In Reference to Temporal Bone Imaging in GJB2 Deafness

Dear Editor:

We read with interest the paper by Propst et al. concerning temporal bone imaging by computed tomography (CT) in patients with GJB2-related deafness. The GJB2 gene, encoding the connexin 26 protein, which is involved in gap junction formation, has been found to be the most common cause of autosomal recessive nonsyndromic sensorineural hearing loss (SNHL) in numerous world populations. Several studies have shown that GJB2-related SNHL is not associated with temporal bone anomalies. Cohn et al. found no inner ear abnormalities on CT scanning in a subset of individuals from 24 families who were either homozygous or compound heterozygous for GJB2 mutations. Denoyelle et al. scanned 23 deaf children from 17 families with GJB2 mutations and found no inner ear malformations. And, most recently, Yaeger et al. reported that in 40 individuals with enlarged vestibular aqueduct (EVA), none had GJB2 mutations.

In contrast, there are a few studies that have identified temporal bone anomalies in a small number of deaf persons carrying GJB2 mutations. Kenna et al. described one patient with bilateral EVA and Mondini deformity who had biallelic GJB2 mutations, and Preciado et al. identified another patient with radiologic abnormalities in a cohort of 29 patients with two GJB2 mutations. We conducted a similar study in our laboratories (Molecular Otolaryngology Research Laboratories) as a retrospective analysis of 1,315 patients with SNHL in whom CT of the temporal bones was available for review and found that inner ear abnormalities were present in 30% (397) of patients. Three hundred nine patients had either unilateral or bilateral EVA, and 88 patients had Mondini dysplasia. Genetic testing identified two GJB2 mutations in 155 of 1,315 patients, 14 (9%) of whom had temporal bone abnormalities (4 unilateral EVA, 8 bilateral EVA, 2 bilateral Mondini dysplasia). Our results and those of other investigators suggest that the probability of an association between GJB2-related SNHL and temporal bone anomalies is below 10%, much less than the 72% figure reported by Propst et al.

On the basis of their results, Propst et al. recommend temporal bone imaging as a routine part of the evaluation of patients with GJB2-related deafness. We disagree. We believe routine CT scanning in persons with biallelic GJB2 mutations is neither evidence based nor cost justified if the clinical history and audio profile are consistent with data reported on large cohorts of patients with GJB2-related deafness. Certainly, temporal bone imaging is needed as part of the preoperative evaluation for cochlear implantation. Genetic testing also provides the opportunity to reduce the stress associated with exhaustive testing, to offer appropriate genetic counseling, and to share with the child's family meaningful prognostic information.

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BIBLIOGRAPHY


In Reply:

We thank Drs. Azaiez and Smith for their interest in our paper on temporal bone imaging in GJB2 deafness. Although we appreciate the complexities inherent in formulating clinical care algorithms, our recommendation to include computed tomography (CT) imaging in the assessment of individuals with GJB2-related hearing loss (HL) was based on our finding that temporal bone abnormalities are