Am. J. Hum. Genet. 67:1621–1625, 2000

Report

Parental Attitudes toward Genetic Testing for Pediatric Deafness

Jeanne Weir Brunger,[1] Gail S. Murray,[2] Maryann O'Riordan,[3] Anne L. Matthews,[1] Richard J. H. Smith,[5,6] and Nathaniel H. Robin[1,2,3,4]

Departments of [1]Genetics, [2]Otolaryngology and Head and Neck Surgery, [3]Pediatrics, and [4]Reproductive Biology, Case Western Reserve University School of Medicine, University Hospitals of Cleveland, Cleveland; and [5]Department of Otolaryngology and Head and Neck Surgery, and [6]Interdepartmental Genetics PhD Program, University of Iowa, Iowa City

Recent molecular genetic advances have resulted in genetic testing becoming an option for deaf individuals and their families. However, there is little information about the interest in such testing. To investigate this issue, parents with normal hearing who have one or more deaf children were surveyed about their attitudes toward diagnostic, carrier, and prenatal genetic testing for deafness. This population was chosen because it represents the majority of individuals who are encountered in clinical practice, given that 90%–95% of deaf individuals are born to persons with normal hearing. Of 328 surveys distributed, 96 were completed and returned. Of the respondents, 96% recorded a positive attitude toward genetic testing for deafness, including prenatal testing, although none would use this information to terminate an affected pregnancy. All respondents had a poor understanding of genetics, with 98% both incorrectly estimating the recurrence risk of deafness and misunderstanding the concept of inheritance. Notably, these findings were similar in the group who had had genetic testing for their children and in the group who had not, suggesting either that the parents who received genetic testing did not receive genetic counseling or that the counseling was not effective. On the basis of these results, it was concluded that this population is interested in the use of genetic testing and that testing should not be done without first providing formal genetic counseling. Appropriate counseling can help parents to understand the risks, benefits, and limitations of genetic testing.

Recent advances in understanding the molecular genetic basis of deafness have made genetic testing an option for hard-of-hearing and deaf individuals and their families. (The term "deaf" refers to a severe-to-profound hearing inability, and "hard-of-hearing" refers to more mild hearing inability.) Testing for mutations in *GJB2*, the gene for connexin 26 (Cx26), is commercially available, and many more deafness-related genetic tests undoubtedly will follow in the near future. A Cx26 mutation can be detected in ~30% of sporadic cases of prelingual hearing impairment. The likelihood of detection of a Cx26 mutation increases to >50% in families with identified autosomal recessive transmission (Maw 1995; Zelante 1997). However, little attention has been given to the value and impact of this test, as perceived by the public. In the only study that has so far attempted to investigate this issue, Middleton et al. (1998) surveyed a group of Deaf adults to elicit their attitudes toward genetic testing for deafness. (Please note that "Deaf" refers to individuals who identify with the Deaf culture, whereas "deaf" refers to individuals with severe-to-profound deafness.) Not surprisingly, Deaf adults had a predominantly negative attitude toward genetic testing for deafness, with the majority stating that they believed that such tests would do more harm than good.

Although this information is helpful in understanding the perspectives of the Deaf community, it cannot be generalized to individuals who do not consider themselves "culturally" Deaf. The majority (90%–95%) of deaf children are born to parents with normal hearing (National Information Center for Children and Youth with Disabilities 1998; Deaf World Web 2000). Inevitably, these parents search for a reason for this unexpected event and often arrive at an inaccurate conclusion (Israel 1995). In some cases, genetic testing could pro-

Received August 11, 2000; accepted for publication October 10, 2000; electronically published November 2, 2000.

Address for correspondence and reprints: Dr. Nathaniel H. Robin, Lakeside 1500, Cleveland, OH 44106-6506. E-mail: nhr2@po.cwru.edu

0002-9297/2000/6706-0032$02.00

vide an etiology for the child's deafness, thereby addressing these questions. In addition, genetic testing may provide information to assist parents of deaf children in making informed decisions concerning medical management and appropriate educational interventions for their children (Cohn 1999).

What remains unknown is whether these parents themselves feel that there are benefits in knowing their child's genetic status. Numerous studies have shown a predominantly positive attitude, among the public, toward genetic testing in general (Rowley et al. 1984; Williamson et al. 1989; Decruyenaere et al. 1992; Mennie et al. 1993; Hietala et al. 1995). It follows then that individuals who do not identify with the Deaf community may have attitudes toward genetic testing for deafness that are different from those of individuals who consider themselves part of the Deaf community.

In an effort to determine the attitudes and levels of interest among hearing parents of deaf children toward diagnostic, carrier, and prenatal testing for deafness, we surveyed a large cohort of parents with normal hearing who had a deaf child. A total of 328 individuals were asked to participate in the study, and 106 (32%) responded. Ten respondents were excluded, because they reported in the questionnaire that one parent was deaf. Of the remaining 96 respondents, 32 (33%) had already had genetic testing for their deaf child. Of those whose children had had genetic testing, 30 respondents were ascertained through the University of Iowa Hospitals and Clinics (UIHC), and 2 through the University Hospitals of Cleveland (UHC). The 64 respondents whose children had not had genetic testing were ascertained through a school for the hearing impaired (33 respondents of 163 surveys sent), a regional center for persons with hearing impairment (8 of 10 surveys), UIHC (42 of 100 surveys), and UHC (23 of 54 surveys).

Questionnaire topics included demographic information, familiarity with genetic testing, and level of interest in results of such testing. Parents whose children had already had genetic testing were asked why they decided to have testing and, from their perspective, what impact the test results had had. For those who had not had testing, questions were asked regarding familiarity with genetic testing and whether testing had been offered. In the cases in which the parents had declined testing, reasons for that decision were explored. A summary of selected results is presented in table 1.

We found that the vast majority (96%) of respondents had an overall positive attitude toward genetic testing for deafness, including 94% (60/64) of those who had not had genetic testing. This response was significantly different ($P < .001$) from the response reported by Middleton et al. (1998), who found that members of the Deaf community felt that genetic testing for deafness was not beneficial to deaf individuals. The majority of our respondents felt that genetic testing would provide benefits to their child. Of 96 respondents, 96% (92) expressed approval for genetic testing for deafness, and 76% (70/92) stated that they were interested in having testing themselves. The most common reason given for wanting testing (93% [65/70]) was to identify a cause of deafness. Other common reasons included determining the recurrence risk (RR) and refining the affected child's future medical management and/or treatment. Slightly more than half (54% [38/70]) of parents who were interested in genetic testing for their deaf child were interested in having genetic testing themselves, and 92% (35/38) stated that they wanted to find out what their chances were for having another deaf child. Of the parents who were interested in genetic testing for their deaf child but not for themselves, 53% (17/32) specifically stated that there was no purpose in having such testing, and 8 of the 17 made the statement "I am not deaf" to explain their sense that such testing served no purpose.

Of parents who were interested in genetic testing for their deaf child, 44% (31/70) were interested in having genetic testing for their other children—with 68% (21/31) wishing to find out whether their other children were carriers, in order to determine those children's chances of having a deaf child. Three parents stated that they would like to have testing for their children but felt that by waiting until their children were older they could allow each child to then make an informed decision.

Twenty of the 92 individuals who expressed general approval of genetic testing stated that, although they approved of genetic testing for others, they were not interested in testing for themselves or their children. Half (10/20) of these individuals did not feel that their child's deafness was genetic. Three participants responded that they did not approve of such testing because they did not believe that genetic testing should be offered for any reason. One stated, "[I] am opposed to eugenics." None of these three persons considered themselves or their children to be members of the Deaf community.

Of the individuals who were interested in genetic testing, 87% (61/70) believed that prenatal genetic testing for deafness should be offered. Most (72% [44/61]) believed that this option should be available to parents who want to prepare for a deaf child (e.g., by taking sign-language classes and finding pediatric deafness specialists). Of those not interested (nine), six felt that there would be no purpose in having such testing, and three felt that it would "allow for abortion to be an option." Some parents stated that genetic test results might alter their decision to have additional children, but no parents stated that they would terminate an affected pregnancy. This issue involves a major concern of the Deaf community—that is, that genetic testing will influence family

Table 1

Summary of Selected Results

Variable	Proportion (%)
Surveys returned/distributed:	
UIHC	42/100 (42%)
UHC	23/54 (43%)
Regional school for hearing impaired	33/163 (20%)
Regional center for hearing impaired	8/10 (80%)
Overall	106/328 (32%)
Surveys excluded[a]	10/106 (9%)
Had genetic testing for child:	
UIHC	30/32 (94%)
UHC	2/32 (6%)
Overall	32/96 (33%)
Had not had genetic testing for child because:	
Not offered	47/64 (73%)
Offered but did not follow through	14/64 (22%)
Saw no purpose	12/14 (86%)
Overall	64/96 (67%)
Positive toward genetic testing for deafness:	
Had genetic testing	32/32 (100%)
Had not had genetic testing	60/64 (94%)
Overall	92/96 (96%)
Interested in genetic testing of deaf child	72/96 (75%)
Perceived benefits of testing deaf child:[b]	
Identify cause	65/70 (93%)
Learn accurate RR	65/70 (93%)
Alter medical management	41/70 (59%)
Interested in testing hearing sib:	
Identify sib's risk for having deaf child	21/31 (68%)
Will let sib make decision as adult	3/31 (10%)
Overall	31/70 (44%)
Not interested in testing hearing sib:[c]	
"My child's deafness is not genetic"	10/20 (50%)
Do not approve of any genetic testing	3/20 (15%)
Overall	20/70 (29%)
Interested in testing for self (parent):	
To identify RR	35/38 (92%)
Overall	38/70 (54%)
Not interested in testing for self (parent):	
"Because I am not deaf"	17/32 (53%)
Overall	32/70 (46%)
Interested in prenatal testing:[c]	61/70 (87%)
Could prepare for having deaf child	44/61 (72%)
Not interested in prenatal testing:	
Saw no purpose	6/9 (67%)
Did not want abortion to be an option	3/9 (33%)
Overall	9/70 (13%)
Inaccurate estimate of own (parent's) RR:	
RR estimated as 0	52/90 (58%)
Overall	83/90 (92%)
Inaccurate estimate of child's RR:	
RR estimated as 0	43/90 (48%)
Overall	88/90 (98%)
Child had negative Cx26 test result:	
Misunderstood test results	6/19 (32%)
RR for self estimated as 0	19/19 (100%)
RR for deaf child estimated as 0	3/19 (16%)

[a] For explanation, see text.

[b] Of those interested in testing. Two respondents did not answer questions about benefits of testing.

[c] Of those not interested in testing.

planning and lead to a decrease in the number of congenitally deaf children (Middleton et al. 1998).

In spite of this study population's strong interest in genetic testing for deafness, we found that they had a very poor understanding of genetics and the inheritance of deafness. The majority (83/90) inaccurately estimated their risk of having another deaf child, and most (58%) underestimated their RR as zero. Furthermore, 98% (88/90) had a poor understanding of their deaf child's chance of having future children with deafness, with 48% (43/90) stating that it was zero. Notably, there was no difference among those whose child had undergone genetic testing and those whose child had not.

Results of genetic testing were also frequently misinterpreted. Some parents (32% [6/19]) of children who received negative Cx26 test results specifically stated, "My child does not have the gene that causes deafness." All thought that they had a zero chance of having another deaf child, and three thought that their deaf child had a zero chance of having children with deafness. Studies have estimated that the actual RR for normal hearing parents of a child with nonsyndromic deafness is 10%–18% (Koehn et al. 1990), and the risk that an individual with nonsyndromic deafness will have a deaf child is ~5% (Smith 1991). As further illustration of this poor understanding of genetics, some parents believed that their child had inherited deafness from them but felt that they, as well as their child, had a zero chance of having other children with deafness. Other parents stated that they had a 25%–50% chance of having another deaf child but that they did not believe that their child's deafness was inherited from either parent.

Most (67% [64/96]) respondents' children had not had genetic testing for deafness. Most (73% [47/64]) of the group with untested children stated that they never had been offered such testing. Fourteen stated that they had been offered genetic testing but had not followed through, with 86% (12/14) stating that they felt there was no purpose. When asked how they anticipated that genetic test results might change their actions, 39% (25/64) reported that it might change future medical management and/or treatment of their deaf child. Sixteen (25%) anticipated that the results might change their decision and/or their child's decision to have additional children.

Of the 32 respondents who had Cx26 genetic testing for their child, the majority (91%) stated that they had decided to have such testing because they wanted to identify a cause. Surprisingly, 63% anticipated that the results might change their decision to have future children and/or their child's decision to have children. One parent stated, "My child might want to use the results to decide to adopt a child rather than risk having a child who is deaf," supporting the Deaf community's fear re-

garding the impact of genetic testing for deafness on family planning.

The comments made by one of the three participants who did not approve of genetic testing for any reason illustrate how public attitudes may lead people to be fearful of genetic testing and to be apprehensive about how test results might be used by the medical community. This apprehension is consistent with other studies that have focused on public attitudes toward genetic testing, which suggest that peoples' misconceptions and misinformation can cause some individuals to be so fearful of modern medical technology that they would decline any opportunity for genetic testing (Chapple et al. 1995; Hietala 1995).

These inaccurate beliefs provide clear evidence of the importance of formal pre- and posttest genetic counseling. By providing appropriate and accurate information, counselors can help individuals to make an informed decision as to whether they wish to undergo such testing. Furthermore, counseling allows those who receive genetic test results to have a clearer understanding of their meaning, including how the results may or may not modify estimates of a family member's RR.

Unfortunately, we do not know whether the participants in this study received genetic counseling. If they did, it was not helpful, because these individuals did not have a clear understanding of the limitations, risks, or benefits of genetic testing. Previous studies have shown that persons receiving genetic counseling should be encouraged to give feedback so that their understanding of genetic test results is apparent (Chapple et al. 1995). These activities may have been neglected for this study population.

Acquiring feedback after genetic testing is also crucial in understanding how results affect patients and families (Chapple et al. 1995). For example, we found that parents of children who tested positive for Cx26 deafness-causing mutations felt significantly more fearful after receiving these results, compared with parents of children without Cx26 deafness-causing mutations ($P < .004$; Fisher's exact test). A geneticist or genetic counselor should explore these feelings during a counseling session. Otherwise, these parents may be left with "serious feelings of guilt, 'spoiled identity,' or confusion" (Kenen and Schmidt 1987).

The present study demonstrates that parents with normal hearing who have deaf children view genetic testing for deafness as positive and beneficial. However, these parents typically have a poor understanding of genetic principles, suggesting either that they did not receive genetic counseling or that the genetic counseling they received was inadequate. Our data suggest that, as genetic testing for deafness becomes more widespread, it should be combined with appropriate genetic counseling, to ensure that parents and patients are given useful information that they can understand and use.

Acknowledgments

This study was supported in part by National Institute on Deafness and Other Communication Disorders research grant DC02842 (to R.J.H.S.).

Electronic-Database Information

The URL for data in this article is as follows:

Deaf World Web (2000) http://dww.deafworldweb.org/

References

Chapple A, May C, Campion P (1995) Lay understanding of genetic disease: a British study of families attending a genetic counseling service. J Genet Couns 4:281–300

Cohn ES, Kelley PM, Fowler TW, Gorga MP, Lefkowitz DM, Kuehn HJ, Schaefer GB, Gobar LS, Hahn FJ, Harris DJ, Kimberling WJ (1999) Clinical studies of families with hearing loss attributable to mutations in the connexin 26 gene. Pediatrics 103:546–550

Decruyenaere M, Evers-Kiebooms G, Denayer L, Van den Berghe H (1992) Cystic fibrosis: community knowledge and attitudes torwards carrrier screening and prenatal diagnosis. Clin Genet 41:189–196

Hietala M, Hakonen A, Aro AR, Niemela P, Peltonen L, Aula P (1995) Attitudes toward genetic testing among the general population and relatives of patients with a severe genetic disease: a survey from Finland. Am J Hum Genet 56: 1493–1500

Israel J (1995) An introduction to deafness: a manual for genetic counselors. Gallaudet University and National Society of Genetic Counselors Special Projects Fund, Washington, DC

Kenen RH, Schmidt RM (1978) Stigmatization of carrier status: social implications of heterozygote genetic screening programs. Am J Public Health 68:1116–1120

Koehn D, Morgan K, Fraser FC (1990) Recurrence risks for near relatives of children with sensori-neural deafness. Genet Couns 1:127–132

Maw MA, Allen-Powell DR, Goodey RJ, Stewart IA, Nancarrow DJ, Hayward N, Gardner R (1995) The contribution of the DFNB1 locus to neurosensory deafness in a Caucasian population. Am J Hum Genet 57:629–635

Mennie M, Compton M, Gilfillan A, Axton RA, Liston WA, Pullen I, Whyte D, Brock DJH (1993) Prenatal screening for cystic fibrosis: attitudes and responses of participants. Clin Genet 44:102–106

Middleton A, Hewison J, Mueller RF (1998) Attitudes of Deaf adults toward genetic testing for hereditary deafness. Am J Hum Genet 63:1175–1180

National Information Center for Children and Youth with Disabilities (1998) Fact sheet: general information about deafness and hearing loss. National Information Center for Children and Youth with Disabilities, Washington, DC

Rowley PT (1984) Screening and genetic counseling for β-thalassemia trait in a population unselected for interest:

comparison of three counseling methods. Am J Hum Genet 36:677–689

Smith SD (1991) Recurrence risks. Ann N Y Acad Sci 630: 203–211

Williamson R, Allison M, Bentley T, Lim S, Watson E, Chapple A (1989) Community attitudes to cystic fibrosis carrier testing in England: a pilot study. Prenat Diagn 9:727–734

Zelante L, Gasparini P Estivill X, Melchionda S, D'Agruma L, Govea N, Mila M, Della Monica M, Lutfi J, Shohat M, Mansfield E, Delgrosso K, Rappaport E, Fortina P (1997) Connexin 26 mutations associated with the most common form of non-syndromic neurosensory autosomal recessive deafness (DFNB1) in Mediterraneans. Hum Mol Genet 6: 1605–1609