Pediatric Otolaryngologists’ Knowledge and Understanding of Genetic Testing for Deafness

Nathaniel H. Robin, MD; Christin Dietz; James E. Arnold, MD; Richard J. H. Smith, MD

Objective: To assess the level of a cohort of pediatric otolaryngologists’ knowledge and understanding of genetics and genetic testing for deafness and hard of hearing (D/HOH).

Methods: A questionnaire was designed to assess the level of knowledge and understanding of the genetic basis and genetic testing for deafness among a cohort of pediatric otolaryngologists. Three hundred questionnaires were made available to attendees of the 14th (1999) Annual Meeting of the American Society of Pediatric Otolaryngology, Palm Desert, Calif. A series of questions asked to gauge the respondent’s level of knowledge of genetics and hearing impairment addressed estimating recurrence risks for deaf and normal-hearing parents and the likelihood of detecting a mutation in connexin 26 in specific clinical scenarios.

Results: A total of 28 questionnaires were completed and returned. All respondents reported that they regularly saw patients for D/HOH. Almost half commonly refer these patients for genetic testing and counseling. Seventeen (71%) of 24 otolaryngologists stated they offered genetic testing in all situations, while 6 offered counseling only at parental request or to address recurrence risk issues. One otolaryngologist offered genetic testing if there was a deaf sibling. Twelve (67%) of 18 offered pretest counseling, which was most frequently provided by a genetic counselor. Although 3 (19%) of 16 otolaryngologists provided the counseling themselves, 2 (13%) reported that they and a genetic counselor provided the counseling. While 24 (89%) of the 27 correctly stated that nonsyndromic D/HOH is usually autosomal recessive, recurrence risks were incorrectly estimated in several examples.

Conclusions: While the surveyed pediatric otolaryngologists have a good knowledge of genetics and genetic testing for D/HOH, recurrence risks were often inaccurate. Since D/HOH testing is clinically available, it is imperative that physicians are educated about genetics and genetic testing and are able to communicate this to their patients and their patients’ families.


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WE ARE entering a new era in the diagnosis and management of deafness and hard of hearing (D/HOH), as genetic testing is becoming an integral part of the evaluation of these affected patients. In previous work, we have shown that normal-hearing parents of deaf children have a positive attitude toward genetic testing for deafness and recognize the benefits of such testing. However, an unexpected finding was the parents’ lack of understanding of the genetics of their child’s deafness even after genetic testing had been completed. Parents who had had genetic testing for their deaf child were just as likely to have a poor understanding of the recurrence risk of their having another deaf child as those who had not had such testing. Almost all respondents (28/29, 97%) had a poor understanding of their chance to have another deaf child, usually underestimating their recurrence risk at 0%. Parents also failed to understand their child’s recurrence risk. These findings suggested that the testing group had not received genetic counseling or that the counseling was inadequate. Because many families of deaf children interact with pediatric otolaryngologists, we conducted a pilot study of these physicians to assess their level of knowledge and understanding of genetics and genetic testing.

See also pages 927 and 941
PARTICIPANTS AND METHODS

Participants for this study were recruited at the 14th (1999) Annual Meeting of the American Society of Pediatric Otolaryngology, Palm Desert, Calif. A packet that contained a brief explanatory letter, a questionnaire, and a self-addressed stamped envelope was made available to all attendees of this meeting. There were no inclusionary or exclusionary criteria. The study received approval by the institutional review board of University Hospitals of Cleveland, Cleveland, Ohio, but since the questionnaires were anonymous, signed informed consent was not needed. The 3 areas of focus were demographics, practice policies regarding pediatric hearing impairment, and general knowledge of the genetics of hearing impairment.

RESULTS

More than 300 questionnaires were made available at the meeting, but only 28 were completed and returned. Because the questionnaire packet was made available and not specifically mailed to individuals, it is impossible to give a response rate; however, given the small sample size, the significance of these results is not definitive, but rather represents pilot data. The results of the questionnaire are given in the Table.

Most respondents completed their otolaryngology training in the 1980s or 1990s (22/28, 80%), with 96% (27 respondents) training at a university-based hospital. Twenty (71%) of 28 had some fellowship training, with 12 (60%) of the 20 having had a 1-year pediatric fellowship, and 6 (30%) of the 20 having had a 2-year fellowship. More than half (21/28, 75%) responded that they are in an academic practice, and an additional 3 (11%) of the 28 were affiliated with an academic medical center; 4 respondents (14%) were in private practice.

All reported that they regularly saw patients for D/HOH. Almost half the respondents (13/27, 48%) stated that they would refer these patients for genetic testing and counseling. When asked under what conditions they offer genetic testing, 17 (71%) of 24 otolaryngologists stated in all situations, while 6 (25%) offered counseling only at parental request or to address recurrence risk issues. Twelve (67%) of 18 offered pretest counseling. Counseling was most frequently provided by a genetic counselor, although some otolaryngologists provided the counseling themselves (3/16, 19%) or together with a genetic counselor (2/16, 13%).

The series of questions intended to gauge the respondent’s level of knowledge of genetics and hearing impairment addressed estimating recurrence risks for deaf and normal-hearing parents, and the likelihood of detecting a mutation in connexin 26 in specific clinical scenarios. Although almost all respondents (25/28, 89%) stated that most nonsyndromic D/HOH is autosomal recessive, recurrence risks were incorrectly estimated in several cases. For example, when asked what the chance is for 2 deaf parents to have a deaf child, the most common answer (10/27, 37%) was 10%, which is correct, but 18 (67%) responded incorrectly, with 15 (56%) of the 27 giving recurrence risks from 25% to 93.8%.

<table>
<thead>
<tr>
<th>Survey Question</th>
<th>No. of Respondents/ Total No. of Respondents (% of Respondents)</th>
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<tbody>
<tr>
<td>Fellowship training (duration and type)</td>
<td>Yes 20/28 (71) 1-y Pediatric 12/20 (60) 2-y Pediatric 6/20 (30) 4-y Pediatric 1/20 (5) 1-y Otology 1/20 (5) None 8/28 (29)</td>
</tr>
<tr>
<td>Approximately how many patients do you see monthly for SNHI (sensorineural hearing impairment)?</td>
<td>1 4/27 (15) 2-5 11/27 (41) 6-10 5/27 (19) &gt;10 7/27 (26)</td>
</tr>
<tr>
<td>For a patient newly diagnosed with isolated SNHI which of the following do you routinely order (circle all that apply)?</td>
<td>Repeated audiometry 26/27 (96) Audiology on parents 7/27 (26) Thyroid function testing 10/27 (37) Ophthalmology consultation 14/27 (52) Congenital infection screening 6/27 (22) Computed tomographic scan of the temporal bones 17/27 (63) Electrocardiogram 6/27 (22) Genetics referral for counseling/evaluation 13/27 (48) DNA-based genetic testing 5/27 (19)</td>
</tr>
<tr>
<td>Do you routinely provide the parents of a newly diagnosed deaf child with information on support groups?</td>
<td>Yes 16/27 (54) No 11/27 (41)</td>
</tr>
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<td>If you offer DNA-based genetic testing, do you provide pretest genetic counseling?</td>
<td>Yes 12/18 (67) No 6/18 (33)</td>
</tr>
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<td>If yes, who provides genetic counseling?</td>
<td>They do 3/16 (19) Refer to a geneticist/genetic counselor 11/16 (69) Both they and a geneticist 2/16 (13)</td>
</tr>
</tbody>
</table>
100%. The most common recurrence risk estimate given for normal-hearing parents of a deaf child was 25% (15/28), with only 43% (12/28) giving the correct estimate (10%-18%).

Most knew that approximately one third of the deaf children born to normal-hearing parents test positive for a Cx26 gene mutation, but most did not recognize that two thirds of normal-hearing siblings of such children are carriers of a Cx26 mutation. All (28) stated that they would be willing to test the normal-hearing sibs of the Cx26-positive deaf child at the parents’ request.

The results of this pilot study suggest that the surveyed pediatric otolaryngologists have a good knowledge of genetics and genetic testing for D/HOH. For example, most knew that the majority of isolated D/HOH was due to genetic factors and that most pediatric cases were autosomal recessive. Most (4 of 28 respondents) correctly stated that a deaf child born to normal-hearing parents would have an approximately 33% chance of having a Cx26 gene mutation detected. However, one disconcerting issue was the inaccurate perceptions of recurrence risks in specific situations. Most respondents overestimated recurrence risk in the 2 given situations (2 deaf parents or 2 normal-hearing parents with 1 deaf child, Table). Because, as this study suggests, many pediatric otolaryngologists are using genetic testing for D/HOH, it is important ensure that parents and patients receive correct and accurate information relating to these issues. As many studies have shown, pretest genetic counseling is necessary if the parents and patients are to receive the maximum benefits from genetic testing.7 With pretest and posttest counseling, parents and patients have a greater understanding of their test results—both the meaning and limitations of the findings. For example, Brunger et al have reported that parents of deaf children who test negative for Cx26 deafness often mistakenly believe that their deaf child does “not have the deafness gene.”3(p1623) These parents were either never told or did not understand that this test screens only one of dozens of genes that can cause deafness. Thus, instead of a very low recurrence risk, normal-hearing parents of a deaf child who tests negative for Cx26 mutations still have a recurrence risk of approximately 14% (R.H.J., unpublished data, 1999).

Another interesting point is that all respondents stated that they would agree to test the normal sibs of a child with Cx26-related deafness. While there is a 67% chance that each sib would be a carrier, current recommendations are that carrier testing of children for genetic disease should not be done. It is reasoned that carrier testing provides no medical benefit to a child and is only important for reproductive decision making. Such decisions will be made years later, when the child is an adult. As an adult, the child (as an adult making reproductive decisions) has the right not to know their carrier status if they so choose. Therefore, it is believed that decisions concerning carrier testing for genetic disease should, in general, be left to the child as an adult, not their parents.3

**CONCLUSIONS**

We are entering an age in which genetic testing is becoming more common and widely used, and studies such as this one emphasize the need for continuing education and the value of genetic counseling in an effort to
maximize benefits to patients. However, we are not sug-
gest that every patient undergoing genetic testing for
D/HOH needs to be seen by a geneticist or genetic coun-
selor. Rather, it is anticipated that this pretest and post-
test genetic counseling will be done by the primary phy-
sician, the pediatrician, or the otolaryngologist, with
genetics becoming involved in only difficult or atypical
cases. But it is imperative, therefore, that the counseling
be provided in the recommended manner, by knowl-
geable providers and with adequate time allotted for
questions and gaining feedback.6

Accepted for publication February 7, 2001.

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REFERENCES

loss attributable to mutations in the connexin 26 gene. Pediatrics. 1999;103:
546-550.
2. Van Camp G, Smith R. Hereditary hearing loss home page. Available at: http:
3. Brunger JW, Murray GS, O’Riordan MA, Matthews AL, Smith RJH, Robin NH.
In press.
4. Brunger JW, Matthews AL, Smith RJH, Robin NH. Genetic testing and genetic
5. Beauchamp TL, Childress JS. Genetic testing for children and adolescents: who
decides? In: Principles of Biomedical Ethics. 4th ed. New York, NY: Oxford Uni-
study of families attending a genetic counseling service. J Genet Couns. 1995;
4:281-300.