia with lateral SCC dysplasia being most common. However, of the children with inner ear anomalies 26 were exposed in utero to thalidomide, a teratogen known to cause a wide variety of craniofacial and inner ear defects [10]. Among 33 cases of unilateral hemifacial microsomia examined by Rahbar et al. [8], 32 (97%) had some degree of aural atresia, 12 (36%) had hypoplastic oval windows and two (6%) had atretic round windows. A hypoplastic vestibule was seen in two of these patients, and hypoplastic semicircular canals and abnormal cochlea were each present in one case.

Compared to previous studies of children with unilateral or bilateral CAA, anomalies of the inner ear and oval window appear to