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Utilization of diagnostic testing for pediatric sensorineural hearing loss☆

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ABSTRACT

Background/Objective: Sensorineural hearing loss is a common diagnosis among children. The diagnostic workup varies widely among practitioners. This study's aim was to assess the utilization of diagnostic testing for SNHL and determine the yield of each test.
Study design: Retrospective chart review.
Setting: Tertiary care center.
Subjects: 827 patients with a diagnosis of SNHL from January 1, 2011 to January 1, 2015.
Results: 746 patients met inclusion criteria. Temporal bone imaging was performed on 561 (75%) of patients with 224 (40%) having positive results that explained the etiology of the SNHL. Congenital SNHL was more likely to be associated with abnormal imaging than acquired SNHL (109/299 versus 106/316 respectively) ($p = 0.001$). Unilateral SNHL was more likely to be associated imaging abnormalities than bilateral SNHL (101/221 and 123/340 respectively) ($p = 0.028$). Genetic testing was performed on 244 (33%) patients, of which 94 (39%) had abnormalities. Positive genetics results were more common with bilateral than unilateral SNHL (82/191 and 12/53 respectively) ($p = 0.007$). There was no statistically significant difference in the utility of genetic testing for congenital and acquired SNHL ($p = 0.0836$). Cytomegalovirus (CMV) testing was available for 104 (14%) of patients with 13 (12.5%) being positive and consistent with congenital CMV. Electrocardiogram, urinalysis, and Lyme titers were less useful.
Conclusions: Imaging and genetic testing had the highest yield in the evaluation of children with SNHL and were the most commonly performed. CMV testing was valuable in neonates who failed newborn hearing screening.

1. Introduction

Sensorineural hearing loss (SNHL) in children is common in developed countries. The estimated incidence of SNHL is 1–4/1000 [1,2] children at birth and 6/1000 by the age of 18 [3]. A wide range of conditions can lead to SNHL that can be generally categorized genetic versus non-genetic and congenital versus acquired. Genetic etiologies account for approximately 50% of cases of congenital SNHL [2]. Approximately 70% of genetic hearing losses are non-syndromic and 30% are associated with an underlying syndrome [2]. Non-genetic etiologies include infections such as congenital cytomegalovirus (CMV), ototoxic medications, maternal drug use, low Apgar scores, prematurity, and other environmental insults [2,4]. Early detection and treatment is important for speech and language development and meeting academic and social milestones [5,6].

Given the challenging nature of finding an etiology for the hearing loss, a thoughtful diagnostic approach is necessary. Although a complete history and physical examination are important, these often have low diagnostic yield. Therefore, further workup including genetic testing, imaging, lab work, and other diagnostic studies is often employed [1,7]. Many efforts have been directed to provide a diagnostic algorithm [2,4,8], yet different practices have different diagnostic approaches. There is no clear dominant strategy for any specific set of tests [9–12]. Choosing to perform a test can depend on many factors, including severity and type of hearing loss, associated signs and symptoms, and family preference [13].

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This study demonstrates the utility of different diagnostic approaches to SNHL in children at a tertiary care center. Particular attention was paid to the types of testing employed and the results of such testing. The purpose of this study is to assess the protocol for diagnosis and evaluation of SNHL at this institution. Findings were compared to the recent recommendations by the International Pediatric Otolaryngology Group (IPOG). [8] The study then looked at the diagnostic yield of the testing performed, comparing these results across different SNHL categories (congenital versus acquired and unilateral versus bilateral) and SNHL levels.

2. Methods

A retrospective chart review was performed for patients presenting to our institution from January 1, 2011 to January 1, 2015. 827 charts were available for review from the practices of five otolaryngologists. The Massachusetts Eye and Ear Institutional Review Board (IRB) approved this study (IRB protocol 15–025H).

Patients who were newborn to age 18 with a diagnosis of SNHL or hearing loss were included in the initial chart review. The presence of one audiogram in the chart to confirm the diagnosis was required for inclusion. Patients were excluded if there was not an audiogram to confirm the diagnosis or if the hearing was normal on audiometry or not sensorineural in nature.

Data extracted included age at presentation, gender, age at diagnosis of hearing loss, results of the newborn hearing screen, laterality of hearing loss, degree of hearing loss, and onset of hearing loss. If available, diagnostic test results were gathered from computed tomography (CT) scans, magnetic resonance imaging (MRI) scans, electrocardiogram (EKG), urinalysis, cytomegalovirus (CMV) testing, Lyme titers, and genetic testing. History of hearing loss in the family, prematurity, NICU stay, IV antibiotic use, jaundice, maternal insults during pregnancy, perinatal events, meningitis, or other factors that may contribute to hearing loss was obtained. All patient information was de-identified.

Hearing loss was considered congenital if it was diagnosed in the first year of life. In cases of sloping hearing loss, the lowest level of hearing loss recorded was used for classification of the degree of loss. Asymmetric hearing loss was considered bilateral hearing loss for analyses where laterality was considered.

Finally, the diagnostic utilities of individual tests including imaging, genetic testing, EKG, urinalysis, Lyme titers, and CMV were evaluated. A positive CMV result was considered diagnostic only if testing was performed less than one month after birth, or if additional findings correlating to the diagnosis of congenital CMV infection were noted, such as microcephaly or MRI findings suggestive of congenital CMV infection.

A Fisher's exact test was used to compare imaging and genetic testing results for both congenital versus acquired and unilateral versus bilateral SNHL. An alpha of $p < 0.05$ was used to define statistical significance. All analyses were performed using Microsoft Excel.

3. Results

Of the 827 charts reviewed, 746 met inclusion criteria. Patient demographics are listed in Table 1. Fig. 1 shows the levels of hearing loss broken down into unilateral, bilateral, congenital, and acquired. Of note, 20 patients did not have information in the chart to classify the hearing loss as congenital or acquired. Table 2 demonstrates the utilization and diagnostic yield of CT, MRI, and genetic testing across the different levels of hearing loss broken down into unilateral, bilateral, congenital, and acquired.

3.1. Imaging

Imaging was performed on 561 (75%) patients with 224 (40%) having positive results that explained the etiology of the SNHL. CT results were available for 480 (64%) of patients and MRI results were available for 207 (28%) of patients. There were 126 (17%) patients who had both CT and MRI performed. CT was positive in 180/480 (38%) patients. Positive CT results are shown in Table 3. The most common positive finding was cochlea-vestibular abnormalities. This was found in 71/180 (39.5%) of patients with abnormal CT scans. MRI was abnormal in 84/207 (41%) of patients. These findings are detailed in Table 4. The most common positive finding was cochlear nerve deficiency (hypoplasia or aplasia of the cochlear nerve) which was found in 27/83 (33%) patients with an abnormal MRI. For the patients who had both CT and MRI performed, both CT and MRI were abnormal in 40 (32%) patients. Of note, there were 6 CT scans that were normal that had abnormal MRI results. MRI found cochlear nerve deficiency in 4 of the patients (all who had profound SNHL) and inflammation of the cochlear nerve in 2 of the patients. In addition, there were 3 MRIs that did not have abnormal findings, but the CT was abnormal. Dehiscence

Table 1
Patient demographics and Comorbidities.

Age at diagnosis	0-18 (mean 4.3) (median 4)
Male	387 (52%)
Female	359 (48%)
Type of hearing loss	Congenital: 304 (41%) Acquired: 422 (56%) Unknown: 20 (3%)
Level of hearing loss	Mild (15–40 dB): 179 (24%) Moderate (41–70 dB): 213 (29%) Severe (71–90 dB): 128 (17%) Profound (> 90 dB): 226 (30%)
Family history of hearing loss	Yes: 180 (24%) (1st degree relative (123), 2nd degree relative (38), 3rd degree relative (12), 4th degree relative (7)) No: 507 (68%) Unknown: 59 (8%)(7 were adopted)
Family history of genetic abnormality	6 (1%)(5 Connexin 26 mutations and 1 branchio-oto-renal syndrome)
Parents are cousins	1 (< 1%)
Known genetic syndrome (no genetic testing performed)	16 (2%)
Maternal insults during pregnancy	Yes: 49 (6%) (ingestion of substances or medications not approved in pregnancy (12), severe infection requiring hospitalization (11), preeclampsia (7), gestational diabetes (7), known CMV (3), received blood transfusion (3), placental problems (2), trauma/fall (1), severe anemia (1), idiopathic thrombocytopenic purpura (1), or severe hypothyroidism (1)) No: 632 (85%) Unknown: 65 (9%)
Normal neonatal period	Yes: 549 (74%) Unknown: 54 (7%) No: 143 (19%) (see below) *History of Prematurity (< 37 weeks) or NICU stay: 108* *History of IV antibiotic exposure (no NICU stay): 5* *History of Jaundice (no NICU stay): 26* *History of low Apgar score (no NICU stay): 2* *History of seizures after birth (no NICU stay): 2*
Chemotherapy history	3 (< 1%)
Radiation exposure	1 (< 1%)
Chemotherapy and radiation exposure	4 (< 1%)
History of labyrinthitis	1 (< 1%)
Possible endolymphatic hydrops	2 (< 1%)
History of meningitis	7 (1%)
History of chronic otitis media with cholesteatoma	1 (< 1%)

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