

## LETTER TO JMG

# Attitudes of deaf and hard of hearing subjects towards genetic testing and prenatal diagnosis of hearing loss

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Hearing loss is an economically and socially important cause of human morbidity. It is estimated that at least 20% of the population develop clinically significant hearing loss at some time during their lives.<sup>1</sup> Hereditary hearing loss occurs in approximately 1/2000 newborns.<sup>2</sup> During the past five years, dramatic advances have been made in the mapping of more than 60 loci for non-syndromic deafness. Interestingly, quite contrary to the assumption that genetic deafness was caused by a large number of equally rare genes, it has been shown that in many populations mutations at the connexin 26 (*Cx26*) locus can account for as much as 50-80% of recessive genetic deafness.<sup>3</sup> The ability to diagnose specific forms of recessive deafness by molecular testing even in simplex families with only one affected child is rapidly becoming the standard of care for the management of such cases. However, the imminent availability of widespread testing in the deaf community has raised a number of ethical issues, some of which are unique to the genetics of deafness. For instance, some would consider prenatal diagnosis for hearing status unacceptable owing to the unique perspective on hearing loss among different groups.

Differences in the way people view the field of genetics and genetic testing for hearing loss are quite likely related to cultural differences in attitudes about hearing loss or deafness. The medical community tends to view deafness as a disability and a condition to be cured. Hearing subjects tend to share this "pathological" perspective on deafness. While most subjects with hearing loss become well integrated into the hearing world, many deaf people regard deafness and manual communication as distinctive features that define the separate, closely knit culture of the Deaf community (denoted with a capital D).<sup>4</sup> Sociologists, linguists, and anthropologists now recognise Deaf people as a special cultural and linguistic population.<sup>5,6</sup> Deaf and hard of hearing people have developed distinctive behaviour patterns, values, and norms based on their hearing loss and in response to societal attitudes towards them. Members of the Deaf community share a common identity, customs, experience, and, most importantly, language. The primary language of those in the Deaf community in the United States is American Sign Language (ASL).

The Deaf community has traditionally viewed the field of genetics with distrust, owing in large part to the "medicalisation" of deafness and awareness of the eugenics movement of the past, when efforts were made to eradicate Deaf culture.<sup>7</sup> Some Deaf subjects have expressed reluctance to participate in genetic counselling for fear that they will be told not to have children.<sup>8</sup> Failing to share the pathological perspective of deafness, members of the Deaf community often feel threatened by what they perceive as efforts to "cure" them by, for example, genetic research or the use of cochlear implants.<sup>9</sup> Indeed, some Deaf parents welcome the birth of Deaf children and seek genetic counselling to learn the chances of this occurrence.<sup>10,11</sup> It is necessary to understand and accommodate these views during genetic evaluation and counselling. An important step towards achieving this goal is to determine the attitudes of a large, diverse group of people with hearing

loss towards advances in the genetics of hearing loss. Middleton *et al*<sup>12</sup> found a predominantly negative view towards genetic advances in a small sample population comprising delegates at a conference on the "Deaf Nation" in the United Kingdom. More than half of the sample thought that genetic testing would do more harm than good. A much larger survey of 644 Deaf, 143 hard of hearing and deafened, and 527 hearing subjects with either a Deaf parent or a Deaf child ascertained from a variety of sources in the United Kingdom<sup>13</sup> showed that self-identified culturally Deaf participants were significantly more likely than hearing or hard of hearing/deafened participants to say that they would not be interested in prenatal testing for deafness. Of those hearing, hard of hearing/deafened, and Deaf participants who would consider prenatal diagnosis, 62-91% of participants in the various groups said they would use such information for preparing personally or preparing for the language needs of that child. Only a small minority of each group said that they would have prenatal diagnosis to terminate a Deaf fetus, and only 3/132 (2%) of Deaf respondents said that they would have prenatal diagnosis to terminate a hearing fetus in preference of a Deaf one.

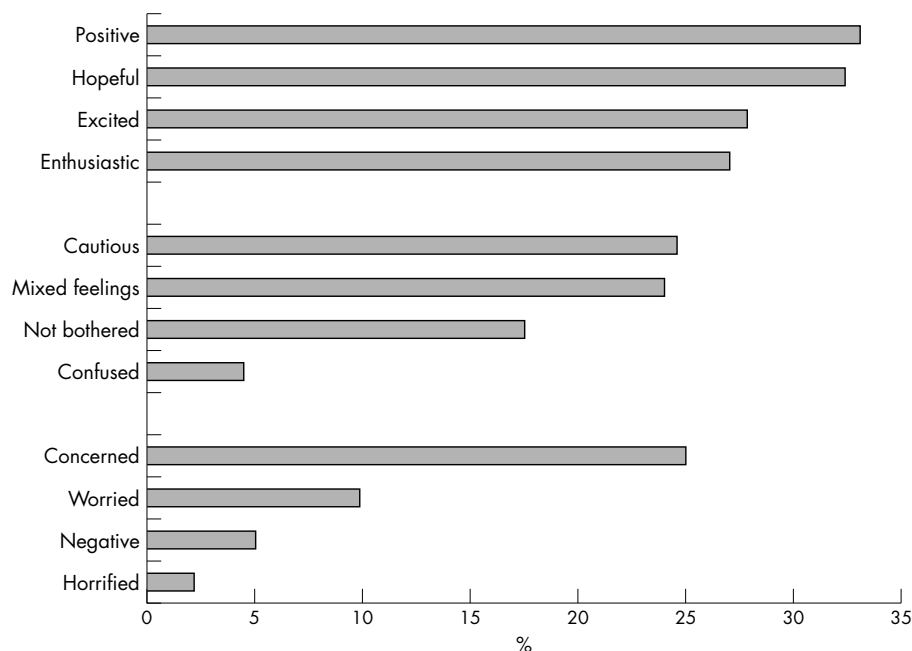
Another recent study of attitudes toward genetic testing in a group of 96 hearing parents of Deaf children in the United States<sup>14</sup> showed that the vast majority had a positive attitude towards genetic testing for deafness including prenatal testing, but none would use this information to terminate a pregnancy. Another interesting finding of this study was that even in the parents who had genetic testing, there was little understanding of genetic mechanisms and recurrence risks, suggesting that either these parents did not receive genetic counselling before testing or that the counselling did not lead to a clear understanding of these concepts. The authors emphasise the importance of appropriate genetic counselling as part of the genetic testing process.

Given these findings, the present study was designed to determine the attitudes towards the field of genetics and genetic testing for hearing loss among Deaf and hard of hearing persons in the United States with differing cultural perspectives.

## METHODS

### Participants

In the interest of studying as diverse a group of people with hearing loss as possible, study participants were ascertained from several sources, including the National Association of the Deaf (NAD), Self Help for Hard of Hearing People Inc (SHHH), the genetics clinic at the Medical College of Virginia (MCV), and the Gallaudet University student body. Two hundred questionnaires were mailed to members of each of the two national organisations residing in the Commonwealth of Virginia. One hundred and fifty-six questionnaires were mailed to patients seen in the genetics clinic at MCV between 1993 and 1999 for hearing loss in themselves or a family member. One hundred and twenty-six questionnaires were distributed



**Figure 1** Feelings about new discoveries in genetics. Percentage of all respondents who checked from a list of 12 adjectives shown when asked to describe how they feel about new discoveries in genetics.

to students in classes at Gallaudet University, a liberal arts college for the deaf in Washington, DC. Of 682 questionnaires distributed, 337 (54%) were returned. The response rate varied widely among the groups from virtually a 100% response when questionnaires were handed out in person, to a more typical response rate of 36% for mailed questionnaires.

Of the 337 respondents, 56% were female, and their ages ranged from 18 to 95. Forty percent of respondents considered themselves deaf, 42% hard of hearing (HOH), 7% late deafened, and 11% hearing. When asked to state whether they were more culturally involved with the Deaf or hearing community, 46% identified with the hearing community, 24% identified with the Deaf community, and 30% indicated that they had an equal involvement in both communities.

### Questionnaire

The questionnaire developed by Middleton *et al*<sup>12</sup> was modified, including minor language changes, the inclusion of a HOH category, and the addition of questions concerning feelings about termination of pregnancy. The adapted questionnaire included 22 closed questions concerning feelings about new genetic discoveries, the expected impact of genetic testing on the Deaf community, the preferred hearing status of participants' children, interest in prenatal diagnosis for hearing loss, and sociodemographic data. The survey was anonymous and voluntary. The introduction to the questionnaire included no information on genetics and informed respondents that the data would be used to help educate medical professionals about consumer opinions. Statistical analysis was carried out using the SAS software package, to test the hypothesis that variation in cultural involvement would affect opinions about genetic testing, preferred hearing status of children, interest in prenatal diagnosis for hearing loss, and feelings about termination of pregnancy for hearing status.

## RESULTS

### Opinions about genetics and genetic testing

Respondents as a whole expressed a positive view of the field of genetics. They were instructed to check as many adjectives as they wished to describe their feelings about new genetic

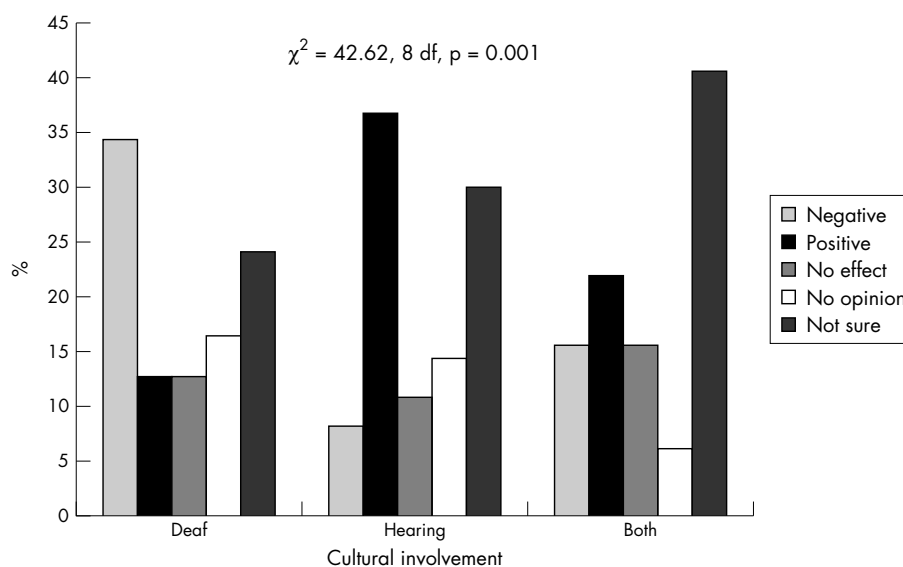
discoveries. As shown in fig 1, the most commonly chosen response was "positive" (33%) and the least commonly chosen response was "horrified" (2%). It is important to note that while the majority of adjectives checked could be described as positive, a significant portion of the sample population checked less positive or neutral adjectives, the most common being "concerned" (25%).

Participants differed in their opinions of the potential impact of genetic testing for deafness on the deaf community (here used with a lower case d to be more inclusive). As shown in fig 2, culturally Deaf respondents were approximately four times more likely to feel that genetic testing would have a negative effect on the deaf community than those who identified themselves with the hearing community (odds ratio = 5.84, 95% CI 2.76-12.38). Among those who had equal involvement in both communities, more than 40% were unsure of the effect. The opinions regarding the effect of genetic testing were significantly different among the three groups ( $\chi^2=42.6$ , 8 df,  $p=0.001$ ).

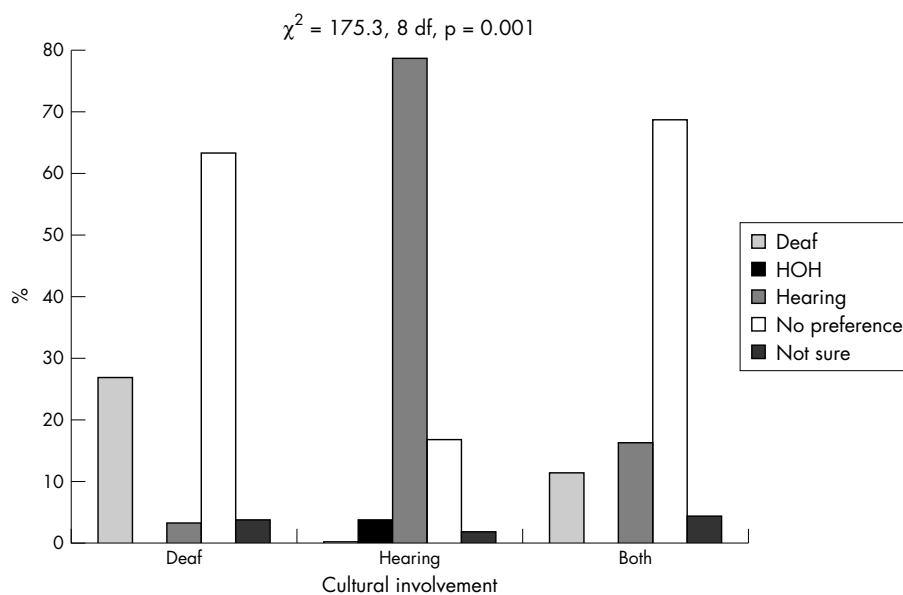
### Preferred hearing status of children and interest in prenatal diagnosis

Overall, the majority of respondents indicated that they had no preference for either Deaf, HOH, or hearing children. However, there were highly significant differences in preference among the cultural groups as shown in fig 3 ( $\chi^2=175.3$ , 8 df,  $p=0.001$ ). The majority of both the culturally Deaf respondents and those in the equal involvement group indicated that they had no preference, while the vast majority of those from the hearing community indicated that they would prefer to have hearing children. Of respondents among the Deaf and equal involvement groups, 27% and 11% respectively indicated a preference for Deaf children.

Interest in prenatal diagnosis for hearing loss was also significantly different with regards to cultural involvement ( $\chi^2=25.5$ , 4 df,  $p=0.001$ ), shown in fig 4. When asked whether they would want to take a test to find out if a baby was deaf, HOH, or hearing before it was born, the majority of participants in the Deaf and equal involvement groups indicated that they would not want to take this test. The majority of participants who identified with the hearing community indicated that they would want to take the test or were



**Figure 2** Relationship between cultural involvement and opinions about genetic testing. Responses to the question “Some people feel that the use of genetic testing for deafness will have an effect on the Deaf community. Others disagree. What do you think?” Percentage of respondents grouped by self-identified cultural involvement (Deaf = more culturally involved with the Deaf community, Hearing = more culturally involved with the hearing community, Both = equal involvement in both communities).

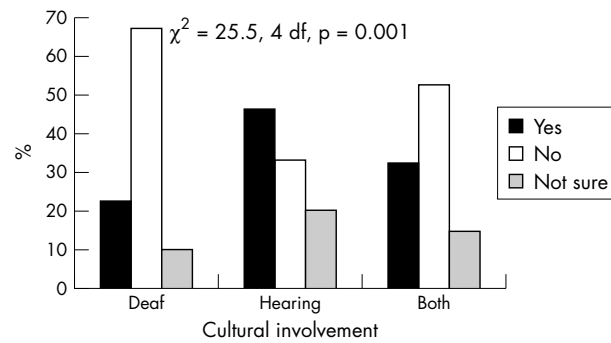


**Figure 3** Relationship between cultural involvement and preferred hearing status of children. Percentage of respondents preferring Deaf, hard of hearing (HOH), or hearing children, or who had no preference or were not sure. Respondents grouped by self-identified cultural involvement (Deaf = more culturally involved with the Deaf community, Hearing = more culturally involved with the hearing community, Both = equal involvement in both communities).

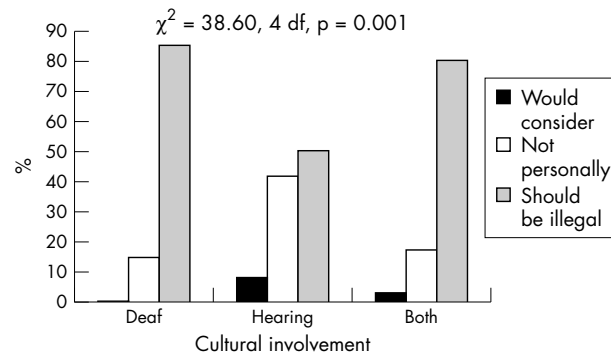
unsure. There were no associations between interest in prenatal diagnosis and the respondent’s sex, whether they already had children, or whether they already had a Deaf or HOH child. Those participants who acknowledged wanting to take this test were asked to choose from a list of reasons that they would want a test during pregnancy. The following responses were chosen: “to prepare myself personally for the child’s needs (79%), to prepare for the language needs of my child (76%), to avoid putting my child through unnecessary medical tests (33%), to consider an abortion if the baby is deaf (9%), and to consider an abortion if the baby is HOH (3%)”. No participants chose the response “to consider an abortion if the baby is hearing.”

**Opinion about termination of pregnancy for hearing status**

Participants were asked their opinion of aborting a Deaf or HOH baby when a hearing baby was preferred. As shown in fig 5, the cultural groups displayed significant differences in their opinions ( $\chi^2=38.6, 4 \text{ df}, p=0.001$ ). The majority of all respondents felt that aborting a Deaf or HOH baby should be illegal, but 42% of respondents in the hearing cultural group indicated that while they would not personally consider this option, it should not be forbidden for others. This is in contrast to only 15% and 18% of respondents in the Deaf and equal involvement groups, respectively. In addition, 8% of respondents in the hearing group indicated that they would consider



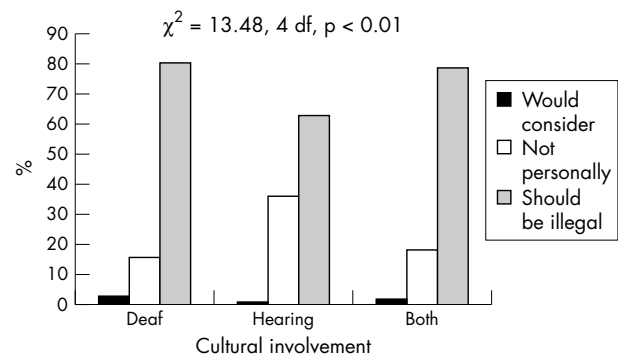
**Figure 4** Relationship between cultural involvement and interest in prenatal diagnosis. Percentage of respondents for the following question: "Suppose you or your partner were pregnant. Would you want to find out whether the baby is deaf or hearing before it is born?" Respondents grouped by self-identified cultural involvement (Deaf = more culturally involved with the Deaf community, Hearing = more culturally involved with the hearing community, Both = equal involvement in both communities).



**Figure 5** Relationship between cultural involvement and opinions about abortion of a Deaf baby when a hearing baby is preferred. Percentage of respondents grouped by self-identified cultural involvement (Deaf = more culturally involved with the Deaf community, Hearing = more culturally involved with the hearing community, Both = equal involvement in both communities). Would consider = would consider aborting a Deaf baby, Not personally = would not personally abort a Deaf baby, but it should not be forbidden for others, Should be illegal = it should be illegal to abort a Deaf baby.

an abortion of a Deaf or HOH baby, while only 2% of respondents in the equal involvement group indicated that they would consider this, and none of the Deaf respondents indicated that they would. Opinions about the abortion of a Deaf baby were not affected by the respondent's sex, whether they already had children, or whether they already had a deaf or HOH child.

The converse of the latter question was also posed to the participants, namely how they felt about the abortion of a hearing baby when a Deaf or HOH baby was preferred. Significant differences among the groups were once again evident ( $\chi^2 = 13.5, 4 \text{ df}, p = 0.01$ ), shown in fig 6. The majority of respondents overall felt that this should be illegal, but those participants in the hearing cultural group were more likely to respond that they would not personally consider this but that it should not be forbidden for others (36% versus 19% in the equal group and 16% in the Deaf group). In addition, 1%, 2%, and 3% of participants from the hearing, equal involvement, and Deaf cultural groups, respectively, indicated that they would consider an abortion for this reason. The opinion about abortion of a baby because it was hearing was not significantly different based on sex, already having children, or having a Deaf or HOH child.



**Figure 6** Relationship between cultural involvement and opinions about abortion of a hearing baby when a Deaf baby is preferred. Percentage of respondents grouped by self-identified cultural involvement (Deaf = more culturally involved with the Deaf community, Hearing = more culturally involved with the hearing community, Both = equal involvement in both communities). Would consider = would consider aborting a hearing baby, Not personally = would not personally abort a hearing baby, but it should not be forbidden for others, Should be illegal = it should be illegal to abort a hearing baby.

## DISCUSSION

This study provides evidence of a disparity in attitudes towards prenatal testing for hearing loss between culturally Deaf subjects and persons with hearing loss who are culturally associated with the hearing community. Culturally Deaf subjects in this study were more likely than others to feel that genetic testing for hearing loss would have a negative effect on the Deaf community. The majority of culturally Deaf subjects and those who identified equally with both the Deaf and hearing communities expressed no preference regarding the hearing status of their children, were not interested in prenatal diagnosis for hearing loss, and were more likely to feel that termination of pregnancy based on hearing status (either deaf/HOH or hearing) should be illegal than were participants who were culturally associated with the hearing community. These results are in agreement with those of Middleton *et al.*<sup>12, 13</sup> The current study also points out the significant differences in attitude between culturally Deaf subjects and persons with hearing loss who identify culturally with the hearing community. Together, these surveys show the concern in the Deaf community over the implications of genetic testing. It is also important to point out that the vast majority of respondents from the Deaf community (97%) indicated that they would not consider the abortion of a hearing fetus. In fact, the majority (>80%) felt it should be illegal to abort a hearing fetus.

These data are particularly important for genetic counselors and clinicians to be aware of when working with clients with hearing loss. These professionals, while firmly based in a non-directive tradition that emphasises patient autonomy, are nevertheless accustomed to viewing genetic conditions such as hereditary deafness as "disorders" or "diseases." When a Deaf couple seeks genetic counselling with the goal of having a deaf child, the genetics professional is put to "the ultimate test of nondirective counselling."<sup>15</sup> A more common clinical scenario involves hearing parents seeking information after the birth of a deaf child. Members of the Deaf community point out that hearing parents who have never met a Deaf adult have difficulty making informed decisions about their Deaf child's education and mode of communication and should be encouraged to interact with Deaf and HOH adults at the time these decisions are being made.<sup>16</sup> This argument is similar to that of the disability community, which advocates that balanced information, including contact with disabled persons outside a medical setting, be presented to parents in

prenatal genetic counselling sessions.<sup>17</sup> Clinicians and counsellors need to be aware of, and sensitive to, the cultural heritage and values of the varied members of the Deaf community in general, in addition to those of the hearing parents seeking information, in order to provide appropriate genetic counselling and medical guidance.

During the past 200 years, the social, economic, and educational circumstances of the Deaf have improved dramatically, as has their fertility.<sup>18</sup> There is a growing recognition that the existence of genetic causes of deafness, in conjunction with relaxed selection and a tradition of intermarriage among the Deaf, may have increased the incidence of deafness in many countries, with selective amplification of connexin 26 deafness, the most common form of recessive deafness.<sup>19</sup> Genotypic, rather than phenotypic, marital selection (that is, the selection of a mate based on the results of a genetic test for connexin 26) would further accelerate this process and represents yet another ethical dilemma posed by these new technologies. By seeking an appropriate partner, some people with specific forms of recessive deafness, such as that caused by connexin 26 mutations, could either avoid or ensure the birth of a deaf child. Those who might be troubled by the eugenic effects of genotypic mate selection should consider the consequences that other forms of non-random mating have had in maintaining the frequencies of traits, such as sickle cell anaemia and Tay-Sachs disease.<sup>20</sup> While we would neither advocate nor discourage mate selection based on the results of a genetic test, it is clearly an example of one way in which genetic knowledge can empower the Deaf community, among whom the birth of a Deaf child is highly valued.

One potential limitation of this study that must be taken into account when interpreting the data concerns the representativeness of the populations from which the study group was ascertained. The majority of the participants were members of organisations or institutions for Deaf subjects. Membership of such a group may, in itself, introduce bias. In order to limit this potential source of bias, a much larger survey of people with hearing loss in the United States, and of those associated with the Deaf community, is currently being designed. Efforts will be made to ascertain subjects who are not affiliated to organisations or institutions for the Deaf and HOH.

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