THE EFFECTS OF GENETIC HEARING IMPAIRMENT IN THE FAMILY

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APPENDIX 15.1

QUESTIONNAIRE 2

TITLE: PSYCHOSOCIAL ASPECTS OF NEUROFIBROMATOSIS TYPE 2

Relative/Significant Other

This is a confidential questionnaire to try and find out about the effects of the diagnosis of NF2 in a family. We are trying to find out what is the most important to family members. There is no right or wrong answer, but what you feel is important to you.

1. *Please could you tell me about the ways in which your partner being diagnosed with NF2 has affected your life. Please could you put a star(*) by the ones you feel are most important.*

2. *Are there any **POSITIVE** effects that the diagnosis of NF2 in your partner has had on your life? Could you write them down.*

Thank you for completing this questionnaire. We hope it will be useful in improving the service to families with NF2.

16 Attitudes of Adults with Otosclerosis towards Issues Surrounding Genetics and the Impact of Hearing Loss

ANNA MIDDLETON, IOANNIS MOUMOULIDIS, GRAEME CROSSLAND, MALLAPPA RAGHU, PRANAY KUMAR SINGH, EVAN REID AND PATRICK AXON

INTRODUCTION

Otosclerosis is a condition that causes hearing loss. It is different from many other causes of hearing impairment in that the loss can often be corrected by surgery, resulting in a restoration of hearing. The psychological impact of this condition has largely been ignored due to the perception that the disability from the condition can be easily 'fixed' (Lemkens, 2005). However, the impact of having a hearing loss cannot be underestimated, particularly if the patient chooses not to have any intervention or if the intervention is inappropriate or unsuccessful. This chapter focuses on the attitudes of people with otosclerosis towards their hearing loss and the impact of this on their lives.

Otosclerosis is a condition that results when the stapes within the middle ear becomes fixed. Typically, the resultant hearing loss is progressive over several years leading to a mild, moderate and sometimes severe or profound loss. Approximately 70% of cases are bilateral and the hearing loss may typically start to become apparent in the late teens or early twenties, with a clinical hearing loss usually picked up in the 20s or 30s. Otosclerosis initially causes a conductive hearing loss, but the latter stages of the disease can affect the inner ear so causing a mixed loss. Surgery and hearing aids are used as effective methods to restore hearing. If the otosclerosis is very far advanced and associated with a profound hearing loss, treatment with a cochlear implant may also be appropriate (House & Cunningham, 2005).

Otosclerosis is thought to have a prevalence of 3 per 1000 in the Caucasian population (Declau et al., 2001). It is also thought, in some families, to have

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an autosomal dominant pattern of inheritance with reduced penetrance and expression, although the majority of cases of otosclerosis appear to be sporadic (Lemkens, 2005).

The study documented in this chapter considers the attitudes of a group of patients with suspected otosclerosis, who have not yet had surgery. They presented to an ENT clinic with diagnostic signs of the condition and more than half of the group had a family history of the condition in other relatives. They therefore had knowledge and experience of coping with otosclerosis and, as this had not been corrected yet by surgery, they were in a position to report the impact of this condition on their lives.

METHODS

A structured questionnaire was designed including 12 closed questions, based around work previously piloted (Middleton, 1999). The questions assessed attitudes towards genetics, having a genetic test for deafness, burden of hearing loss, success of communication and need for support. The questionnaire was offered to patients with otosclerosis attending an ENT clinic in Cambridge, UK, in 2003. Completed questionnaires were received from 205 participants in total, 62% of whom had already had successful surgery for their otosclerosis. The data presented here relates to the 71 participants who had not yet had surgery or for whom surgery was not appropriate, i.e patients currently living and coping with the condition. Socio-demographic data relating to the sample is given in Table 16.1.

Table 16.1 Socio-demographic data

Total participants with otosclerosis (n = 71)	%
Married	75%
Female	66%
Age ranges:	
20–39	28%
40–59	52%
60–79	20%
Degree of hearing loss:	
Mild (21–40 dB)	52%
Moderate (41–60 dB)	47%
Severe (61–80 dB)	1%
Has a family history of otosclerosis	53%
Social class 1–2 (manager, senior official, professional)	35%
Social class 3–7 (Assoc. professional, technical, administration, secretarial, skilled trade, sales, customer services)	41%
Social class 8–9 (Plant and machinery operatives, elementary occupations)	24%

Table 16.2 Responses to each question documenting attitudes towards issues surrounding genetics and impact of deafness

Question	% response
If there was a cure or treatment for deafness, would you want to have it?	
Yes	75%
No	1%
Not sure	24%
If you could have had a genetic test (blood test) when you were younger that would have predicted whether you were likely to develop a hearing loss when you were older, would you have wanted such a test?	
Yes	76%
No	10%
Not sure	14%
If you answered yes, do you think you would have altered your behaviour to protect against going deaf?	
Yes	51%
No	13%
Not sure	21%
Some people with no experience of deafness might assume that this is burdensome for a person who has lost their hearing. Please can you say whether you feel, in reality, an actual burden of having a hearing loss?	
My hearing loss causes no burden to me	3%
My hearing loss causes very little burden	58%
The burden is moderately great, but I can cope with it	34%
The burden is very great, but I can cope with it	3%
The burden is too great and I have difficulty coping with it	1%
I'm not sure	1%
How successfully do you and your partner/significant other communicate with each other?	
Very successfully/Successfully	89%
OK (communicate on a basic level, but are not able to talk about complex issues due to problems with communication)	4%
Poorly	1%
Do you feel you are advantaged/disadvantaged in any way because of your hearing loss?	
Advantaged	3%
Disadvantaged	54%
Neither advantaged nor disadvantaged	39%
Both advantaged and disadvantaged	4%
Some people feel they need extra emotional support with coping with their hearing loss. Do you feel you receive enough support from family, friends and hearing professionals?	
Enough support received from family and friends	76%
Not enough support received from family and friends	1%
Enough support received from health professionals	59%
Not enough support received from health professionals	1%
Would you appreciate more specific emotional support from health professionals?	
Yes	34%
No	24%
Not sure	42%

Percentages for each question may not always add up to 100% due to missing numbers

RESULTS

All sociodemographic data as well as other variables were analysed according to whether patients had a family history or not. There was *no significant difference* between the responses from participants with a family history and without a family history of otosclerosis to any of the questions so the results have been presented as one group (Table 16.2).

DISCUSSION

The majority of participants in the study were female, married and aged 40–59, had a family history of otosclerosis and had a moderate level of hearing loss. This could be considered a typical representation of people with this condition (House & Cunningham, 2005).

The vast majority of the group said that they would want a cure or treatment for their deafness, the assumption from this being that this was a condition that was irritating to live with and they would rather they did not have it. The majority of participants in the present study will be assessed at some point to see if surgery is appropriate. It could be assumed that most of them would be keen to proceed with surgery; however, 25% of the group said they did not want to be treated or were not sure if they would want a cure for their deafness. This indicates that although surgery may be offered to them, perhaps not all would wish to pursue this. It is possible that, if there were alternatives to surgery, these would be preferable. Other research which considered the impact of having surgery on people with otosclerosis has shown that there are differences in 'temperament' and 'optimism – pessimism' scale pre- and post-operatively, in that before surgery people feel more negatively about their condition than after surgery (Gildston & Gildston, 1972). This would suggest that even though there may be fear and uncertainty about the surgical procedure prior to having it, afterwards there is psychological benefit. Larger and new studies looking at the psychological benefits of having surgery for otosclerosis would be useful.

The majority (75%) of the group said that, if it had been possible to predict when they were younger whether they were likely to develop a hearing loss when they were older, they would have wanted such a test. But, only half of the group said that they would have altered their behaviour to protect against going deaf. This indicates that people like to be forewarned as to what may befall them in the future, but that this is mainly for information's sake. It may also indicate that otosclerosis is not perceived as a condition to be avoided at all costs. This attitude was reflected in other questions assessing the perceived burden of the condition. More than 60% of the group said that their hearing loss caused no or little burden to them and 89% said they managed to have very successful or successful communication with their partner or significant other.

However, sadly, one member of the group said that the burden associated with their hearing loss was too great to cope with. It is hoped that this expression of despair has been discussed with the health professionals involved and appropriate support put in place. It also suggests that the psychological impact of otosclerosis should not be underestimated for some people affected with it.

The results of this study can be compared to other research that has documented attitudes towards the same issues, but using different population groups. Deaf and hearing parents of deaf children were asked to comment on the perceived level of burden of deafness for their children (Middleton, 2005). The results from this showed that deaf parents were less likely to perceive a burden of deafness in the children than hearing parents, the assumption being that deafness is perceived as more burdensome by hearing people, who do not have personal experience of deafness in themselves. Most people participating in that research had a severe – profound level of deafness, either in themselves or in their children and so this is a somewhat different situation to the participants in the present study with otosclerosis. However, it would be interesting to ask hearing people, with no knowledge or experience of otosclerosis, for their opinions on the perceived level of burden attached to this condition. One could hypothesise, like in the research already mentioned above, that they might perceive the level of burden to be higher than reported by people actually with the condition.

The majority of participants felt that they had received enough support from family, friends and health professionals in coping with their hearing loss, but 34% said they would have appreciated more specific emotional support from health professionals. This is an interesting finding as, even though most in this study felt that the condition was not too burdensome to deal with, more than a third of them still would have appreciated more emotional support from health professionals. This may be in relation to dealing with a health professional who is empathic to their situation or else spends a little time asking about and listening to the impact of the condition.

When asked about whether participants felt they were advantaged or disadvantaged because of their hearing loss, just over half said that they felt disadvantaged. This indicates that otosclerosis is disabling to some, but not for all people who have it.

Interestingly, 3% said that they felt advantages associated with their hearing loss; all these people had a family history of the condition. This could possibly indicate that having a family history of the same condition offers support and a connection that links people. This has been reported before within numerous other contexts. For example, an anecdotal study by Lemkens (2005) indicated that those with severe otosclerosis that warranted treatment with a cochlear implant, felt that having a family history of otosclerosis was an advantage to them as they felt more prepared for their situation. They also found their problems could be easily discussed in the family and they gained support from this.

Other work done on congenital deafness has shown that having a family history of deafness can help with how people cope with this, in particular in relation to schooling (Stephens, 2005) and whether parents feel there is a burden attached to their child's deafness or not (Middleton, 2005). Many different pieces of research across the world have shown that deaf children of deaf parents are less likely to have emotional and behavioural problems than deaf children of hearing parents (e.g. Meadow, 1980; Satapathy & Singhal, 2001; Polat, 2003). Other work presented elsewhere within this book has shown that those with a family history of late onset deafness or hearing impairment (i.e. different from the congenitally deaf) also have a more positive experience of deafness than those without a family history (Kramer et al., this volume, Chapter 6). Therefore, in the present study, although there was no significant difference in the responses from participants with respect to each question and whether they had a family history or not, those who did have a family history may have been more likely to feel there were advantages of sharing the 'family condition'. It is possible that this offered a connection to their family unit – a shared experience, from which they gained psychological benefit.

In summary, otosclerosis is a condition that the participants in this study found to be bearable. The vast majority felt that the burden associated with the condition was something they could cope with and most felt that their communication with their partners was not affected by the condition. However, there was enough of a burden attached to the condition to feel that a treatment or cure was warranted and most felt that they would have liked to have been forewarned of this before they developed signs of the condition.

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There has been an explosion of studies in the field of genetic hearing impairment in the past decade, associated with major advances in our understanding of the mechanisms and conditions involved. However, a recent review has highlighted the very limited number of studies on the effects of such hearing impairment on the individuals and families of those concerned.

In *The Effects of Genetic Hearing Impairment in the Family*, under the aegis of the European Union GENDEAF programme, the editors have taken the first steps to address this deficit in our knowledge and understanding of this topic. The book addresses the problem by secondary analyses of existing large scale population studies, by prospective investigation of individuals with a family history of hearing impairment and by specific studies on patients with otosclerosis and neurofibromatosis 2 and their families.

In addition several chapters look at the specific impact of deaf culture, ethnicity and religion on reactions to deafness and the specific needs in genetic counselling.

This book represents an important first step in this field and should be an invaluable resource for all professionals involved with people with hearing impairments.

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