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Prenatal Diagnosis for Inherited Deafness—What is the Potential Demand?

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Genetic testing for inherited deafness is now available within some genetics centres. This study used a structured questionnaire to assess the potential uptake of prenatal diagnosis (PND) for inherited deafness, and document the opinions of deaf and hearing individuals toward PND and termination of pregnancy (TOP) for hearing status. Participants were self-selected from the whole of the UK, of whom 644 were deaf, 143 were hard of hearing or deafened, and 527 were hearing individuals who had either a deaf parent or child. The results showed that 21% of deaf, 39% of hard of hearing and deafened, and 49% of hearing participants said they would consider PND for deafness. Six percent of deaf, 11% of hard of hearing and deafened, and 16% of hearing participants said they would consider a TOP if the fetus was found to be deaf. Two percent of deaf participants said they would prefer to have deaf children and would consider a TOP if the fetus was found to be hearing.

KEY WORDS: deafness; prenatal diagnosis; termination of pregnancy; questionnaire; ethics; attitudes.

INTRODUCTION

The majority of deafness in the developed world is caused by genetic factors (Cohen and Gorlin, 1995). Genetics is responsible for both congenital, profound deafness as well as adult-onset, progressive deafness. Research into the molecular genetics of deafness has advanced enormously in recent years (Grundfast *et al.*, 1999; Skvorak Giersch and Morton, 1999). To date more than 60 nonsyndromal deafness genetic loci have been identified (Van Camp and Smith, 2000). Mutations in one particular gene, Connexin 26, are of great interest to clinicians working with

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deaf families since they are thought to be the most common cause of nonsyndromal deafness (Cohn and Kelley, 1999). A consequence of the molecular research means that diagnostic, carrier, and prenatal genetic testing for inherited deafness will become part of routine clinical practice within genetics services. Many different genetics centres around the world now offer genetic testing for mutations in Connexin 26. Research is needed to evaluate the impact of such testing before a clinical service is made available (Garvican, 1998). This can be done by consideration of the opinions of the individuals for whom the tests are relevant (Hietala *et al*., 1995).

It is likely that there will be variation in the attitudes toward genetic testing for deafness, principally because deafness can be perceived in different ways. The "medical model" views deafness as a pathology to be treated or cured, whereas the "cultural model" views deafness as a condition to be understood and preserved (Arnos *et al*., 1991). Culturally Deaf people (written with an uppercase "D") often have very positive attitudes toward their deafness, seeing themselves as part of a distinct cultural community sharing common values and identity (Padden, 1980). For many in the Deaf community, deafness is not viewed as a disability. Rather it is society's attitudes that cause the deaf individual to be considered disabled.

Researchers working in the field of inherited deafness are excited by the prospect of rapid incorporation of molecular genetic developments into routine clinical practice (Reardon, 1998). However, the use of prenatal diagnosis (PND) for inherited deafness should be treated with caution. A consequence of the varying attitudes toward deafness means that it is debatable whether deafness is a serious enough condition to warrant the offer of PND and whether termination of pregnancy (TOP) is an acceptable option for parents who do not want a child of a particular hearing status. It could be assumed that most parents would prefer to have hearing children. However, previous research, which contributed toward the pilot work for this project, has suggested that some deaf individuals would prefer to have deaf children and others feel that the use of genetics threatens the vitality of the Deaf community (Middleton *et al*., 1998). Many hearing persons may find this difficult to comprehend. Our previous study led us to hypothesize that deaf individuals would be less interested in PND for deafness than would hearing parents of deaf children.

Throughout the text of this paper the term "deaf" is used to refer to all individuals affected by hearing loss including the culturally Deaf; the term "Deaf" refers to culturally Deaf individuals only.

SUBJECTS AND METHODS

Subjects

Participants were classified according to how the individual perceived their hearing status; participants had to say whether they viewed themselves as deaf, hard

of hearing or deafened, or hearing with a deaf parent or child. Most participants who considered themselves deaf had a profound, congenital or early onset hearing loss, approximately half of whom used British Sign Language (BSL) as their preferred language. Those who considered themselves hard of hearing tended to have a hearing loss in the mild to severe range, which was congenital or early onset. Those who were deafened had varying levels of hearing loss, which occurred later on in life.

Ascertainment

Participants were ascertained from all over the UK via a total of 28 different sources,which included hospital departments such as audiology, ENT, and genetics, as well as social services, teachers for the deaf, charities for the deaf, schools and colleges for the deaf, and conferences for the deaf. Participants were sent a letter and a consent form via a health/education professional they were in contact with giving them details of the project and asking them to return the consent form if they wished to participate. Participants were given the option of taking part by completing a postal questionnaire or receiving a structured interview with a BSL interpreter if requested. The study questionnaire was also sent out as part of three magazines for the deaf in the UK. Also, articles about the research were written for other deaf magazines and Ceefax and Teletext sites on the TV used by deaf viewers. Recipients of such articles were encouraged to contact the authors if they wanted to take part. Written informed consent was obtained from all participants who were contacted via a professional they were involved with and also from participants who responded to an article about the study. For participants who received a questionnaire through a magazine mailing it was assumed they would only complete and return it if they wanted to take part in the study (i.e. consent was implicit). Ethical approval was granted for the study from the St James's University Hospital Ethics Committee, Leeds, UK, where the study was conducted.

Questionnaire

A questionnaire was developed with input from deaf sign language users so that it was accessible to participants whose first language was BSL. It took approximately 20 min to complete. Themes relevant to the study were obtained from more than 100 different papers published in the medical and social sciences literature, adapted for the study and processed through three pilot studies. The questionnaire contained 21 questions of which only a selection have been chosen for discussion here. These included assessing preference for having deaf or hearing children, interest in utilizing a test in pregnancy (PND) for deafness, reasons for having PND for deafness, Deaf cultural identity, and sociodemographic information. Since the study was of a descriptive nature, statistical analysis involved group comparisons

Table I. Sociodemographic Information for the Sample

Sociodemographic characteristics	Total sample size (1314)	%
Age range		
13–19	36	3
20–29	152	12
30–39	441	35
40–49	390	30
50–59	179	14
60–69	54	4
70–93	32	2
Female	833	63
Had children	958	73
Married or living with partner	930	71
Owned own house/flat	922	70
Had a religious affinity	662	50

using chi-squared analysis with a $p < .0001$ level of significance. The questionnaire consisted of single measures (i.e., yes/no dichotomous responses) and thus a reliability statistic did not need to be calculated.

RESULTS

Completed questionnaires were received from 644 deaf individuals, 143 hard of hearing and deafened individuals, and 527 hearing individuals with either a deaf parent or deaf child. Sociodemographic data is given in Table I. Participants considered culturally Deaf were identified as those with a hearing loss (deaf, hard of hearing and deafened) who used BSL as their first language and said they associated with the Deaf community. Therefore, all participants with any hearing loss were re-classified according to cultural Deafness, details of which are given in Table II.

Eight hundred and thirty five questionnaires were sent to professionals to pass onto potential participants. Out of these, 458 returned completed postal questionnaires and 10 requested an interview, total = 468 (response rate 56%). One hundred and thirty nine questionnaires were sent out to participants who requested them after reading about the study in the press, of these 76 returned completed questionnaires (response rate 55%), and 5,700 questionnaires were sent out in the magazines for the deaf, from which 770 participants returned completed questionnaires (response rate 14%). Therefore, a total of 1,314 questionnaires were completed for the study. Data collection was conducted from June 1998 to June 1999.

Table II. Number of Culturally and Nonculturally Deaf Participants

All participants with a hearing loss	644	
Culturally Deaf	212	33%
Nonculturally deaf	452	66%

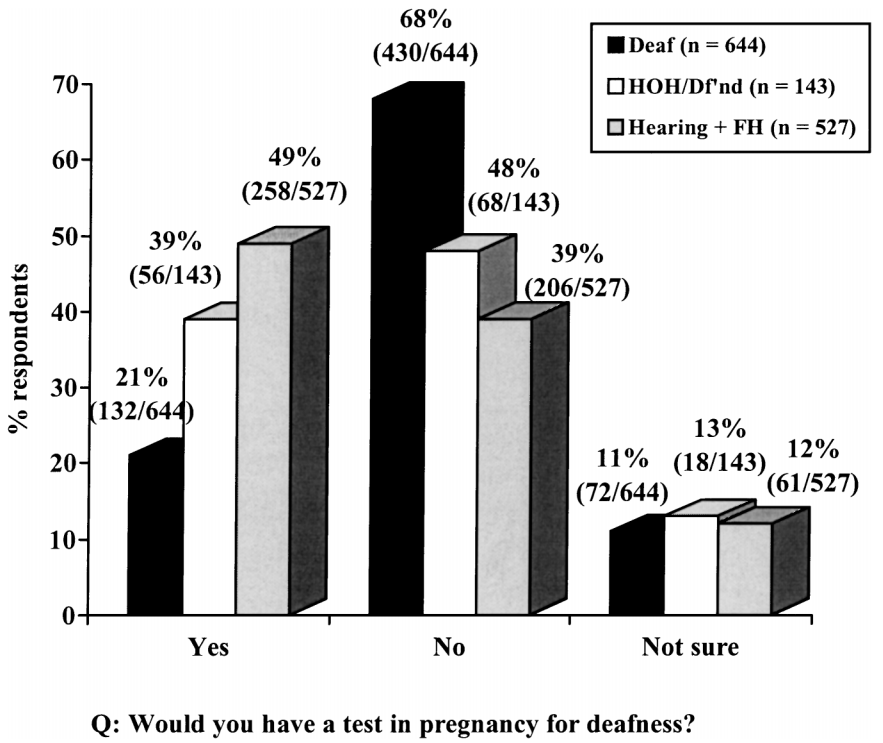

Fig. 1. Percentage of participants who were interested in a test in pregnancy for deafness (*Note*: HOH/DF'nd = Hard of hearing and deafened participants; Hearing + FH = Hearing participants who have either a deaf parent or deaf child).

Interest in Utilizing PND for Deafness

Figure 1 shows that deaf participants were more likely to say "no" to prenatal testing than the other two groups and hearing participants were more likely to say "yes" to prenatal testing than the other two groups ($\chi^2 = 113.1$, df = 4, $p < .0001$). If the results from the deaf and hard of hearing and deafened groups are re-classified according to Deaf cultural affinity, it is clear that the culturally Deaf are more likely than nonculturally deaf participants to say "no" to prenatal testing ($\chi^2 = 21.1$, df = 2, $p < .0001$; Fig. 2).

A subset of the data from Fig. 1 (those who answered "yes" to PND for deafness) was classified with the data from a different question assessing preference for having children of a specific hearing status. The results of this are presented in Fig. 3 and show that while hearing (177/258, 69%) and hard of hearing and deafened (34/56, 61%) participants might use PND for deafness to have hearing children, deaf participants were not so emphatic, most who wanted PND for

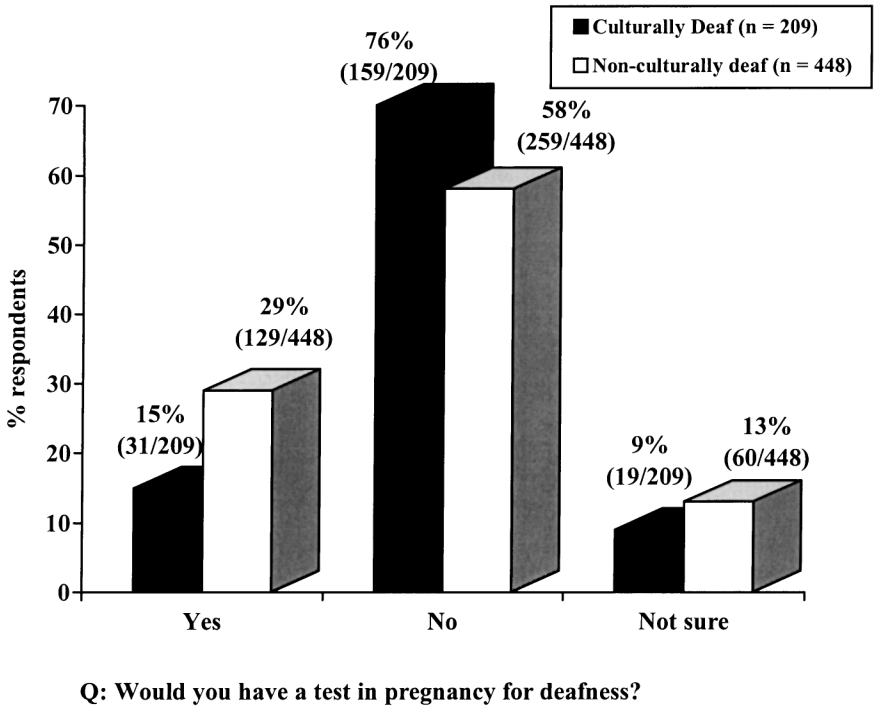

Fig. 2. Percentage of culturally deaf and nonculturally deaf participants who were interested in a test in pregnancy for deafness.

deafness did not mind the hearing status of future children (73/132, 56%; $\chi^2 = 41.3$, df $= 2$, $p < .0001$). Of those deaf participants who had a preference, 48/132 (36%) preferred to have hearing children and 11/132 (8%) preferred to have deaf children. Of those deaf participants who preferred to have deaf children the majority (9/11, 82%) were culturally Deaf.

Reasons for Having PND for Deafness

When participants were asked for specific reasons for having PND, the majority of all participant groups said they would have a test in pregnancy for deafness so they could prepare for the child, either for the language needs of the child or personally (Fig. 4). Most participants indicated that they would not act on the information gained from a prenatal test by having a TOP. However, a minority of each group (8/132, 6% deaf; 6/56, 11% hard of hearing and deafened; 41/258, 16% hearing participants) said they would consider a TOP if the child was found to be deaf. A small minority of the deaf group (3/132, 2%) said they would consider a

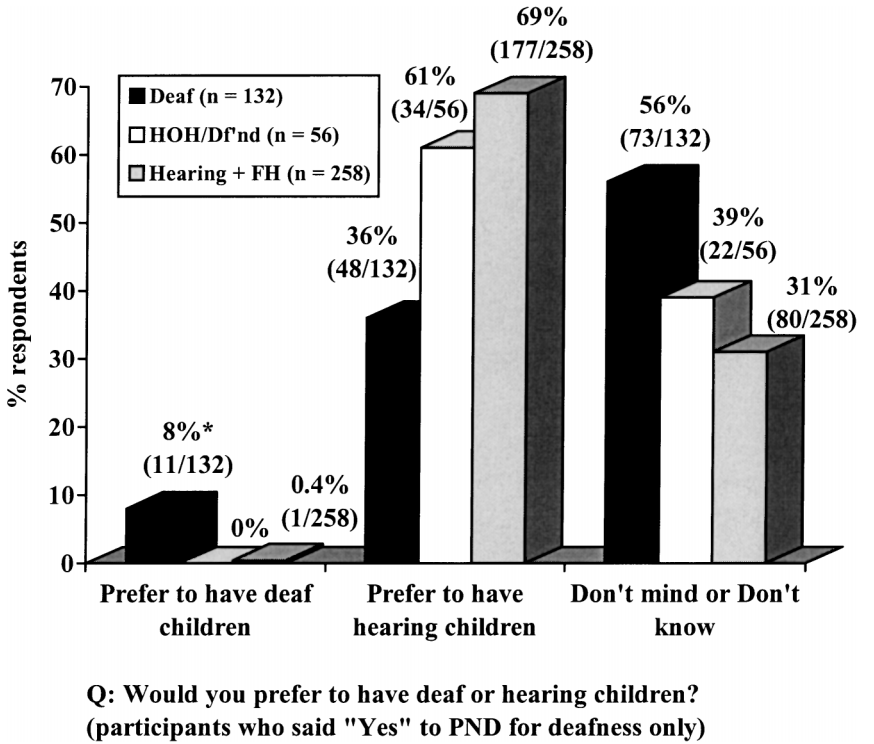

Fig. 3. Percentage of participants who wanted PND and preferred to have a child of a specific hearing status (*of these 8% (11/132), 82% (9/11) were culturally Deaf).

TOP if the baby was found to be hearing. Of these three participants, two were culturally Deaf and one was nonculturally deaf.

DISCUSSION

The aim of this research was to determine attitudes toward prenatal genetic testing for inherited deafness. By gaining a better understanding of the potential uptake of PND and TOP the question can be asked about whether PND for inherited deafness should be available.

Hearing participants with a family history of deafness were more likely than the other groups to be interested in using PND for deafness, presumably because deafness is seen more negatively by people who do not personally have a hearing loss. Culturally Deaf participants were the least likely to be interested in PND for deafness; this is likely to be because prenatal genetic testing is perceived to threaten the continuation of the Deaf community (Middleton *et al*., 1998).

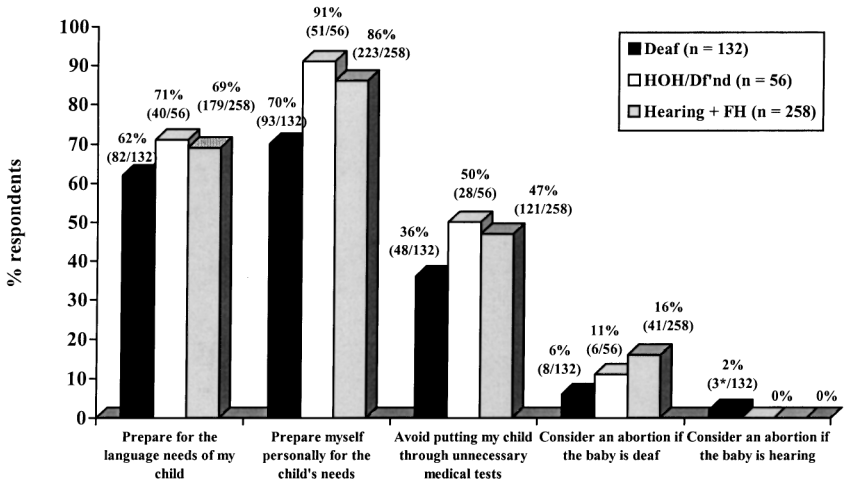

Fig. 4. Reasons participants gave for having PND for hearing status (*of these three given in the final column, two were culturally Deaf and one nonculturally deaf).

The majority of hearing and hard of hearing and deafened participants who were interested in PND for deafness preferred to have hearing children, they may therefore seriously consider using PND with selective TOP to ensure that they have hearing children. The results indicate that deaf participants were not as interested in this. The majority of deaf participants did not mind the hearing status of future children, presumably because they cope adequately with their own deafness and so do not see having deaf children as a problem. However, 8% of deaf participants who wanted PND for deafness preferred to have deaf children. Such participants were, in the majority, culturally Deaf. It is logical that culturally Deaf people would wish to pass on deafness to the next generation since this would keep their language and culture alive (Jordan, 1991). The remaining deaf participants who preferred to have deaf children may have done so because they worried about how they would teach speech to a hearing child. Some already had deaf children and so may have wanted another so that their children could experience the deafness together. This may also be the reason why one hearing participant also preferred to have deaf children. A preference for having deaf children is not a new phenomenon; other studies have also reported this finding both anecdotally (Jordan, 1991) and statistically (Middleton *et al*., 1998). However, such previous research has concentrated on the views of those who were culturally Deaf, without consideration of the opinions of those utilizing the medical perspective of deafness. The present study has ascertained the views of the wider deaf community and in

doing so has attempted to avoid being culturally biased (in a Deaf sense). This means that the figures given in this study might be a closer representation of the real situation.

Although it is obvious that some participants may be keen to use PND for deafness with selective TOP, most participants from all groups said they would not act on the results of a prenatal genetic test. The majority said they would only have PND for deafness in order to prepare for the language needs of the child or else prepare personally for the child's needs. Since invasive PND procedures involve an increased risk of miscarriage, the role of PND needs to be clear. Is it acceptable for prenatal genetic diagnosis to be offered just for the information it provides when it is known that the parents have no intention of considering a TOP? Or should PND only be made available if parents intend on acting on the information provided by having a TOP? On the other hand, since attitudes do not necessarily predict behavior, individuals who say that they would not have a TOP, when actually faced with a pregnancy, may behave differently. It would be useful in future studies to document the views of participants who are currently pregnant and where the option of PND is a realistic possibility. Since the attitude–behaviour prediction is questionable, decisions over whether PND should be available should not be decided on the basis of attitude studies alone.

A minority of all groups said they wanted to have PND for deafness so they could abort a fetus of the "wrong" hearing status. More hearing participants were interested in TOP for deafness and only deaf participants were interested in TOP for a hearing fetus. The fact that even a small percentage of deaf participants said that they would abort a healthy hearing fetus because they preferred to have deaf children shows the strength of feeling on this subject. Two out of the three deaf participants who said that they would have a TOP for a hearing fetus were culturally Deaf. This fits in with previous assumptions about Deaf cultural attitudes, that is, that to be deaf and to pass on deafness to children is a positive experience. It is possible that a deaf person who said he/she preferred to have deaf children would in reality be content with having a hearing child and would not go as far as terminating a hearing fetus. Actively choosing to abort a wanted pregnancy because it has the "wrong" hearing status would be traumatic for any couple. However, if preimplantation genetic diagnosis for inherited deafness were offered, it is possible that couples might be more open to selecting an embryo with a specific genetic hearing status than they would be to having PND with selective TOP.

The ethics of prenatal testing in general is well documented (Burgess, 1994; Kapp, 1994). The American Medical Association (1994) states that

> The dilemma posed by new genetic technologies is the question of how far parents' authority over their children should extend and, in particular, how completely parents should be able to control the genetic composition of their children.

If PND for deafness becomes part of routine clinical practice then all clients who wish to use it should be able to, whether they are deaf, Deaf, or hearing. However,

it is logical to question whether it is acceptable for a healthy hearing fetus to be aborted because its parents want a deaf child. Davis argues that deliberately closing options for a child is unethical, that is, it is morally wrong to actively choose to have a deaf child (Davis, 1997). The decisions to perform PND for nonlethal genetic conditions such as deafness rely on the discretion of the individual clinician. There are currently no guidelines, which recommend whether PND for deafness is acceptable, or not. An international study of geneticists and genetic counsellors looking at the attitudes toward PND for many different scenarios including deafness showed that 9% of British and 35% of American genetics professionals would perform PND for a deaf couple wanting to have deaf children (Wertz, 1999). Therefore, there is no obvious consensus of opinion on this subject.

The medical genetics profession finds itself in an increasingly impossible position if it totally subscribes to the model of nondirective counseling. If a deaf person requests PND for hearing status with the intention of aborting a hearing fetus, then for a geneticist to be truly nondirective, respecting client autonomy, he/she should allow them to do this. However, if genetics professionals are not comfortable offering PND for indications such as deafness, the "wrong" sex, or a treatable physical defect, then they could be criticised for denying some individuals access to services or options they feel strongly that they should have a right to use.

As genetic testing technology becomes possible for more nonlife-threatening conditions, has the time come to decide whether genetics professionals will be led by the demand for such testing? Or perhaps decisions should be made to refuse PND for certain conditions?

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