ELECTRONIC LETTER

Genetic information but not termination: pregnant women's attitudes and willingness to pay for carrier screening for deafness genes

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About 1 in 1000 British children are born deaf.[1] Although the true genetic contribution to the aetiology is unknown, it is estimated that genetic causes account for at least 60% of cases.[2] Of genetic cases, 70% of deafness occurs in the absence of other clinical features and is termed "non-syndromic". Recessive genes account for 80% of such deafness, which represents about one-third of all hereditary deafness. At the time of writing, 33 recessive non-syndromal deafness genes have been mapped and 17 have been identified.[3] Despite this genetic heterogeneity, sequence variations in the connexin26 (or gap junction beta-2, *GJB2*) gene account for up to 50% of cases of non-syndromic, prelingual, sensorineural hearing loss in some populations.[4 5] Connexin26 mutations account for 17% of severe and 30% of moderate, non-syndromal deafness in the UK.[6] Its contribution to hearing loss world wide is variable, but the 35delG allele alone accounts for approximately 10-20% of cases of hearing loss in white people of northern European descent, and approximately 30–40% of cases in Mediterranean regions.[5] The heterozygote frequency of 35delG varies from 1-3% in white populations of north European origin, and is about 1 in 30 in those of Mediterranean descent.[5]

Connexin26 testing is now available for affected subjects and their families on a diagnostic basis in the United States and Europe, and its use in prenatal genetic counselling is being piloted in Greece.[7] Antenatal screening for connexin26 mutations alone is unlikely to be considered in the UK as only a minority of non-syndromic deafness would be detected.[6] However, as advances in mutation screening technology such as gene chips take place, it is possible that carrier screening for a wider number of recessive deafness genetic variants may become technically feasible within the next few years. Such carrier testing could be offered either before or early in pregnancy. Carrier couples identified pre-pregnancy would have the options of taking their chances, avoiding pregnancy, preimplantation diagnosis, or gamete donation. Those identified during pregnancy would have the opportunity of prenatal diagnosis and termination of any affected fetus, or preparation for the health and educational needs of the neonate.

While it is widely recognised that genetic screening and prenatal diagnosis should not reflect a eugenic policy,[8] there is no consensus concerning what conditions justify carrier screening or termination. Furthermore, there has been concern that advances in human genetics are being determined by professionals, with little consideration being given to the views of the general population. While there are some studies looking at attitudes of given populations to prenatal testing and/or gene therapy for some diseases,[9] little is currently known about the public's attitudes towards either the introduction of carrier screening for deafness, or the termination of any affected fetus. Indeed, there is controversy among genetics professionals as to whether prenatal diagnosis and termination are justified on the grounds of deafness alone.

What is clear from the limited research is that the deaf and hearing often have different views towards genetic testing for deafness. Further, the culturally Deaf (those positive about deafness, proud to be deaf, and belonging to a community with their own language, history, culture, and identity[10]) see deafness positively. For example, Middleton *et al*[11] conducted a survey to establish the attitudes of deaf adults in the United Kingdom towards genetic testing for deafness. The survey was administered at an international conference of the "Deaf Nation", and thus sampled respondents from the "Deaf community" (denoted with a capital D). The results of this study indicate that the Deaf community have a negative attitude towards genetic testing for hereditary deafness, with one-third of respondents preferring to have a deaf child. In a later study Middleton *et al*[12] extended this study to investigate the attitudes of the deaf, hard of hearing or deafened, and the hearing who had either a deaf child or parent. The latter group has a more positive attitude towards both prenatal diagnosis for inherited deafness and termination of pregnancy if the fetus was found to be affected. In this study, 2% of deaf respondents said they would prefer to have a deaf child, and would consider termination if the fetus was of hearing status.

Key points

- With improving molecular genetic techniques, it is likely that carrier screening for recessive deafness genes will be possible within the next few years. Decisions about the provision of such a test will therefore have to be made. This study investigated the attitudes and willingness to pay of non-deaf pregnant women for deafness carrier screening.
- The design was a structured self-completion questionnaire and the setting was Aberdeen Maternity Hospital, Scotland. The participants were 104 non-deaf pregnant women attending for their 12-13 week booking visit scan.
- Seventy-two percent of respondents would want to know if they were a carrier of a deafness gene and 74% would have prenatal diagnosis of the fetus's hearing status. Only 7% of respondents would terminate an affected pregnancy. Mean willingness to pay for deafness carrier screening was £42. Willingness to pay was related to age, perception of risk of having a deaf child, views on antenatal serum screening, and positive attitudes to deafness carrier testing.
- While non-deaf pregnant women value carrier screening for deafness, the majority would not abort an affected fetus. A number of ethical and research implications of this study are discussed.

Similar results have been found in the United States. Brunger *et al*[13] investigated the attitudes of parents with normal hearing who have one or more deaf children. A positive attitude towards genetic testing was recorded for the majority of respondents, though none would use the information to terminate a pregnancy. Stern *et al*[14] investigated the attitudes of deaf and hard of hearing subjects towards genetic testing and prenatal diagnosis of hearing loss. Consistent with the work of Middleton *et al*,[12] this study found differences in attitudes between culturally Deaf subjects (who had a negative attitude) and those with a hearing loss who are not associated with the "Deaf community" (who had a more positive attitude).

As technology advances, priorities for provision of new carrier testing programmes will have to be set. Such priorities will be determined by a number of factors, including ethical and moral questions, legal factors, professional views, consumer demand, and economic considerations. This study considers the latter two factors, consumer demand and economic considerations. We wondered if pregnant women in the UK, with no family history of congenital deafness, would take up such a screening programme and, if so, what value they would place on the opportunity of a "recessive deafness gene carrier screen".

METHODS

Ethical committee approval for the study was obtained from Grampian Joint Ethics Committee. Consecutive women attending Aberdeen Maternity Hospital for their 12-13 week booking scan were asked to read an information sheet about involvement in the study and complete a consent form. They then read a second information sheet about genetic deafness and carrier testing and completed a structured questionnaire (the information sheets, consent form, and questionnaire are available from the authors). Respondents were informed that subjects who were defined as being at high risk following carrier screening would be given the option of prenatal testing. Given other studies, recruitment was restricted to clinics on certain days. The questionnaire was either completed at the clinic or returned by post (a stamped addressed envelope was included and one reminder was sent). A pilot study was carried out on 16 subjects to assess whether respondents understood the questionnaire. Following this, 135 subjects were asked to take part in the main study (which formed the data for the analysis).

The questionnaire was in three sections. The first section elicited respondent's attitudes to carrier screening for deafness, prenatal diagnosis concerning deafness, and termination for recessive deafness. Respondents were presented with five statements and asked to respond on a five point Likert scale ranging from "definitely not" to "definitely yes". Space was provided for respondents to explain their responses in a qualitative manner.

The second section used the economic instrument of willingness to pay (WTP) to assess the value respondents placed on deafness carrier screening. This technique is based on the premise that the maximum amount of money a person is willing to pay for a commodity is an indicator of the value to them of that commodity. In the absence of a market to determine maximum WTP, such information is collected from responses to hypothetical questions.[15] Respondents were asked to imagine they lived in a country, such as America, where people have to pay for medical tests. They were informed that the aim of the question was to place a value on deafness carrier screening. They were then presented with a payment card with the following ranges: £0, £1, £2, £4, £6, £8, £10, £12, £16, £20, £30, £50, £75, £100, and £100+. These ranges were initially based on a previous WTP study looking at the value of carrier screening for cystic fibrosis. The pilot study indicated that this range was broad enough, with no respondents in the pilot study ticking the £100+. Respondents were asked to tick "Yes" for amounts they were sure they would be willing to pay for a deafness carrier test, to tick "No" for amounts they were sure they would not be willing to pay, and to circle the maximum amount they would be willing to pay. Respondents who stated they would be willing to pay more than £100+ were asked their maximum willingness to pay. Following this, respondents were presented with space to state the reason for their willingness to pay response. Finally respondents who answered zero were asked why they did so, with possible responses being "do not value a genetic screening test", "cannot afford to pay", and "object to paying". The latter were interpreted as protest responses and removed from the data.

The final section collected information on respondents' hearing status, experience of deafness, general attitudes towards serum screening, perceptions of own risk of having a deaf child, and socioeconomic characteristics.

Information collected in sections 1 and 3 of the questionnaire were assessed in terms of their relationship with WTP. A number of a priori hypotheses were identified.

- Respondents more in favour of carrier screening and prenatal diagnosis (as indicated by responses to statements and questions concerning general attitudes towards serum screening) will value carrier screening more highly, and hence have a higher WTP.
- Respondents with knowledge of deafness (including own hearing status, member of family being born deaf, knowledge of anyone born deaf) will differ in their values from those with no knowledge
- Respondents who perceive themselves to have a high chance of having a deaf child will value carrier screening more highly.
- Higher income respondents will have a higher WTP.
- No a priori hypotheses were made concerning age. Where sample size permitted, tests (*t* test, F test of linearity, chi-squared (c2), and Kruskal-Wallis (Z)) were used to see if mean willingness to pay was related to these variables.

RESULTS

Of the 135 women approached, 91 women agreed to complete the questionnaire in the clinic and 37 were given a questionnaire to return by post. Eighty-eight women successfully completed the questionnaire at their appointment and 16 women returned the questionnaire by post. The characteristics of respondents are shown in table 1. The majority had normal hearing, were not aware of any member of their family being born deaf, and perceived their own risk of having a deaf child as being "low" or "very low". One quarter of respondents knew someone who was deaf.

The results from the attitude statements are shown in table 2. Seventy percent of respondents would like to know if they were a carrier of a deafness gene. The most common explanation for such responses was the value of information, whereas those against stated that deafness was not a major disability, and expressed concern regarding "society's desire for the perfect child". Seventy-four percent would have prenatal tests to see if their child was deaf. Reasons for and against prenatal diagnosis were similar to those for carrier screening. Only 7% of respondents would have a termination if the test indicated the baby would be deaf, supporting the hypothesis of value in information.

The positive attitudes towards deafness carrier screening were supported by the WTP responses. Ninety-three percent of respondents (n=97) completed this question. Three of these were protest bids, leaving 94 valid responses. Mean WTP was £42, with a median and mode of £20, and a range from £0 to £250. Mean willingness to pay was significantly related to perceptions of chance of having a deaf child, intention to have other screening tests, low maternal age (table 1), and "pro-screening" attitudes (table 2).

Table 1 Characteristics of respondents and willingness to pay (WTP) for carrier screening for deafness genes

Variable	Values	% (numbers)	Mean WTP (£), (numbers)	Test
Knowledge of deafness				
Hearing status	Normal hearing	94.2 (98)	43 (90)	Insufficient variation
	Mild hearing loss	1.0 (1)	20 (1)	
	Moderate hearing loss	0	–	
	Deaf	0	–	
	Other (partial deafness, no hearing aid)	1.9 (2)	10 (2)	
Are you aware of any member of your family being born deaf?	No	92.3 (96)	43 (88)	Insufficient variation
	Yes	3.8 (4)	28 (4)	
	Don't know	1 (1)	30 (1)	
Do you know anyone who was born deaf?	No	72.1 (75)	45 (72)	t=1.54, p=0.130
	Yes	25.0 (26)	31 (21)	
Perceptions of chance of having a deaf child				
What do you think is your possibility of having a deaf child?	No possibility	4.8 (5)	52 (5)	F=72.3*, p=0.001
	Very low possibility	30.8 (32)	32 (32)	
	Low possibility	44.2 (46)	44 (41)	
	Medium possibility	10.6 (11)	48 (9)	
	High possibility	0 (0)	–	
	Very high possibility	0 (0)	–	
Attitudes towards serum screening				
Do you think that you will have the 16 week blood test for spina bifida/Down syndrome in this pregnancy?	No	14.4 (15)	28 (11)	F=67.44, p=0.001
	Don't know	20.2 (21)	38 (21)	
	Yes	60.6 (63)	47 (59)	
Socioeconomic characteristics				
Age (grouped responses)	20 and under	9.6 (10)	56 (10)	F=51.68, p=0.001
	21–25	24 (25)	47 (22)	
	26–30	42.3 (44)	39 (42)	
	31–35	20.2 (21)	37 (17)	
	36 and over	2.9 (3)	23 (3)	
Household income (before tax)	Less than £6000	8.7 (9)	37 (9)	χ^2=5.88, p=0.318
	£6001–£10 000	5.8 (6)	28 (6)	
	£10 001–£15 000	9.6 (10)	60 (10)	
	£15 001–£20 000	15.4 (16)	32 (14)	
	£20 001–£25 000	11.5 (12)	24 (10)	
	More than £25 001	45.2 (47)	48 (42)	

*For this test "no possibility" and "very low possibility" were grouped together, giving a mean willingness to pay of £35.00.

DISCUSSION

As the human genome project leads to the identification of increasing numbers of disease causing genes, and high throughput genotyping technology develops, the range of conditions amenable to carrier screening will increase. As indicated by this study, this will raise ethical and economic challenges (to name but a few). This study indicates a positive attitude of non-deaf pregnant women towards prenatal deafness screening. While this is the first study assessing non-deaf pregnant women's attitudes to genetic testing for deafness, given findings from elsewhere, it is likely that opposite results would have been found if this study had been carried out on the Deaf community (pregnant or not), but similar results would have been found for hearing parents of deaf children (pregnant or not).[11–14] This difference in viewpoint of different groups in society highlights important ethical issues that will be raised within the health service as decisions are made concerning the implementation of genetic developments. Within the context of this study, such ethical issues relate to questions such as "is deafness a sufficiently severe disability that warrants carrier testing" and "if it is acceptable for the hearing to undergo prenatal testing for deafness, can the Deaf community use testing to ensure a deaf child"?

From an economic perspective, the study indicates important points regarding the value of prenatal screening. While the majority of respondents to the questionnaire wanted, and valued, deafness genetic carrier screening, and would go on to have prenatal diagnosis if they were found to be carriers, they would not seek termination of an affected pregnancy. Qualitative responses indicated that women value screening for the information provided and not the opportunity to terminate the pregnancy. This result is consistent with other studies.[16–18] and supports the findings of Jones[19] concerning the value of genetic testing generally, recently reported in the *BMJ*. Such findings have important implications for future economic evaluations of genetic deafness screening programmes and screening programmes more generally. Many studies evaluating such programmes adopt either a resource savings approach[16] or a cost effectiveness approach,[20 21] thus assuming that the only benefit is the detection and termination of an abnormal fetus (that is, benefits are defined in medical terms). Future evaluations of screening programmes, and indeed any proposed health care interventions based on genetic testing, should take a cost-benefit approach (where costs and benefits are measured in the same terms, usually money).[20 21] This is the only way for all potential (dis)benefits to be considered, including the value of information, better preparation for the delivery of a deaf child, and any improved functional outcome and quality of life for the child resulting from earlier detection and management. (These latter benefits would be derived from neonatal hearing screening for all newborns.) Consideration should also be given to any implications of prenatal diagnosis for the delivery of postnatal care.

Mean WTP for carrier screening for deafness genes for non-pregnant women was estimated to be £42. From an economic perspective, questions regarding whether carrier screening

Table 2 Pregnant women's attitudes and willingness to pay (WTP) for carrier screening for deafness genes

Statement	Definitely not % (number) WTP (£) (number)	Probably not % (number) WTP (£) (number)	Don't know % (number) WTP (£) (number)	Probably yes % (number) WTP (£) (number)	Definitely yes % (number) WTP (£) (number)	Test
If the test was available, do you think you would like to know if you were a carrier of a deafness gene?	2.9 (3)	11.5 (12)	15.4 (16)	46.2 (48)	24.0 (25)	
Mean WTP	0.00 (2)	28.00 (10)	33.00 (14)	38.00 (44)	63.00 (24)	Z=5.07, p=0.027
If you and your partner did have tests, and were both found to be carriers for a deafness gene, do you think that you would ask for tests to find out if the baby would be born deaf?	3.8 (4)	7.7 (8)	14.4 (15)	43.3 (45)	30.8 (32)	
Mean WTP	0.00 (3)	10.00 (6)	23.00 (14)	37.00 (40)	66.00 (31)	Z=17.36, p=0.001
If in pregnancy your genetic tests indicated that the baby would be deaf, do you think that you would have a termination?	47.1 (49)	26.0 (27)	20.2 (21)	0.8 (6)	1.0 (1)	
Mean WTP	26.00 (42)	50.00 (25)	67.00 (20)	33.00 (6)	20.00 (1)	Z=4.75, p=0.032
If you and your partner were both found to be carriers, do you think that this knowledge would affect your decision to have more children in the future?	23.1 (24)	29.8 (31)	28.8 (30)	14.4 (15)	3.8 (4)	
Mean WTP	31.00 (21)	43.00 (29)	41.00 (26)	46.00 (14)	75.00 (4)	Z=2.10, p=0.151
Even if you would not want a carrier test yourself, do you think that the test should be available for other people?	0 (0)	2.9 (3)	1.0 (1)	46.2 (48)	47.1 (49)	
Mean WTP	–	15.00 (2)	10.00 (1)	25.00 (43)	60.00 (46)	Z=12.65, p=0.001

should be provided are related to (1) do benefits outweigh costs? And, if so, (2) how does the ratio of costs to benefits compare with other competing interventions where costs outweigh benefits? Future research is needed in this area.

While people who knew a deaf person were willing to pay less than those who did not, the difference was not significant. This finding is however interesting. One interpretation is that knowledge of deafness informed respondents that deafness is not a major disability. This raises questions concerning appropriate population groups in future studies eliciting the public's views on genetic developments, and whether education of the public about the quality of life of the hearing impaired might reduce the perceived value of prenatal testing.

CONCLUSION

Advances in genetic technology will make it possible to predict both disease and individual characteristics of unborn children. The extent to which such developments should take place continues to be a subject of great controversy, resulting in economic, social, and ethical dilemmas. This study found that while non-deaf pregnant women value carrier screening for deafness genes, and would undergo prenatal diagnosis, the majority would not abort an affected fetus. While more studies eliciting public views are required, the potential lack of agreement across society suggests difficulties for those striving to make genetic screening policy decisions that reflect public opinion.

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