THE EFFECTS OF GENETIC HEARING IMPAIRMENT IN THE FAMILY

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ANNA MIDDLETON

OVERVIEW

This chapter discusses the practicalities of seeing d/Deaf[1] clients within a clinical setting in the UK. This is considered within the context of issues surrounding genetic counselling, psychology of deafness and Deaf culture.

There have been numerous excellent reviews of how to conduct an evaluation of the genetic/inherited basis of hearing loss within genetic counselling (e.g. see Israel, 1989, 1995; Arnos et al., 1991, 1992, 1996; Israel & Arnos, 1995; Gorlin et al., 1995; Mueller, 1996; Arnos & Pandya, 2003; Smith et al., 2004) and so these will be considered the background to this chapter and will not be addressed in any specific detail here.

The culturally Deaf client may have a different perspective on genetics issues and also different communication needs from those who identify with the hearing world. So there may be particular considerations pertinent to a clinical service involving such clients. Therefore, this chapter gives attention to

[1] **Terminology**
Within this chapter the terms 'deaf' and 'deafness' refer to people with an audiological loss within severe/profound levels, 'hearing loss' is an all-inclusive term that refers to any level or type of audiological hearing loss. 'Deaf' written with an uppercase 'D' refers to a deaf person who is culturally Deaf, i.e. uses sign language (e.g. British or a National Sign Language) as their first or preferred language and has a positive identity attached to being deaf. The term d/Deaf refers to deaf people who identify with both the Deaf community and the hearing world; this term is generally used in relation to adults rather than children. The term 'hearing-impaired' is avoided as many Deaf people would not view themselves as 'impaired' in any way; however, it is acknowledged that this term is widely used among health professionals as a generic term instead of 'deafness' used in this context. The deaf community is an inclusive term to refer to all people with any level and perception of hearing loss. The Deaf community or culture is a specific term that refers to culturally Deaf people only. NSL (National Sign Language) is used as a general term to refer to the main signed language from any country (e.g. British Sign Language or French Sign Language). SSSL (Signed Supported Spoken Language) refers to the sign language, which is a literal translation of spoken language for any country (e.g. Signed Supported English).

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these issues and offers information to help provide a Deaf-friendly genetic counselling service.

The author does not make the assumption that all d/Deaf people *should* participate in genetic counselling – potential clients are free to decide themselves as to whether they wish to access such services. There is also no underlying agenda to reduce deafness in society as an outcome of genetic counselling.

Firstly, an overview of the frequency of deafness is given, with reference to Deaf culture and how genetic counselling is relevant to d/Deaf people. Secondly, a historical picture is offered which gives a background to some of the attitudes of culturally Deaf people towards genetics. Thirdly, practical ideas suggest methods for effective communication with d/Deaf people in a clinic consultation. Finally, counselling issues relevant to d/Deaf people are discussed depending on the context of the family background.

The attitudes of those hearing people who have lost their hearing later on in life, due to genetic or inherited causes (the 'hard of hearing' or 'deafened'), as well as people who have specific needs due to syndromal deafness, are a different group that are not referred to specifically in any detail in this chapter, although some of the issues discussed will be relevant.

This chapter is introduced with a short account of the experience of working with Deaf people. A researcher or clinician from the genetics community may experience certain difficulties working with members of the Deaf community if this is not handled with insight and preparation. The following describes the author's initiation into such work.

INTRODUCTION

As culturally Deaf adults are often from large d/Deaf families, i.e. people with an inherited or genetic deafness, there is a huge resource here for understanding the molecular genetic basis of hearing loss as well as the psychological dynamics between members of a Deaf family (i.e. research that genetic counsellors might be interested in). However, given some of the strongly negative attitudes towards the perceived misuse of genetic technology (see later), it is unlikely that many culturally Deaf adults would seek out participation in molecular or psychological genetic research studies. Yet, when asked, d/Deaf families are often interested to know what the genetic basis is of their deafness and are also keen to be asked their views about genetics issues. Through transparent and sensitive explanation, and acknowledgement of the historical context within which the genetic services are placed, it is possible to work well as a genetic researcher in the Deaf community looking at either molecular genetic work or psychological studies.

When I originally started working in research with d/Deaf people the word 'genetic' in my job title seemed to be the codeword that closed doors to me. I was interested in documenting the views of d/Deaf and hearing parents of

deaf children towards genetic technology, and yet just asking people's opinions about genetics issues was enough for Deaf people to be suspicious of me – the assumption being that if I worked in the field of genetics then I would view d/Deaf people from the 'medical model' and would advocate the demise of the Deaf community, e.g. via genetic testing in pregnancy and selective abortion of deaf foetuses. As a practising genetic counsellor I subscribe to a non-directive model which means I work with the values and direction of the client, so the idea that I would advocate the 'demise' of any community was alien to me.

I worked hard to gain trust and offer accurate information about what modern-day genetics services offer and by doing this carefully and sensitively I was gradually able to establish myself as less of a threat. By enrolling in British Sign Language classes and through this making an attempt to be linguistically and culturally adept I tried to engage with the Deaf community by visiting Deaf clubs, support groups, charities, schools and universities as well as seeing d/Deaf people in their homes.

I approached people who could be considered 'Deaf community leaders' – key policy-makers and advocates with levels of influence in the community. These people were invited to contribute to my research and were offered an opportunity to express their views to the medical profession. They helped me with my questionnaire design and gave their approval. Without this the process would have been much more difficult, also it would have been very easy to discredit my research as the Deaf community is very small and so expressed disapproval from an influential member would have been devastating to me personally (but also practically for the study). It is imperative that different d/Deaf people are involved in the design and creative stages at the beginning as well as participating in any study so that the process is transparent and culturally sensitive. It is also advisable that researchers learn the National Sign Language (NSL) for their country so that they can respectfully initiate conversations with Deaf people, even if an interpreter is used too.

There are many academics doing interesting work within the Deaf community. Those who are d/Deaf themselves have the advantage of being able to communicate on so many different levels, both in terms of language and culture, with the Deaf study participant. I had the 'disadvantage' of being both hearing and also working in genetics (a perceived threat!) and so I had to give much consideration as to how I conducted my research.

Work with the Deaf community can be enormously rewarding and I have felt privileged to be able to meet and learn from Deaf people across the world. I am indebted to the hundreds of d/Deaf people who have taken the time and commitment to offer their opinions. This has helped me to think through how genetic counselling services could be improved and how we, as health professionals, have an obligation to do this well. The present chapter offers a brief summary of some of the knowledge and experience I have been fortunate enough to gather.

EPIDEMIOLOGY OF DEAFNESS

There are many different causes of deafness; these include environmental and genetic factors. Out of the approximately 1 in 1000 children born with a severe – profound, congenital or early-onset deafness (Davis, 1993) more than 50% have a genetic cause (Parving, 1996; Smith et al., 2004). There are over 400 genetic syndromes that involve deafness as part of the phenotype (Gorlin et al., 1995). Approximately, 30% of pre-lingual hearing loss consists of syndromal deafness, the remaining 70% consists of non-syndromal deafness (Smith et al., 2004). Most people affected by syndromal and more than half with non-syndromal deafness would be eligible for referral to genetic counselling services. Therefore, within the UK alone there are likely to be several hundred thousand people for whom genetic counselling is relevant; within the world this number could reach millions.

People with an inherited form of deafness may have numerous similarly affected relatives within their family and may use a National Sign Language (NSL) (e.g. BSL) as their preferred language. They may also choose to mix and socialise with other d/Deaf people and as such may choose to have a partner who is d/Deaf. Some researchers have suggested that approximately 90% of Deaf individuals marry another d/Deaf person (not including individuals with late onset deafness) (Schein, 1989, in Prezioso, 1995). It is thought that 70% of d/Deaf couples who have only deaf children are thought to have their deafness because of alterations in the Connexin 26 gene (Nance et al., 2000).

MEDICAL OR CULTURAL MODEL?

The 'pathological' or 'medical' model perceives deafness as a medical defect to be treated, corrected or cured. For example, an ENT surgeon would advocate the use of cochlear implants and an audiologist prescribes hearing aids, both taking the perspective that to be hearing, or as close to this as possible, is the preferred option for the client. However, this perspective starkly contrasts the way deafness is perceived via the 'cultural' or 'linguistic' model. Here deafness is not viewed as a disability, but rather a way of life, often identified via communication using sign language. People who consider themselves 'culturally Deaf' do not feel disabled or 'impaired' with respect to this. They feel empowered by their language, they have a positive identity attached to their deafness and they tend to mix and socialise with many other Deaf people (Padden, 1980; Arnos et al., 1991; Christiansen, 1991). Deaf identity is something that evolves over time, the process of establishing an identity is influenced by the interactions deaf people have with other deaf people and also their hearing peers (Ohna, 2004).

Although exact figures are not known it is thought that there are at least 50,000 deaf people in the UK who use British Sign Language (BSL) as their first or preferred language (RNID, 2006a), and therefore may consider themselves 'culturally Deaf'. It is likely that many of these people come from families where there are numerous relatives with an inherited deafness. There is a large and vibrant 'Deaf culture' in many countries across the world, e.g. in the UK, USA, Netherlands, Sweden, Norway, Germany, Australia etc.

Being a member of the Deaf community is not determined by audiological level of hearing loss (Woll & Ladd, 2003). Although most people will have a congenital or early onset, profound level of deafness, there are many people with this audiological assessment who would consider themselves more associated to the hearing world. Conversely there are culturally Deaf people who have a relatively mild level of hearing loss and residual hearing.

Ninety per cent of deaf children are born to hearing parents (Cohen & Gorlin, 1995). Such deaf children may not have easy access to Deaf role models if they do not automatically have similarly affected relatives. This means that they may not develop their Deaf identity until they start school and begin to mix with other d/Deaf children through groups and clubs. If they are brought up in a mainstream school and an oral environment then they may not have an affinity with the Deaf community at all, or not until adulthood. However, studies have shown that those d/Deaf people who are able to accept, mix and work with the values of both the hearing world and the Deaf community appear to have the highest levels of self-esteem (Bat-Chava, 1994, in Calderon & Greenberg, 2003). Calderon and Greenberg (2003) argue that Deaf role models are vital throughout the education of deaf children, whether they are part of a hearing or deaf education system.

NEWBORN HEARING SCREENING PROGRAMME

As the majority of deaf children are born into hearing families a diagnosis of deafness may be delayed – due to parents and health professionals neither anticipating nor specifically looking out for it. The Newborn Hearing Screening Programme offers the opportunity to screen all newborn babies audiologically for deafness and, as such, means that deafness can be diagnosed much earlier than ever before (Cone-Wesson, 2003). The hope of this is that appropriate communication and educational tools can be implemented as early as possible thereby giving the deaf child the best possible chance of 'normal' development (Sass-Lehrer & Bodner-Johnson, 2003). By delaying a diagnosis, this may delay the acquisition of effective language. The knock-on effect of this on emotional and cognitive development can be enormous.

There is discussion as to whether genetic testing, e.g. for Connexin 26, should be an automatic part of the Newborn Hearing Screening Programme, so that

both the audiological hearing loss and also the genetic cause are identified (Arnos & Pandya, 2003). There is some slight resistance to this, however, due to concern that such testing, although useful for parents to know what caused their child's deafness, may give the impression that pre-natal genetic testing for the next pregnancy should be utilised (Middleton, 2002a) and there is much resistance to the use of this from both deaf and hearing people (Middleton et al., 2001).

GENETIC COUNSELLING

Deaf individuals are often interested to know if and how they have inherited their deafness and what the chances are of passing this on to children (Arnos et al., 1992). These are questions that can be addressed by the clinical service of genetic counselling. Such a service is provided by genetic counsellors and clinical geneticists working in the Clinical Genetics department, found in most major teaching hospitals across many areas in the UK and elsewhere throughout the world.

Genetic counselling in general is 'the process by which patients or relatives at risk of a disorder that may be hereditary are [informed] of the consequences of the disorder, [and] the probability of developing or transmitting it' (Harper, 1993). Genetic counselling not only offers information about issues relating to genetics and inheritance, it also offers a supportive and non-judgemental environment, following a 'non-directive' code, where clients are neither advised nor coerced with regards to decisions.

Both geneticists and genetic counsellors undertake a genetic evaluation. It is usual for there to be overlap between the work that both these health professionals do. However, broadly speaking, one of the main differences between the roles is that any physical or diagnostic examination would be done by the doctor (geneticist) and, once a diagnosis is established, longer-term follow-up and support as well as information giving can be provided by the genetic counsellor. A medical history is taken and also a physical examination is carried out on the client to evaluate whether there could be a syndromal cause to the deafness. Medical records for relatives may also be collected for comparison and the obstetric history of the client's mother is documented. Genetic testing via a blood sample may be offered, which may confirm the clinical investigations.

Several hundred genes are known to be involved with deafness (Van Camp & Smith, 2006). Alterations in the Connexin 26 gene, are thought to account for up to 50% of genetic cases of childhood deafness, with 1 in 31 people carrying this gene in certain populations (Estivill et al., 1998; Kelley et al., 1998). Deafness resulting from Connexin 26 gene alterations is typically severe – profound and congenital (Mueller et al., 1999); however, there are also reports of people with mild – moderate loss too (Cohn et al., 1999). A result of the

molecular genetic research means that, for certain families, it is possible to define whether a specific gene alteration has caused a person's deafness and subsequently, what the chances are of passing this on to children. Such testing and information relating to this is provided within the genetic counselling service.

Some people request genetic counselling with the aim of preventing genetic disorders from being passed on in their family, others simply want information so that they are better informed of the chances of passing on a specific genetic condition. Families may be interested in finding out the medical basis to their hearing loss, just for information's sake to 'piece together the jigsaw' or because they want to make specific decisions relating to having children.

Pre-natal genetic testing for deafness is not a service that is routinely available within genetic counselling and requests for this are few and far between. Most families are just interested to know if their deafness is genetic and what the chances are (for preparation) of passing this on to children (Middleton et al., 2001). However, pre-implantation genetic diagnosis for Connexin 26 deafness has been requested, where two hearing parents wanted to avoid having deaf children (Australasian Bioethics Information, 2002; Kelly, 2002).

There are often myths surrounding why deafness is present in a family. Many people make reproductive decisions based on assumption rather than medical information. The following case studies are examples of this.

CASE STUDY 1

One deaf couple known to the author through her work as a genetic counsellor were so frightened of having deaf children that they had decided not to have children. The burden that they attached to their own deafness meant that they felt a heavy responsibility to not 'inflict' this on their children. However, through genetic testing it was revealed that their chances of having deaf children were minimal. They were delighted with this news.

CASE STUDY 2

Another Deaf couple had assumed that their deafness was not inherited because they both came from hearing families; they were then surprised when their two children were born deaf. Genetic testing revealed that they were both deaf due to an alteration in the Connexin 26 gene and as a consequence all their children would be born deaf, they were also delighted with this news. They had a strong Deaf identity and, although their hearing families hoped that deafness would not be inherited, as a couple, they were really pleased to pass on their deafness, their language and culture to their children.

Both couples welcomed the opportunity to discuss their concerns about family planning and the process of genetic counselling and testing meant that they were better informed about their genetic heritage. This in turn meant that

they were better able to psychologically engage in their future. They also had the opportunity to confidentially express the burden and responsibility they felt with respect to passing (or not) deafness on to their children. This was provided within a sensitive environment away from the perceived 'pressure' from their family and community.

The process of genetic counselling for deafness is therefore of direct relevance to the millions of d/Deaf people across the world with inherited deafness. However, generally the uptake for genetic counselling from such d/Deaf clients in the UK is very low. There are many possible reasons why this might be: d/Deaf people may just not be interested in knowing why they are deaf or what the chances of having deaf children are, although clinicians working with d/Deaf adults would indicate otherwise (Arnos et al., 1992). Other reasons may relate to fear of genetic services; this issue is addressed below.

DEAFNESS, EUGENICS, GENETICS AND ATTITUDES

Culturally Deaf people may often have quite negative attitudes towards genetic technology (Middleton, 2002b). The views of a collective group of culturally Deaf people attending a conference called the 'Deaf Nation' at the University of Central Lancashire, UK, in 1997 were studied to ascertain attitudes towards genetics (Middleton et al., 1998a, 1998b). Delegates were asked to complete a questionnaire which documented their views about genetic technology and how they felt about its use with respect to deafness (e.g. genetic testing in pregnancy for deafness). Of the 87 delegates who completed questionnaires, 55% thought that genetic testing for deafness would 'do more harm than good'; 46% thought that its potential use 'devalued d/Deaf people', and 49% were concerned about new discoveries in genetics (Middleton et al., 1998a, 1998b). This group indicated that they felt really threatened by the perceived 'misuse' of genetic technology, the biggest fear relating to pre-natal genetic testing for deafness followed by selective termination of pregnancy if the foetus was deaf. If this fear were realised then the net result of such actions could be the demise of the Deaf community.

A much larger study has since been completed (n = 1314), which replicated many of the above views. Here the attitudes of d/Deaf, hard of hearing and deafened adults as well as hearing parents of deaf children were documented (Middleton et al., 2001). This study indicated that Deaf people have quite different attitudes from those who do not identify with the Deaf culture including hard of hearing/deafened adults and hearing parents of deaf children. Those who mix more in the hearing world tend to have quite positive attitudes towards genetic technology. The majority of all participant groups indicated that not many people would actually be interested in using pre-natal genetic testing for deafness with selective termination of pregnancy involving a deaf foetus, which is a fear of the Deaf community. This work was completed in the

UK and has also been replicated in the US (Stern et al., 2002), with similar findings. Therefore, it is very unlikely at the moment that the Deaf community would diminish through the use of genetic technology. Nevertheless the perceived fear in relation to this is enormous.

HISTORICAL CONTEXT

Throughout history there have been numerous attempts to suppress and even deliberately destroy the Deaf community. Alexander Graham Bell (inventor of the telephone and leader of the eugenics movement) delivered a paper in 1883 called 'Memoir Upon the Formation of a Deaf Variety of the Human Race' to the National Academy of Sciences. In this he advocated that deaf people should marry hearing people (as opposed to other deaf people) so that they could reduce the chances of passing on deafness to their children (Bell, 1883). Despite his great respect for d/Deaf people (his own mother was deaf and so too was his wife) he took the view that deafness was a great disability and should be avoided if at all possible. Hitler during the Second World War advocated that d/Deaf children and adults should be sterilised so that they could not pass on deafness to their children; indeed 16,000–17,000 deaf people suffered sterilisation. In addition to this, other d/Deaf people were killed as part of 'Operation T4' the Nazi programme designed to destroy disabled citizens – all part of the eugenic pursuit of the perfect Aryan race (Biesold, 1999, in Schuchman, 2004).

Given the evidence above and many other attempts throughout history to prevent d/Deaf people from having children – all with the (often incorrect) assumption that deafness is always inherited, it is not surprising that d/Deaf people are often suspicious of modern-day genetics services. The very fact that pre-natal genetic testing for deafness with selective termination of pregnancy for a deaf foetus is even possible is enough for Deaf people to feel that there is another eugenic agenda being impressed upon them. There is a feeling that, historically, genetics services (and 'why should modern-day services be any different!') have devalued the role of Deaf people in society. It is therefore imperative that genetic counsellors and geneticists are mindful of the context within which they practise.

It is important to offer a 'culturally neutral' genetic counselling service (Arnos & Pandya, 2004), where Deaf clients are neither judged nor stereotyped. Assumptions should not be made about preferences for having deaf or hearing children and genetic counsellors should be aware of the historical sensitivity of such issues.

DEAF PEOPLE'S CLINICAL SERVICE REQUIREMENTS

The following sections consider the requirements of a clinical service for d/Deaf people. The UK Disability Discrimination Act (1995) gives some

guidance on specific issues to consider in relation to communication and access to services.

THE DISABILITY DISCRIMINATION ACT (DDA)

The Disability Discrimination Act (DDA) 1995 in the UK prevents d/Deaf people from being discriminated against by any service providers, including the Health Service and hospitals (RNID, 2006b). This means that the health profession needs to ensure that communication issues are addressed, for example through the installation of text-based and video-based information and telephone systems as well as providing access to qualified interpreters supporting the preferred language of deaf clients. In an ideal world all health professionals would have Deaf awareness training and those working regularly with deaf clients would be proficient in signed language and lipspeaking.

The following sections give consideration to the different forms of communication tool that d/Deaf people may use.

COMMUNICATION

Deaf and hard of hearing individuals use a variety of different forms of communication: speech, National Signed Language (NSL), Signed Supported Spoken Language (SSSL), which refers to the sign language which can be a literal translation of spoken language for any country, lip-reading, writing, reading, cued speech, use of non-verbal cues through gesturing and facial expressions. Particularly within a counselling context, effective communication does not always have to mean fluency in language – the use of non-verbal cues, facial expressions and body language all offer a form of communication that can express what a person is feeling sometimes more than a language can. NSL has its own grammatical structure and is different from SSSL which usually follows the pattern of speech.

Lip-reading

It is important to give clear lip-patterns when speaking to a d/Deaf person, without obstructing these features (e.g. by chewing gum, eating food, or covering the mouth with hair or a hand or even a beard or moustache). It is also important to maintain eye contact and not repeatedly look away, for example, at a computer or set of patient records.

Speech

Profoundly d/Deaf people may not always be able to effectively communicate using speech. Individuals from large culturally Deaf families may use very little speech, if any. This means that conversations in a clinic setting that are totally

focused around speech can be difficult. Deaf people will often have very good voice control and their speech may be quite clear. However, this can sometimes be rather misleading to the hearing person, who wrongly assumes that all they are saying is being understood. As with any conversation, where one person is communicating in a different language, it is important not to make assumptions about the level of understanding. Checking this out throughout the conversation can help. The focus of good communication not only applies to the consultation, it needs to be in place right from the moment the individual or family are referred, through to when they walk through the door of the genetics clinic, including the interaction with receptionist. The staff in the genetics clinic should know how to use IT that Deaf people use (e.g. by being familiar with TypeTalk or similar telephone relay services or having a mobile phone texting or video-phone service to inform about changes to clinic times). The receptionist needs to make sure they approach the Deaf person sitting in the waiting room to let them know visually of their consultation. Simple things like not calling out the client's name in the waiting room are easily overlooked yet so easy to put right.

Reading/writing skills

It has sometimes been the case that deaf children fall behind their hearing counterparts in reading and writing skills (Holt et al., 1992, in Ralston & Israel, 1995). Some older research has indicated that the average reading age of an 18–19-year-old deaf student fits that of an 8–9-year-old hearing student (Paul, 1998, 2003; Traxler, 2000). This may be due to the learning environment within which the deaf person was taught, or may be because English is the second language (with signed language as the preferred language thus using a different grammar and sentence construction). It is possible therefore that some deaf adults have difficulty in reading forms or questionnaires and written instruction. It is important not to assume that these difficulties are due to any problems with intellect. More recent research from Europe has suggested that deaf children who have deaf parents are more likely to have better educational achievement than deaf children with hearing parents (Kramer, 2005). The assumption here is that having a positive role model in the family who understands how to solve communication issues leads to better academic achievement. However, aside from this it is still worth making sure that any written instruction from the clinic is clear, brief (short sentences) and Deaf-friendly (by checking with someone fluent in sign language).

The genetic counselling teams can produce information in NSL for delivery via DVD and video. The information can be given in NSL and also voice-over in spoken language with subtitles (Belk & Middleton, 2004). This is a very useful tool for providing equal access to services and also complies with the Disability Discrimination Act (1995).

Communication over the telephone

Deaf people often have high levels of technological literacy. This may involve the routine use of the computer, text messaging and videophones, as well as more traditional text telephones (see Harkins & Bakke, 2003, for an overview). This technology can be incorporated into clinical practice.

Relay telephone systems also exist in the UK with information relayed to the deaf person via their text telephone through an operator.

Communication in a clinic setting

When choosing a sign language interpreter it is important to first check what sort of language is to be used. Interpretation of NSL is different from SSSL, which is different again from lipspeaking. It is also important to double-check whether the client would rather bring their own interpreter. As the local Deaf community may be small, confidentiality may be difficult to maintain and so clients may prefer to choose someone they know already. Alternatively they may prefer to use someone completely unconnected and not part of their local community (hearing interpreters are often involved in the Deaf community, and may be hearing children of Deaf parents themselves).

Whatever the situation, it is important to check whether the interpreter has interpreted genetic or even medical consultations before. If not, then it would be important to speak or meet with them beforehand to check their understanding and discuss ways that they intend to use when interpreting terms that they may not have encountered before. It is not sufficient to assume that the medical consultation will be interpreted word for word or even concept for concept with the inflection and tone of speech. There will almost certainly be differences, which unless specifically asked about, and checked, the hearing clinician will be unaware of.

Most hospitals in the UK use an agency of registered interpreters, or alternatively local freelance interpreters (agencies charge a booking fee, all interpreters charge travel costs and a minimum call-out charge on top of their fee). Interpreting is demanding and breaks are needed every half-an-hour or so (RNID, 2006d). Although ideally two interpreters should be booked, if a whole afternoon of interpreting is needed, this is not always possible. It is useful to discuss with the interpreter and the Deaf client the seating arrangements and the lighting before the consultation. It is important to talk directly to the Deaf person and maintain eye contact with them at all times. It is important not to ask the interpreter for opinions as they are meant to be neutral rather than an advocate for the Deaf person. Afterwards, as part of the feedback process, check with the Deaf person as to whether the interpreting arrangements were satisfactory (BDA fact sheet, 2005b)

Interpreters take recognised qualifications after many years of approved training (RNID, 2006d). They are highly qualified professionals and will often

specialise in specific types of work, e.g. medical, theatre, law courts etc. It is important to use someone who is registered through a national agency or has an accreditation for interpreters in the chosen NSL. There is usually a directory of qualified interpreters in each country.

Lipspeakers

'Lipspeakers' are interpreters who help d/Deaf people use speech and lipreading. The lipspeaker sits next to the hearing person who is speaking, they repeat what is being said (without using their voice) using clear lip patterns that the lip-reader may find easier to follow. They can also use fingerspelling, gesture and facial expression as well as other cues that show the phrasing and emphasis of the spoken work. Normal speech uses up to 200 words a minute. It may be very difficult for a person lip-reading to compute this many words, so a lipspeaker can use less words without losing the intended meaning. Lipspeaking is skilful and involves detailed training (RNID, 2006e).

Electronic notetakers

There are different forms of note taking, all very similar. An electronic notetaker uses a laptop to type up a summary of spoken language, not every word is typed, the notetaker summarises what is being said. The d/Deaf person could network their computer to the notetaker's so that they can also communicate to each other. As a notetaker is summarising the spoken conversation the written interpretation is delayed and does not happen in real time.

Speech-to-text (STT) reporters use a specially designed keyboard that enables every spoken word to be phonetically transcribed by a software programme into text. This makes it quicker and easier to keep up with the pace of spoken language and requires the d/Deaf person to be able to read at high speed. STT reporters use Palantype® or Stenograph® in the UK (RNID, 2006f, 2006g).

GENETIC COUNSELLING CONSULTATIONS

Timing of consultations

Most genetic counselling consultations in the UK last between 45 minutes and an hour. As there is often much technical and clinical information to explain as well as emotional issues to address, it is usual for a post-clinic letter or leaflet to be sent afterwards that summarises the consultation. However, as mentioned above, if reading and writing skills are different from those of hearing counterparts, then this method for summarising information may not be very helpful. In addition to this, within the clinic consultation if memory-processing skills are being employed in the interpretation of language, then these will

not immediately be so readily available to reinforce the technical information. Therefore, in these sorts of consultations, it is important to keep them shorter than normal and more frequent. So, instead of having a 1-hour consultation, it might be more useful to have two half-hour sessions instead. It would also be helpful to revisit the same concepts several times and rephrase them in different ways to help them embed in the d/Deaf client's memory.

Use of language

If a genetics professional is aware that a d/Deaf client does not view having a deaf child as a problem, then it would be insensitive to talk to them in terms of there being a 'risk' of having a deaf child or else referring to deafness as 'abnormal' and hearing as 'normal' within the genetic counselling process. Instead the geneticist or genetic counsellor would talk about the 'chance' of having a deaf child and use the terms 'deafness' and 'hearing' as they are without saying either is 'abnormal'. In addition to this, terms like 'mutation' and 'gene fault' also have negative connotations attached to them and so could be replaced with gene 'alteration' or 'change' instead.

Taking a pedigree

In order to make a genetic evaluation for a d/Deaf client the first piece of information collected is the family tree or pedigree; this should cover at least three generations. The hearing status and health of each individual is documented. For clients who are not aware of the details of their relatives, genetic evaluation is still possible as other data is collected too. The ethnic background of the family is relevant and so too is whether there is consanguinity (cousin or intermarriage in the family).

The experience of delayed or difficult communication between a deaf child and his/her hearing parents may lead to a feeling of exclusion in the home, a consequence of this is that there could be less knowledge about the family history (Israel & Arnos, 1995). Therefore, d/Deaf adults from hearing families may have less information about family relations to offer within pedigree taking than one might expect. It is not unusual for the genetic counsellor to be asked to telephone the hearing family on behalf of the d/Deaf client, to find out details for the pedigree. But d/Deaf adults from deaf families, who have grown up with a closeness to their relatives via a shared language, are more likely to have easier access to personal information about their family pedigree.

COUNSELLING ISSUES

Hearing children learn to express their emotions through voice and language; they are also taught to label their feelings via spoken interaction with their

parents. However, sometimes deaf children born to a hearing family may have a delay in acquiring their communication skills and therefore may have a delay in emotional and cognitive processes (Henderson & Hendershott, 1991, in Ralston & Israel, 1995). This means that as d/Deaf adults they may find it more difficult to express and describe emotions (although it is important not to overgeneralise this issue). Describing and expressing emotion can be a part of the genetic counselling consultation and so it is important to be aware that d/Deaf adults may do this differently from hearing adults.

There is much written about the social and emotional development of deaf children (e.g. Greenberg & Kusche, 1989, in Calderon & Greenberg 2003). Calderon and Greenberg (2003) summarise some of this work: 'deaf children are often delayed in language development, tend to show . . . poorer emotional regulation, and often have an impoverished vocabulary or emotion language'. Not everyone agrees with this negative labelling and can provide many examples of positive emotional expression amongst deaf children. The literature on the emotional development of d/Deaf people is somewhat controversial and there is much research to demonstrate that d/Deaf adults are resilient and able to overcome negative influences – the deafness may not impact negatively if the family environment is supportive, if the parents adapt and cope with the deafness and if there are adequate community and education resources available (Calderon, 2000; Stinson & Foster, 2000, in Calderon & Greenberg, 2003).

With regards to the emotional engagement within a clinical setting, d/Deaf adults may have a different emotional language and expression from hearing clients, depending on their life experience. However, this difference should not be viewed as deficient in any way.

Genetic counselling for deafness is of relevance to all sorts of people with differing backgrounds, many of whom will have different perspectives and experiences of deafness. The following groups and the specific nuances relating to each group are all considered in turn in the following sections.

Hearing parents of deaf children

The birth of a deaf child to hearing parents with no experience or understanding of deafness can be perceived as devastating to the parents and their extended family (Luterman & Ross, 1991, in Israel, 1995). There are many factors that may influence the grieving process as parents try to make sense of their situation. Eventual acceptance of the child as deaf may be influenced by these factors: prior perceptions of deafness, expectations and attitudes of friends and relatives, economic issues, stress factors in the family, previous coping strategies and relationships with health professionals and education network (Calderon & Greenberg, 1993, in Israel, 1995).

Hearing parents of deaf children are often very keen to understand what has caused their child's deafness, they may blame themselves and look to the

pregnancy to see what could have gone wrong. Some research has suggested that the unexpected birth of a deaf child may cause parents to feel they are being punished in some way (Vernon & Andrews, 1990, in Israel, 1995). It is therefore very important that accurate and sensitive information is given about the causes of deafness, and this can be done via genetic counselling: 'parents must know, when possible, the cause of the child's deafness to realistically face issues about which they would otherwise fantasize' (Mindel & Feldman, 1987, in Israel, 1995).

With respect to genetics issues, hearing parents of deaf children generally have positive views. In a study looking at the attitudes of a group of 527 hearing adults with a family history of deafness (most of whom were parents of deaf children), the majority chose positive as opposed to neutral or negative words to describe their feelings about new discoveries in genetics, the most frequently chosen word was 'hopeful' (Middleton et al., 2001).

Four hundred and thirty-two parents of deaf children were asked specific questions about their family and children and attitudes towards testing in pregnancy for deafness; 69% said they preferred to have hearing children (as opposed to not minding the hearing status of future children); 53% said they would be interested to find out whether a baby is deaf or hearing before it was born (i.e. have a pre-natal genetic test); most of these said they would just want this information for preparation purposes rather than so that they could have an abortion if the foetus was deaf; however, 16% said they would consider this. The majority (67%) felt their deaf children were disadvantaged because of their hearing loss (which was not the case for many d/Deaf parents), and most felt there was some to great 'burden' for them attached to having a child who is deaf. More than 80% said, if it were possible, they would want a cure for their child's deafness. When asked about support at the time of the deafness diagnosis more than half the group (52%) said they felt they did not receive enough support from the health professionals. However, most said they received the required support from family and friends (Middleton, 2005).

Therefore, for this group of clients, attending a genetic counselling consultation, there tends to be quite a lot of interest as to why the deafness is present which is coupled with negative emotions surrounding the deafness. This group is most commonly referred for genetic counselling.

Deaf adults with hearing parents

The experience of growing up in a hearing family may be daunting for deaf children if the parents and extended family are unsure how to cope and adapt to the specific needs associated with deafness. If parents struggle to communicate with their child and the child never really feels understood by their parents then this can lead to a very difficult experience that could conceivably impact on the d/Deaf person as an adult. This could also mould their own

attitudes towards having deaf children. However, if hearing parents make every attempt to establish communication channels (e.g. by learning sign language or helping children to lip-read and encourage their speech) and so too does the extended family, then this will help in all aspects of the child's development.

The perceived success of communication between parents and their deaf children has been documented. A study of 108 deaf/hard of hearing parents of deaf children reported that 67% felt that they communicated 'very successfully' with their deaf children, whereas only 33% of the 432 hearing parents of deaf children felt this was true. The vast majority of hearing parents felt the communication with their deaf child was less than perfect. Indeed 18% of this latter group said they felt communication issues were only OK or even poor (Middleton, 2005). Deaf adults who have hearing parents may feel an emotional distance between themselves and their parents, particularly if the hearing parents struggled to communicate with them when they were children.

Given the issues documented in the previous section about hearing parents' attitudes towards the impact of deafness on their children, it is easy to see how deaf children may develop low self-esteem as they grow into d/Deaf adults. Deaf and hearing researchers have suggested this can be overcome by developing positive interactions with deaf and hearing peers at school (Antia & Kriemeyer, 2003) and also through the provision of specific education systems and the incorporation of Deaf role models (Calderon & Greenberg, 2003).

Some deaf parents have said that they would choose not to have deaf children if it could be avoided (Middleton, 2005). One participant in this research said they '*would not wish deafness on [their] worst enemy*'. This highlighted the negative personal experience they had while growing up with a hearing loss and struggle they had within a mainstream hearing society. But other Deaf parents of deaf children felt the experience was positive – they were lucky to have the opportunity to pass on their language, history and culture, as well as deafness, to their children and they were proud of this (Middleton, 2005).

Deaf parents of deaf children

Ten per cent of d/Deaf couples have deaf children (Cohen & Gorlin, 1995). The process of genetic counselling for deafness can be complicated as Deaf people often marry and have children with other Deaf people. As there are so many different genetic causes behind deafness, two people within a couple (particularly when there have been multiple d/Deaf relationships within the same family) may have complex and multiple genetic predispositions. This means the calculation of 'genetic risk' and the inheritance pattern may not be straightforward. A study of the frequency of Connexin 26 gene changes showed that families where there was deafness in both the parents

and their children (n = 43), 42% had this due to Connexin 26 (Pandya et al., 2003).

The birth of a deaf child to d/Deaf parents may not be a total surprise but may still elicit a mixed response. Much depends on the d/Deaf parents' own values and beliefs about their deafness and the place of deafness in society.

Deaf parents of deaf children are much more likely than hearing parents of deaf children to feel that their deaf children do not place a burden on the family (Middleton, 2005). They are also more likely to feel that the deafness in their children is more of an advantage than disadvantage: one deaf parent (who did not identity with the Deaf community) said she felt an advantage in having deaf children as '*I could share my skills and knowledge of deafness. I could understand her needs better*'. Another deaf parent of deaf children said: '*being deaf myself, the children were advantaged as I knew what the problems were and knew what to do*'. Finally, one culturally Deaf parent of deaf children said: '*at home we're all deaf so [the children] never felt left out. It's society without "deaf awareness" that made them feel disadvantaged! Otherwise we are all happy and [a] close-knit family with [the] same rich language [and] culture*' (Middleton, 2005).

Hearing children and adults with deaf parents

Approximately 90% of d/Deaf couples have hearing children (Israel, 1995). The birth of a hearing child to d/Deaf parents can often lead to a feeling of confusion (Hoffmeister, 1985, in Israel, 1995). In a family that only uses sign language it is much easier for a hearing child to learn sign language first and spoken language second. Research has shown that normal speech and language can develop in a hearing child from a d/Deaf family if that child has contact with hearing speakers approximately 5–10 hours per week (Schiff-Myers, 1988, in Israel, 1995).

A hearing child born to d/Deaf parents may be used by their d/Deaf parents as the link between the Deaf and hearing world. Hearing children may be used as interpreters for their Deaf parents and this may be inappropriate as well as appropriate in different situations. Hearing children within d/Deaf families may be perceived as having the 'best of both worlds' – they can participate in the Deaf culture with their family, but also have access to the hearing world too. However, in order to develop a 'healthy psychosocial perspective' hearing children/adults of d/Deaf parents need to maintain a balance in the relationship between these cultures (Myers & Marcus, 1993, in Israel, 1995).

ETHICAL CONSIDERATIONS: CHOOSING TO HAVE DEAF CHILDREN

For culturally Deaf families, where there are many relatives in the family who are d/Deaf, there may be a preference for having deaf children. This concept

is not new and has been well documented in the past (Hoffmeister, 1985; Dolnick, 1993; Erting, 1994; Israel, 1995; Middleton et al., 1998a). Research from the author has indicated that a very small number of d/Deaf people may consider the application of pre-natal genetic testing for deafness with selective termination of pregnancy if the foetus was likely to be hearing. One participant in this study indicated that she wanted to avoid having hearing children as she worried they would not learn speech and be taken away from her by social services (UK) (Middleton et al., 2001; Middleton, 2004).

Deaf adults may be interested to use genetic counselling so that they can find out their genetic heritage and use this to choose a suitable d/Deaf partner with whom they can have deaf children. At Gallaudet University, Washington, DC, the author met many d/Deaf students who were interested in the process of genetic counselling. One student said that she knew her deafness was due to having two gene alterations in the Connexin 26 gene; she said she would be interested to know if any future partners also had their deafness due to Connexin 26 as she wanted to ensure that her children would be deaf.

At the time of publication no readily available, published medical evidence indicates whether any d/Deaf parents have chosen to actually use pre-natal genetic testing with selective termination of pregnancy for a hearing foetus. However, there are unsubstantiated suggestions within the genetics field to suggest that this may have been done. Given the worldwide negative press that Deaf people have received in relation to this issue, it is not surprising that neither d/Deaf parents nor the genetics professionals seeing them would advertise such an issue openly.

In 2002 a Deaf, lesbian couple from the US decided that they wanted to have another deaf child. Their deliberate choice to have a deaf child caused great debate across the world (e.g. Anstey, 2002; Fletcher, 2002; Levy, 2002; McLellan, 2002; Savulescu, 2002; Spriggs, 2002). The following are some comments from these articles:

> Couples who select disabled rather than non-disabled offspring should be allowed to make those choices, even though they may be having a child with worse life prospects. (Savulescu, 2002)

> Deaf people are behaving like hearing people. They feel good about themselves and want to have babies like them. Why should they be morally blamed? (Fletcher, 2002)

> Cultures are simply the kind of things to which we are born, and therefore to which the children of deaf parents, hearing or deaf, normally belong. Thus these parents are making a mistake in choosing deafness for their children. Given their own experience of isolation as children, however, it is a mistake which is understandable, and our reaction to them ought to be compassion, not condemnation. (Levy, 2002)

VIEWS OF GENETICISTS AND GENETIC COUNSELLORS

It is not clear to what extent parents should be allowed to externally control the genetic makeup of their own children (American Medical Association, 1994). Within genetic counselling practice it is considered best practice to offer a 'non-directive' service where clients are not told what to do nor directed to make certain decisions. Therefore, it should be possible for a d/Deaf couple to have a pre-natal genetic test with selective termination of pregnancy for the absence of the gene faults for deafness (i.e. if the foetus is likely to be hearing). Offering preimplantation genetic diagnosis with active selection for embryos that have the gene faults for deafness, could also be possible. However, it is debatable whether hearing geneticists and genetic counsellors would feel comfortable with such a use of genetic technology.

Wertz and Fletcher (1999) asked genetics professionals across the world to comment on whether they would offer pre-natal genetic diagnosis to a d/Deaf couple wanting to have deaf children. Of those who said they would offer pre-natal genetic diagnosis with selective termination of pregnancy, 43% were from Cuba, 35% were from the US, 18% were from Canada, 9% were from the UK and 0% were from Norway. Those who were in favour of this used the 'autonomy' argument – i.e. if this is what the parent chose, and they were able to make a fully informed autonomous decision, then this was acceptable to the genetics professional.

THE HUMAN RIGHTS ACT

The Human Rights Act 1998 brings the European Convention on Human Rights into UK law (RNID, 2006c). This is particularly relevant to d/Deaf parents. The Act protects the rights of d/Deaf parents not to be discriminated against. For example, a d/Deaf couple should not be told they couldn't have children because they might pass deafness on. They must also not be told to end a pregnancy if there is a chance their baby might be deaf. Deaf couples also obviously have a right to fertility treatment. In terms of whether they could use the Act to gain support for actively creating a deaf child, via implementation of genetic technology, it is not clear whether this would be covered.

THE BRITISH DEAF ASSOCIATION POLICY ON GENETICS

The 'Sign Community' or British Deaf Association (BDA) is 'the UK's largest national organisation run by Deaf people for Deaf people' (SignCommunity website) It does stress concern over the use of pre-natal genetic testing with the selective termination of 'deaf' pregnancies and *demands* that: 'all genetic counsellors should receive Deaf awareness training to ensure a clear understanding of the Deaf community and Deaf culture . . . [and that] . . . parents

are not formally or informally pressured to take pre-natal tests or to undergo termination where it is discovered that the foetus is deaf' (BDA, 2005a).

Therefore, the BDA believe that d/Deaf and hearing parents attending a genetic counselling consultation in the UK currently do not receive adequate information to enable them to make informed decisions about deafness and intend to rectify this by implementing more Deaf awareness training among genetics professionals.

THE UK NATIONAL DEAF CHILDREN'S SOCIETY POLICY ON GENETICS

The National Deaf Children's Society also has a Policy on Genetics. In it they advocate choice and information:

> The Society . . . recognises the rights of potential parents from families who have a history of deafness to take advantage of genetic testing and ante-natal diagnosis and to use the results of such tests in a way that suits the individual family. If asked for advice, the Society will ensure that the family receives positive information about deafness in order to enable them to make an informed choice. (NDCS, 2005)

To date there is no consensus across the world on whether deliberately choosing to have deaf children should be endorsed by medical science. It is of interest and useful to know that Deaf parents may prefer to have Deaf children so that, within a clinical setting, there is awareness of and preparation for such attitudes. However, it is not useful to focus entirely on this view; only a very small number of Deaf people may ever consider this option.

CONCLUSIONS

Genetic counselling services for d/Deaf people and their relatives require a specialist knowledge of deafness, Deaf culture and the role that genetics has played within history for d/Deaf people. It is imperative that communication and language differences are embraced as well as attitudinal differences. Training in Deaf Awareness would be valuable for any health professional wanting to start working in this area.

Deaf people and their families are often very interested in the services offered by genetic counselling. With prior consideration of the nuances specific to the Deaf culture it is possible for genetics professionals to offer a culturally sensitive service.

Working with deaf people who use sign language as their first language is both interesting and rewarding. All health professionals who engage in this work enjoy learning from their clients. Hopefully this chapter has offered some

ideas for health professionals thinking of entering this field as well as providing an overview for existing practitioners.

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THE EFFECTS OF GENETIC HEARING IMPAIRMENT IN THE FAMILY

EDITED BY DAFYDD STEPHENS & LESLEY JONES

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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