Ocular Findings in Children With Congenital Sensorineural Hearing Loss

Derek D. Mafong, MD; Steven D. Pletcher, MD; Creig Hoyt, MD; Anil K. Lalwani, MD

Objective: To examine the yield of ophthalmologic examination in the diagnostic workup of unexplained sensorineural hearing loss (SNHL) in children.


Setting: Tertiary care university hospital.

Participants: Children 18 years or younger presenting with unilateral or bilateral SNHL.

Outcome Measures: Ophthalmologic findings.

Results: Of the 49 patients with SNHL for whom ophthalmologic examination results were available, 15 (31%) had ocular abnormalities. Hyperopia was the most common abnormality, present in 7 patients (46%). Myopia was found in 2 patients (13%) and astigmatism in 1 (2%). Two other patients had multiple abnormalities: one with hyperopia and astigmatism and the other with myopia and astigmatism. The remaining 4 patients had the following abnormalities: Lisch nodules, esotropia, ptosis, and allergic conjunctivitis. As a result of ophthalmologic examination, 5 interventions were performed in 4 children: 2 children received prescription lenses; 2 children underwent surgery; and 1 child was treated with eyedrops. Ophthalmologic examination in 2 children contributed to the diagnosis of a hearing loss syndrome.

Conclusion: In children with SNHL, ophthalmologic examination is useful in evaluating visual acuity and determining or confirming the cause of hearing impairment.


ESTIMATES OF the prevalence of permanent, moderate to severe bilateral sensorineural hearing loss (SNHL) fall between 1 and 2 per 1000 live births.1,2 Significant bilateral hearing loss during the first years of life can impede adequate development of literacy skills, school achievement, and social or emotional development.3,4 Studies of hearing-impaired populations indicate a genetic basis in approximately 50% of all hearing loss.5 Approximately 15% to 30% of hereditary hearing impairment involves other organ systems and occurs as a syndrome.6 Identifying syndromic causes of hearing loss requires a thorough evaluation that includes a complete case history, physical examination, audiologic testing, radiographic studies, consultation with other specialists, and multiple laboratory testing. While studies have shown the diagnostic yield of routine laboratory evaluation to be low, abnormal radiologic findings in children with SNHL range from 7% to 39%, indicating that computed tomography (CT) of the temporal bone is important in their evaluation.7-10

In hearing-impaired children, early identification of other sensory deficits and prompt intervention are essential for cognitive and emotional development. Because hearing-impaired children are extremely dependent on vision to compensate for the loss of auditory input, it is crucial to establish normal visual acuity.11 Previous studies have shown the incidence of visual problems to be higher among the deaf.11-13 In addition, abnormal ocular findings are associated with certain common deafness syndromes (Table 1). Since visual and auditory channels are responsible for more than 95% of information acquisition,14 it is crucial to maximize visual function in children with SNHL. In this report, we review our experience with the utility of routine ophthalmologic examination in the diagnostic evaluation of children with unexplained SNHL.

From the Division of Otolaryngology–Head and Neck Surgery (Drs Mafong, Pletcher, and Lalwani), and the Division of Pediatric Ophthalmology (Dr Hoyt), University of California, San Francisco.

©2002 American Medical Association. All rights reserved.

Downloaded From: https://jamanetwork.com/ on 06/20/2022
We conducted a retrospective analysis of children with unidentified causes of SNHL seen between 1998 and 2000 at the University of California, San Francisco Medical Center. The study was approved by the Committee on Human Research. Patients with SNHL attributed to recurrent otitis media, maternal cytomegalovirus, rubella, and toxoplasmosis were not included in this group. The following clinical information was retrieved from clinical notes, hospital charts, and outside records and recorded on a clinical data sheet: demographic information; pertinent prenatal, perinatal, and postnatal factors (eg, gestational diabetes, low birth weight); family history of hearing loss; CT and magnetic resonance imaging (MRI) findings; and ophthalmologic consultation. A cause for the hearing loss was noted if the clinical staff was able to establish a diagnosis.

The following clinical information was included in this group. The following clinical information was included in this group. We conducted a retrospective analysis of children with unilateral SNHL attributed to recurrent otitis media, maternal cytomegalovirus, rubella, and toxoplasmosis. The severity of hearing impairment was moderate to profound hearing loss in 81% of patients, while the remainder had mild hearing loss. Most had SNHL; however, mixed hearing loss was found in 6 patients. Members of our research group have previously reported the outcome of laboratory and radiologic evaluation in these same children.

A total of 114 patients 18 years or younger with SNHL of unknown cause were identified; 54 were girls and 60 were boys. The age range was 1 to 18 years, with a mean and median age of 9 years. Audiometric results were available in the medical records of 111 of 114 patients: 95 (83%) had bilateral SNHL and 13 (11%) had unilateral SNHL. The severity of hearing impairment was moderate to profound hearing loss in 81% of patients, while the remainder had mild hearing loss. Most had SNHL; however, mixed hearing loss was found in 6 patients. Members of our research group have previously reported the outcome of laboratory and radiologic evaluation in these same children.

Records of 49 ophthalmologic examinations were available for review; nearly all of the records available were for children undergoing ophthalmologic evaluation at our institution. Limited success was encountered in obtaining outside ophthalmologic records. A total of 15 patients (31%) had ocular abnormalities. Hyperopia was the most common abnormality and was found in 7 patients (46%). Myopia was found in 2 patients (13%). Astigmatism was present in 1 patient (7%). Two other patients had multiple abnormalities: one with hyperopia and astigmatism and the other with myopia and astigmatism. The remaining 4 patients had the following abnormalities: Lisch nodules, esotropia, ptosis, and allergic conjunctivitis.

In 2 children, ophthalmologic findings aided in the diagnoses of a hereditary hearing loss syndrome. A child with moderate bilateral SNHL and bilateral large vestibular aqueducts (LVAs) on CT was found to have Lisch nodules on ophthalmologic examination and diagnosed with neurofibromatosis type 1. The second child with multiple congenital anomalies (including renal agenesis and bilateral inguinal hernias) was found to have optic nerve hypoplasia and esotropia on ophthalmologic examination, and infant of diabetic mother syndrome was diagnosed.

Nearly all of the patients had undergone radiologic imaging; more importantly, their radiologic studies were available for review (Table 2). Forty-six of these patients had CT and 2 had MRI results available; 1 patient did not have radiologic studies. Of the 15 patients with ocular abnormalities, 5 (33%) had abnormal radiologic findings; bilateral LVA was the most common finding (n=2). Of the 7 patients with hyperopia, 6 had CT scans; normal ear anatomy was most common (n=4) followed by hypoplastic cochlea (n=1) and lateral semicircular canal dysplasia (n=1). One of the 2 patients with myopia had bilateral LVA. The 2 patients with multiple findings had normal CT findings. Of the patients with Lisch nodules, esotropia, ptosis, and allergic conjunctivitis, the patient with Lisch nodules showed bilateral LVA, while the remaining radiologic studies were normal. The MRI in the patient with esotropia showed butterfly vertebrae.

As a result of findings on ophthalmologic examination, 4 patients underwent medical or surgical intervention. Prescription lenses were given to 2 myopic children, one of whom also underwent an operative ptosis repair. A bilateral medial rectus recession surgery was performed in a child with esotropia. Another child was given anti-inflammatory drops for an allergic conjunctivitis.

Ophthalmologic consultation in children with SNHL serves 2 goals. The first goal is to determine visual acuity and identify visual deficits requiring intervention. Normalization of visual acuity is critical in these patients who already have auditory sensory deficit and therefore have greater dependence on visual input. Early identification of children with dual sensory deficits allows early intervention and maximization of quality sensory input critical for development. The second goal is to aid in the identification of hereditary hearing loss syndromes that are associated with ocular findings. Early identification of syndromes gives patients and their families the comfort of a diagnosis, may allow identification of other syndrome-associated abnormalities, and will have implications for

| Table 1. Ocular Findings in Hereditary Deafness Syndromes |
|-----------------|-----------------|
| Syndrome        | Ocular Findings |
| Autosomal dominant | Lisch nodules of iris and optic gliomas |
| Neurofibromatosis type 1 | Juvenile posterior cataracts |
| Neurofibromatosis type 2 | Blue sclera |
| Osteogenesis imperfecta | Myopia, retinal detachment, cataracts |
| Stickler syndrome | Coloboma of lower eyelids, downward slanting palpebral fissures |
| Treacher Collins syndrome | Heterochromia irides, lateral displacement of medial canthi |
| Waardenburg syndrome | Progressive external ophthalmoplegia and retinopathy |
| Autosomal recessive | Retinitis pigmentosa |
| Usher syndrome | Optic atrophy |
| Wolfman syndrome | Congenital or progressive blindness, pseudoglioma |
| Sex-linked disorders | Opacification, ocular degeneration resulting in microphthalmia |
| Norrie syndrome | Progressive external ophthalmoplegia and retinopathy |
| Mitochondrial disorders | Progressive external ophthalmoplegia and retinopathy |
| Kearns-Sayre syndrome | Intestinal keratitis |

©2002 American Medical Association. All rights reserved.
In 1987, Fillman et al. examined 210 hearing-impaired patient population and definition of what is abnormal by Fillman et al. This discrepancy may be explained by the nature but significantly lower than that noted in the study. The incidence of ophthalmologic abnormalities noted in the present study was in the middle of the range reported by the literature.

Previous studies investigating ophthalmologic abnormalities in children with SNHL have reported prevalence ranging from 12% to 58%. The variability in these studies is likely secondary to differences in patient population and definition of what is abnormal. In 1987, Fillman et al. examined 210 hearing-impaired students and found that 44% had abnormal ophthalmologic findings. Of the 92 patients with visual findings, 12 interventions were performed (corrective lenses and patching), and 3 patients were found on electroretinogram (ERG) to have retinitis pigmentosa. The incidence of ophthalmologic abnormalities noted in the present study is in the middle of the range reported by the literature but significantly lower than that noted in the study by Fillman et al. This discrepancy may be explained by a difference in patient population. In the present study, patients with known causes of SNHL such as rubella were excluded. In the study by Fillman et al., multiple patients were found to have both ocular and otologic manifestations of rubella. Similar to 6% in that study, 8% of patients who underwent ophthalmologic examination in the present study were treated with some intervention.

Other investigators have promoted measurement of corneal-retinal potentials via ERG testing as part of the routine ophthalmologic evaluation of children with SNHL to identify Usher syndrome. The ERG is a noninvasive test that has the potential to identify retinitis pigmentosa before the onset of fundoscopic and visual abnormalities. Mets et al. found that 5 (10%) of 48 children with severe to profound preverbal SNHL screened with ERG were found to have Usher syndrome. The investigators therefore recommended that all children with severe to profound prelingual SNHL should be screened for Usher syndrome by ophthalmologic examination, including ERG. Absence of Usher syndrome in the present study is likely because ERG testing was not performed as part of ophthalmologic evaluation. As ERG testing often requires sedation in children, we have not routinely performed it unless the syndrome was clinically suspected. Because early retinal changes are uncommon, it may be prudent to obtain a follow-up ophthalmologic examination during teen years in these children in whom an ERG is not performed to detect retinal pigmentary changes associated with Usher syndrome.

A weakness of the present study is that the results of ophthalmologic examinations were unavailable for many of our patients. Many of our patients live far from our medical center and were examined by their local ophthalmologists. The results of these examinations were often unavailable. It is unclear how many of our patients, all of whom were referred for ophthalmologic examination, actually were examined by an ophthalmologist. This introduces the possibility of selection bias. Despite this limitation, we believe that our data support routine ophthalmologic examination for children with congenital SNHL.

In addition to ophthalmologic examination, the clinical evaluation of children with congenital SNHL at our institution also includes laboratory testing and imaging studies. Abnormal findings in traditional laboratory and ancillary testing for children with congenital SNHL (complete blood cell count with platelets, thyroid function tests, microscopic and macroscopic urinalysis, and electrocardiogram) have proven to be quite uncommon. On the other hand, there is a high incidence of abnormal findings on CT scan in patients with congenital SNHL. In the earlier review of this cohort, radiologic abnormalities were present in 38 (39%) of 97 cases, with 33 CT scans (37%) and 7 MRIs (33%) identified as abnormal.

The most common radiologic abnormality was LVA. Likewise, evaluation of patients with ocular findings turned up a similar incidence of radiologic abnormality. In other words, in individuals with ocular findings, radiologic abnormality is not more or less likely than in patients with normal findings of eye examination.

Newer, genetically based diagnostic tests, such as CX26 mutation testing, are becoming available that may change our evaluation of children with congenital SNHL. Connexin 26 (CX26) is a gap junction protein believed to have a role in 50% of patients with congenital hearing loss. CX26 is ideal for genetic testing: it not only accounts for a large fraction of childhood SNHL, but it is small, and there are 2 common prevalent mutations (35delG and 167delT). Ocular findings have not been reported in patients with CX26 mutations. Therefore, it may be that as genetic testing becomes routine, normal CX26 results may obviate the need for laboratory, radiologic, and ophthalmologic evaluation. However, until clear genotype-phenotype correlation data are fully available, children with congenital SNHL should undergo ophthalmologic examination to allow early detection and treatment.

Table 2. Ocular Findings and Radiologic Results in Children With Sensorineural Hearing Loss

<table>
<thead>
<tr>
<th>Ocular Finding</th>
<th>CT Finding</th>
<th>MRI Finding</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hyperopia</td>
<td>Normal</td>
<td>None</td>
</tr>
<tr>
<td>Myopia</td>
<td>Normal</td>
<td>None</td>
</tr>
<tr>
<td>Astigmatism</td>
<td>Normal</td>
<td>None</td>
</tr>
<tr>
<td>Myopia +</td>
<td>Normal</td>
<td>None</td>
</tr>
<tr>
<td>Ocular Findings</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Esotropia</td>
<td>None</td>
<td>None</td>
</tr>
<tr>
<td>Ptosis</td>
<td>None</td>
<td>None</td>
</tr>
<tr>
<td>Allergic</td>
<td>None</td>
<td>None</td>
</tr>
<tr>
<td>conjunctivitis</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

*CT indicates computed tomography; MRI, magnetic resonance imaging; SCC, semicircular canal; and LVA, large vestibular aqueduct.
ment of ocular abnormalities and identification of hereditary hearing loss syndromes.

Accepted for publication May 20, 2002.

This study was presented at the American Academy of Pediatrics National Conference and Exhibition, San Francisco, Calif, October 21, 2001.

Corresponding author and reprints: Anil K. Lalwani, MD, Division of Otology, Neurotology, Skull Base Surgery, Department of Otolaryngology–Head and Neck Surgery, University of California, San Francisco, 400 Parnassus Ave, A730, San Francisco, CA 94143-0342 (e-mail: lalwani@itsa.ucsf.edu).

REFERENCES


CME Announcement

CME Hiatus: July Through December 2002

CME from JAMA/Archives will be suspended between July and December 2002. Beginning in early 2003, we will offer a new online CME program that will provide many enhancements:

- Article-specific questions
- Hypertext links from questions to the relevant content
- Online CME questionnaire
- Printable CME certificates and ability to access total CME credits

We apologize for the interruption in CME and hope that you will enjoy the improved online features that will be available in early 2003.