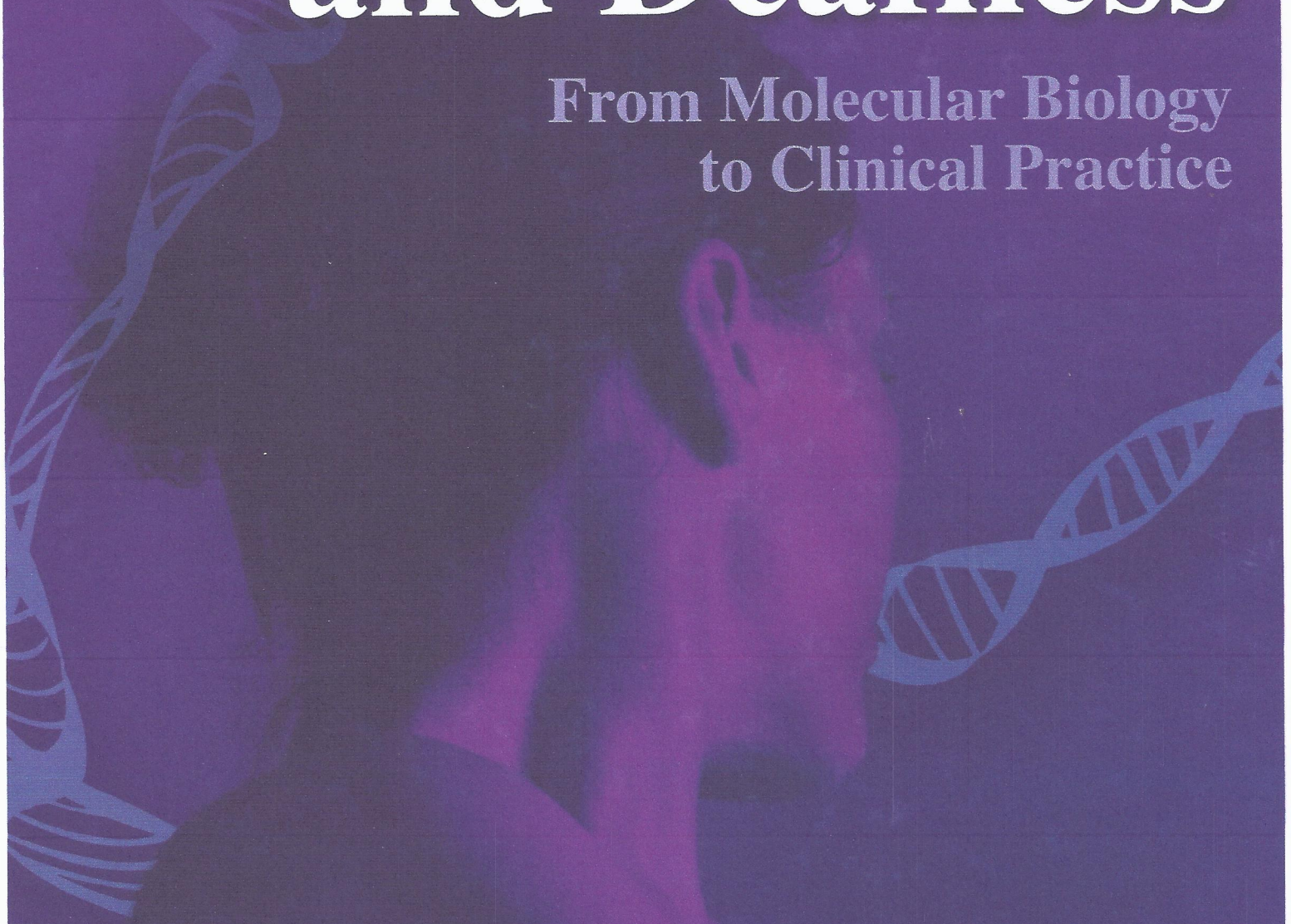
Genes, Hearing, and Deafness

From Molecular Biology to Clinical Practice



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11 Attitudes of deaf people and their families towards issues surrounding genetics

Anna Middleton

Introduction

Genetic health services in general could be improved with more insight into the particular concerns and fears of patients with different genetic conditions. Previous research has documented the lay understanding of genetics (1,2) and has looked at case-study discussion of the experience of living with a genetic disorder (3). However, more research is needed to fully explore the experience and specific demands that deaf patients and their families have with respect to genetic issues.

This chapter provides an overview of some of the research that has been done to investigate the attitude of deaf people and their families towards genetics and genetic testing. Before this is covered, it is introduced with an overview of the different perspectives of deafness. This is followed by more practical sections on genetic testing services and what happens within genetic counselling. Then, a brief summary is given on the historical context to issues surrounding genetics, eugenics, and deaf people.

Perspectives of deafness

Deafness can develop at any stage of life, the clinical consequences may vary, and this may impact in different ways on the individual's daily functioning. Deaf people may have to alter their use of language and communication in order to function effectively in the hearing world. For a mildly deaf person, this could be through the use of speech with additional lipreading or for a profoundly deaf person this could be through the use of a sign language or its derivatives.

The "pathological" or "medical" model views deafness as a medical defect, which needs treatment or correction. For example, a cochlear implant or hearing aid aims to restore hearing as much as possible, with the view that to be hearing is the preferred option for the patient. However, this perspective is in stark contrast to the way deafness is viewed as part of the "cultural" model. Within this, deafness is not a disability, but rather an experience that is just different, and certainly not defective. Here, the main form of communication is often sign language. People who consider themselves "culturally Deaf" (written with an upper case D) will often not perceive that they have a disability or impairment. They feel positive and empowered by their language and have a strong Deaf identity (4). They also tend to mix and socialize with many other Deaf people (5,6). Deaf identity evolves over time, the process is influenced by the interactions deaf people have with other deaf and hearing peers (7).

Within the United Kingdom, it is thought that there are at least 50,000 deaf people who use British Sign Language (BSL) as their first or preferred language (8), such people may consider themselves culturally Deaf. These people may come from families where there are several relatives who are deaf. Such a "Deaf culture" exists in many countries across the World, e.g., in the United Kingdom, United States, Netherlands, Sweden, Norway, Germany, and Australia.

When there are numerous similarly affected relatives with profound deafness in the same family, there is often a shared use of sign language (e.g., BSL in the United Kingdom). Such individuals may also choose to mix, socialize, and work with other Deaf people and may also choose to have a partner who is Deaf. Approximately 90% of Deaf individuals are thought to marry another Deaf person (not including individuals with late onset deafness) (9).

The audiological level of deafness is not always a direct determinant of membership of the Deaf community (10). Although most people have a congenital or early onset, profound level of deafness, there are many people with this level of deafness who associate themselves more with the hearing world. Conversely, there are people with a mild level of deafness and residual hearing who consider themselves part of the Deaf community.

When a baby is born to deaf parents, there may be an anticipation that it will have inherited its parents' hearing loss. The reaction to this may be mixed. Much depends on the d/Deaf parent's own values and beliefs about their deafness and their experience of being deaf within the wider mainstream society.

Research by the author looking at these issues has shown that deaf parents are much more likely than hearing parents to feel that their deaf children do not place a burden on the family (11). They are also more likely to feel that there are advantages to being deaf within a deaf family; one such deaf parent in the author's research commented: *"I (can) share my skills and knowledge of deafness. I (can) understand her (daughter's) 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."* 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"* (11).

Preferring to have deaf or hearing children

In 2002, a deaf lesbian couple from the United States chose to have donor insemination from a male deaf friend with the hope that this would increase their chances of having a deaf child (12). Although not actively using genetic intervention, they hoped that genetic inheritance would be favourable for them, as they wanted to increase the chances of passing deafness on. This case caused international debate about the ethics of deliberately creating what some people felt was a "disabled" child (12–17).

The issue of deaf parents preferring to have deaf children is not a new phenomenon; it has been well documented in the past. Passing on deafness to the next generation would keep the Deaf culture alive and would mean that the Deaf community would continue to thrive (18,19). Dolnick (19) comments on this in "Deafness as Culture": *"So strong is the feeling of cultural solidarity that many deaf parents cheer on discovering that their baby is deaf."*

Deaf people, who do not have ties with the Deaf community, but who nevertheless still prefer to have deaf children, may have this opinion because the thought of having hearing children fills them with worry. This may lead them to asking: "How will I cope?" "How will I teach the child to speak?" "What school will they go to?" The psychological reaction of a deaf parent to having a child of unexpected hearing status (either deaf or hearing) may be very similar to a hearing parent having a deaf child (9). There can be feelings of disbelief, fear, and loss. It is possible that another deaf child would fit better into the family unit if other deaf children were already present, a hearing child may just feel isolated. One hearing individual met by the author indicated that she would actually prefer to have deaf children even though she was personally hearing. This was because her family was all deaf with several generations of deafness and as a hearing person among them, she found it hard to cope with being different from the rest of the family.

Some deaf parents have said that they would choose not to have deaf children, if it could be avoided (11). One participant in the author's 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 the struggle they had within a mainstream hearing society. Whereas other d/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 (11).

Several different pieces of research have shown that deaf parents usually do not mind the hearing status of future children, whereas most hearing parents prefer to have hearing children (11,20,21). This implies that deaf parents may be flexible about coping with either a deaf or a hearing child. They may also have a greater awareness of what deafness in a child would mean and therefore could be more ready to accept this than someone with no such personal experience.

It would be logical to conclude from this that more hearing people than deaf people would be interested to find out whether a baby was likely to be deaf or hearing, via the use of a prenatal genetic test. They may also feel more anxious to learn as soon as possible if their baby is likely to be deaf so that they can have a choice as to whether to continue with the pregnancy or not. Attitudes towards such a use of technology are documented in later sections.

Genes, deafness, and genetic testing services

Deafness can result from different factors, including environmental and genetic causes (22). Out of the 1 in 1000 to 2000 children with severe-profound, congenital, or early onset deafness, between 20% and 60% are thought to be deaf due to genetic causes, 20% to 40% due to environmental causes, and the remaining of unknown cause (23–25). Between 59% and 85% of

cases of genetic deafness are thought to be caused by autosomal recessive genes, 15% to 33% by autosomal dominant genes, and up to 5% by X-linked or mitochondrial genes (26–28).

Several hundred genes are known to play a part in inherited deafness (29). Alterations in the *connexin 26* gene are thought to account for up to 50% of childhood genetic deafness, with 1 in 31 people carrying alterations in this gene in certain populations (30,31).

The deafness that results from alterations in the *connexin 26* gene is typically congenital and severe-profound (32), although mild-moderate deafness has also been reported (33). Advances in the molecular genetic research into deafness mean that, for certain families, it is possible to offer a genetic test to define whether a person's deafness is genetic and subsequently, what the chances are of passing this on to children. Such testing and information relating to this is can be obtained via genetic counselling services.

Genetic testing

Genetic testing is a general term that can refer to different types of testing, e.g., diagnostic, carrier, prenatal, and predictive.

- Diagnostic testing is used to diagnose whether a deaf person has a gene alteration(s), which causes his/her deafness.
- Carrier genetic testing tells a hearing individual whether he/she is carrying a gene alteration, which when also carried by their partner, would usually give them as a couple, a one in four chance of having a deaf child.
- Prenatal genetic testing tells a pregnant mother, via an invasive test such as amniocentesis or chorionic villus sampling, whether the foetus has a gene alteration(s) that could cause deafness. The invasive test involves an approximately 0.5% to 1% risk of miscarriage of the pregnancy. Information from a prenatal genetic test could then be used by the parents to decide whether the pregnancy should be continued or not. If not, the mother could have a termination of pregnancy (TOP) from this point on. Prenatal genetic testing is a form of diagnostic testing but it is performed in the prenatal phase, it is also known as prenatal genetic diagnosis (PND).
- Predictive genetic testing could tell a hearing person whether they have a gene alteration(s) that could predispose them to developing deafness later in life.

As more genes linked to deafness are identified and the clinical basis understood, it will become easier to incorporate genetic testing for deafness within routine clinical services. Many clinicians are excited by this prospect (34), but, others may prefer to treat this with some caution. Prenatal testing with selective TOP for deafness raises ethical concerns in relation to whether deafness is a "serious" enough condition to warrant such a course of action. Just because a test is technically possible, does this mean it should necessarily be available? Before such testing becomes routine, it is helpful to consider the longer-term consequences of this procedure.

Genetic counselling for deafness

There is often interest from Deaf individuals to know if and how they have inherited their deafness and what the chances are of passing this on to their children (35). These are issues that can be covered within the clinical service of genetic counselling. Such services are available from genetic counsellors and clinical geneticists working in clinical genetics departments across many parts of the world.

Genetic counselling has been described as "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" (36). Genetic counselling offers clinical information about different genetic conditions and their heritability within a supportive and nonjudgmental environment.

Some deaf parents worry that they would be told that they should not have children if they came for genetic counselling (37). This would not happen within the present-day genetic counselling services in the United Kingdom as the service is "nondirective," i.e., the genetic counsellor does not tell the client what to do nor give advice. The focus of genetic counselling for deafness is now on the individual needs of the patient and their family and does not have a wider agenda to prevent deafness within larger mainstream society (35). However, aside from this, there is still often the misconception that genetic counselling has an ulterior motive, Das (38) states that: "*The high incidence of genetic causes* (of deafness) *indicates that steps should be taken to facilitate Genetic Counselling and conceivably to reduce the numbers affected*" (38). Therefore, there is an assumption that the process of genetic counselling will inevitably reduce the numbers of deaf children born, which may or may not be the case in reality. Aside from this, the actual focus of genetic counselling is on the provision of information and choice. This means that Deaf parents who prefer to have deaf children would be able to access information about genetics and inheritance in relation to this.

Some patients (deaf and hearing), however, do request genetic counselling because they would rather avoid passing on deafness in their family; others simply want information so that they are better informed of the chances of this happening, just for the sake of information.

Requests for PND for deafness are few and far between. There are limited numbers of people who feel that deafness is a serious enough condition to need to find out about during pregnancy or to opt for a termination if the foetus was likely to be deaf. When asked for their opinion on this subject, the majority of deaf and hearing individuals interested in having a test in pregnancy for deafness said they would only do so just to be prepared (39,40). However, in thinking about having a "nondisabled" child, created outside a natural conception, preimplantation genetic diagnosis could be a viable alternative. Such testing for *connexin 26* deafness has been requested, where two hearing parents wanted to avoid having deaf children, preimplantation genetic diagnosis was requested to select the

embryos that did not have the deafness-causing genes with the aim that these would be implanted in the mother (41,42).

Different individuals have different opinions about passing on deafness to the next generation. One deaf couple, known to the author through her work as a genetic counsellor, were so fearful of passing on deafness to their children that they had decided not to have children. The negative personal experience they had in relation to being deaf meant that they felt a heavy responsibility to not "inflict" this on their children. However, the process of diagnostic genetic testing and knowledge of inheritance patterns revealed that their chances of having deaf children were minimal. They were delighted with this news. Another Deaf couple had assumed that because their families were hearing and that their deafness could not be inherited, they were then pleasantly 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 consequently all their children would be deaf. They had a strong Deaf identity and were really pleased to pass on their deafness, language, and culture to their children.

Both couples welcomed the opportunity to discuss their concerns about family planning. This in turn meant that they were more fully informed about their genetic heritage and consequently better able to engage in their future. Genetic counselling also offered them the opportunity to confidentially express the burden and responsibility they felt with regards 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.

Potential outcomes of genetic research

For families who test positive for a specific gene alteration that could cause deafness, it is possible to identify whether hearing parents or siblings are also carriers of such a gene alteration and to offer more specific information about the chances of having deaf children. It could also offer a quick and early diagnosis of deafness in a newborn baby in addition to the audiological testing that they might currently have. Therefore, as more work is done on the molecular genetics of deafness, more accurate information can be offered to families.

Identifying the genetic processes that interplay within the inner ear may lend itself eventually to gene therapies for deafness. This could replace the need for cochlear implants in children, and the obvious pain and risks that major surgery brings. It has also been suggested that, within the next 50 years, hair cell regeneration within the cochlear will be possible (43).

The potential impact of genetic research on families with deafness is summarized by Arnos et al. (35): *"Advances in molecular genetics will eventually bring about new options for prenatal diagnosis of deafness and prenatal or postnatal treatment. Deaf and hard-of-hearing people and parents of deaf children will surely have different feelings and may make different choices regarding the options that will be available to them. Some of the issues that arise may be similar to those that have come up as genetic technology has been applied to the diagnosis and treatment of other hereditary conditions. The sociocultural aspects of deafness will lend additional considerations to these discussions"* (35).

Genetics, eugenics, and deaf people

There have been many attempts throughout history to prevent deaf people from having children so that the numbers of deaf people would be reduced within society. Alexander Graham Bell, inventor of the telephone and also a leader in 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. Here he advocated that deaf people should not be allowed to marry other deaf people, but should marry hearing people so that the chances of passing on deafness to their children would be limited (44). At that time the inheritance of genetic conditions was poorly understood and he mistakenly made the assumption that this would be an effective way of preventing deafness from being passed on. In fact, even if a deaf adult married a hearing partner, if the deafness was due to a dominant gene alteration there would be a 50/50 chance of passing this on to any children. Bell had a great respect for deaf people (his own mother was deaf and so too was his wife), but still felt that deafness was a disability and should be avoided if at all possible. This view, although derived from well-meaning intentions, is seen as insulting by many culturally Deaf people. As such this work has been discussed among British, European, and American deaf studies academics and lay people for over a hundred years since (45).

Another key event in history that involved deaf people related to Hitler's regime in the Second World War. In the Nazi programme, that advocated the eugenic pursuit of the perfect Ayrian Race, Hitler ordered deaf children and adults to be sterilised so that they could not pass on deafness to their children, and this happened to 16,000 to 17,000 deaf people. In addition to this, other deaf people were killed as part of "Operation T4" the Nazi programme designed to "wipe out" disabled citizens (46). Again, the incorrect assumption was made that deafness is always inherited and also another assumption was that deaf people will pass it on to their children. In fact, the majority of deaf children are born to hearing parents.

Given the historical context to the misuse of genetic knowledge, it is not surprising that d/Deaf people are often suspicious of modern day genetics services. The very fact that PND for deafness with selective termination for a deaf foetus is technically possible is sufficient for Deaf people to feel that there is another eugenic agenda being impressed upon them. There is often a sense that genetics services in the past have "devalued" the role of Deaf people in society. With this in mind, it is

therefore imperative that genetic counsellors and geneticists are mindful of the historical context within which they practice in the present day.

It is important that a "culturally neutral" genetic counselling service is available to deaf people and their families (47), where Deaf patients 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.

General attitudes to the medical model of deafness

As deafness can be viewed from different perspectives, there are often differing beliefs about appropriate medical intervention in relation to this. Deaf people may be sensitive to technology that aims to "cure" deafness and, as such, there has been clear resistance to cochlear implants (48). Here, the view is taken that deaf children should not be put through extensive, painful surgery to try and make them hearing when, to them, being deaf is not insurmountable.

Wheeler, from the Deafness Research Foundation, United States, believes that there is still compatibility between the preservation of the Deaf community and search for a cure/effective treatment for deafness (43). He suggests that by removing communication barriers, so that sign-language users have equal access to *"learning and enjoyment of life,"* a better quality of life will be achieved. At the same time those who wish to use treatments or cures can do so. However, the real argument from many Deaf people is that as most deaf children are born into hearing families, decisions to have treatments or cures will be made by hearing people who probably are not aware of the cultural model of deafness. Such hearing people, with their ignorance of the Deaf World, will make decisions for their deaf child according to their "hearing" perspective. Therefore, such deaf children are "cured" of their deafness before they are old enough to make choices for themselves, so missing the opportunity to be part of a community they could have naturally belonged to.

Having an awareness of what the Deaf community offers is something that many Deaf people aim to educate hearing people about, so that hearing parents are able to make informed decisions about their child's future. However, input from d/Deaf people about the medical or educational management of deafness has largely been ignored in the past (49). This situation is improving but still has a long way to go to create a working partnership between parents of deaf children, the Deaf community, and professionals working in deafness (50).

The British Deaf Association (BDA) or Sign Community is "the U.K.'s largest national organisation run by Deaf people for Deaf people" (51). The BDA has a policy on genetics (updated in May 2003) that stresses concern over the use of PND with selective termination of "deaf" pregnancies. In addition they "demand" 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 prenatal tests or to undergo termination where it is discovered that the foetus is deaf"*(52).

Therefore, the BDA believes that d/Deaf and hearing parents attending a genetic counselling clinic in the United Kingdom do not at present receive enough information to enable them to make informed decisions about deafness. The BDA intends to rectify this by implementing more Deaf awareness training among genetics professionals.

The National Deaf Children's Society (NDCS) also has a policy on genetics. In this, they advocate choice and information: *"The Society. . .recognizes the rights of potential parents from families who have a history of deafness to take advantage of genetic testing and antenatal 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" (53).*

Support groups such as the BDA and NDCS consist of deaf and hearing individuals with an interest in the current clinical, educational, and support services in place for deaf people and their families. These groups are a powerful force that aims to help prevent discrimination and promote acceptance of deafness, whether perceived from the medical or cultural perspective.

Attitudes towards genetics may sometimes be seen as linking in with cultural identity. Those Deaf people who are against the eugenic practices of the past will often have negative views towards modern day genetics services (54). Such attitudes have been well documented over the last ten years, the following gives an overview of some of this work.

Attitudes towards genetics

The views of a collective group of culturally Deaf people attending a conference called "Deaf Nation" at the University of Central Lancashire, United Kingdom in 1997 were studied to ascertain attitudes towards genetics (55,56). Delegates were asked to complete a questionnaire which asked for their views about genetic technology and how they felt about its use with respect to deafness (e.g., for 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. Some of this group indicated that they felt threatened by the perceived "misuse" of genetic technology, the biggest fear relating to prenatal diagnosis for deafness followed by selective termination if the foetus had the genes for deafness. The worry was that if such actions were utilized to any great extent, then the Deaf community would diminish.

A much larger study ($n = 1314$) has since been completed by the same authors. Here, the attitudes of d/Deaf, hard-of-hearing, and deafened adults as well as hearing parents of deaf children were documented (11,39,40). Participants were collected from medical and educational sources, social services, charities, and support groups for the deaf, i.e., a wider selection of participants were ascertained than gathered in the Deaf Nation study. However, the same findings were replicated among the culturally Deaf participants—involving negative attitudes towards genetic technology. On the other hand, those participants who identified with the wider mainstream hearing society tended to have quite positive views about the use of genetic technology.

Participants were given a list of positive, neutral, and negative words and asked to tick those from the list that described their feelings about new discoveries in genetics. The results showed very different attitudes between groups (Fig. 11.1). Deaf participants were more likely to select negative words ($\chi^2 = 42.2$, $df = 6$, $P < 0.001$). The most frequently ticked word was "concerned," and just under half of the group ticking this was culturally Deaf. Hearing participants were more likely to select positive words ($\chi^2 = 156.7$, $df = 8$, $P < 0.001$), the most frequently ticked word being "hopeful." Hard-of-hearing and deafened participants were more likely to tick a mixture of words, the most popular was "cautious."

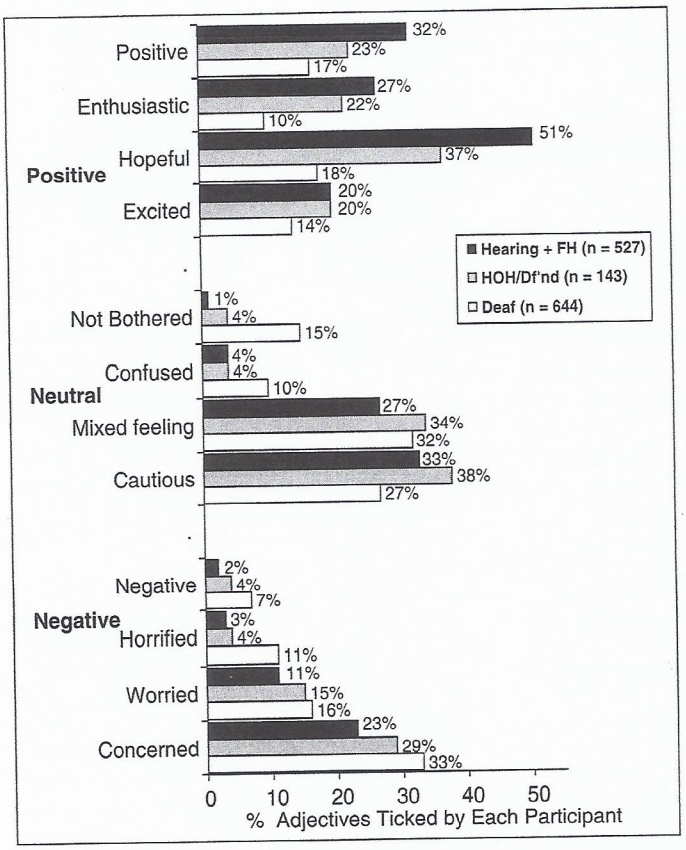

Figure 11.1 Percentage of participants who ticked different adjectives to describe their feelings about new discoveries in genetics. *Abbreviations*: HOH/Df'nd, hard-of-hearing and deafened participants; Hearing + FH, hearing participants who have either a deaf parent or a deaf child. *Source*: From the Journal of Genetic Counseling.

Participants were given the opportunity to comment on their feelings about new discoveries in genetics. The following are a selection of these.

Some participants felt that new discoveries in genetics would be positive:

> *"We must go forward in genetics to help us understand causes of deafness and other disabilities caused through genes."*
> (nonculturally deaf participant)
>
> *"I think it is a good idea—to stop the genes passing on into the next generation."*
> (nonculturally deaf participant)

Some had negative comments about new discoveries in genetics:

> *"Angry at people trying to mess with nature and interfering with deaf people - leave us alone!"*
> (culturally Deaf participant)
>
> *"My hands is little nerve* (I feel nervous). *To think it is worst soon* (I feel this is the worst situation)"
> (culturally Deaf participant, who used BSL as first language, translated their feelings from BSL into written English.)

And some comments were mixed:

> *"Interested but do not feel involved"*
> (nonculturally deaf participant)
>
> *"Enthusiastic about benefits it can bring—early diagnosis, treatment to improved levels/quality of hearing, BUT concerned it will be used to increase abortion."*
> (hearing parent of deaf children)

Attitudes towards genetic testing as part of the newborn hearing screening programme

A diagnosis of deafness within a hearing family always has the risk of being delayed, due to neither the parents nor health professionals anticipating or specifically looking out for it. The Newborn Hearing Screening Programme offers the opportunity to obtain a diagnosis as early as possible by screening all newborn babies for audiological deafness (57). The earlier the diagnosis, the sooner that appropriate communication and education tools can be implemented thus giving the d/Deaf child the best possible chance of "normal" development (58). A delayed diagnosis may impact on the acquisition of effective language and this in turn may affect emotional and cognitive development.

Adding genetic testing for the *connexin 26* to the programme and thus making it an automatic part of Newborn Hearing Screening has been discussed (59); this is already in place in some countries. There is some resistance to this, however, due to concern that such testing, although possibly useful for parents to know a genetic cause to their child's deafness, may make it seem implicit that prenatal diagnosis should be utilised in the next pregnancy (60). Therefore, careful consideration of the impact of this should be given before genetic testing services are automatically added onto the audiological testing.

Research looking at the attitudes of deaf adults towards the use of genetic testing as part of the Newborn Hearing Screening Programme has shown that attitudes are generally positive (61) with most deaf participants perceiving the genetic and audiological testing offered as a useful way of diagnosing deafness earlier than has been done in the past. Another study looking at the attitudes of deaf, hard-of-hearing, and hearing participants also showed that most agreed that newborn genetic testing for deafness was appropriate (20). Deaf and hearing people alike appear to agree that the earlier the diagnosis of deafness, the better the outcome for the deaf child.

Attitudes towards genetic testing for deafness from the general public with no knowledge and experience of deafness

Ryan et al. documented the views of 91 pregnant women attending their 12- to 13-week booking scan at a maternity hospital in Scotland towards having a personal carrier test for deafness and subsequent prenatal test should both members of the couple be carriers for a deafness-causing gene (62). The majority had no personal experience of deafness either in themselves or in their relatives. Respondents indicated that the vast majority were interested in carrier and prenatal testing and if found to have a baby that was likely to be deaf, most said they would not have a TOP. This study is interesting as it places a value of what deafness means for people who do not have a family history of it. This study appears to show that people from the general public may perceive it as a condition to know about but not necessarily one to justify the ending of a pregnancy.

Prenatal testing for Cx26 gene alterations is already available to pregnant women having chorionic villus sampling in a pregnancy in Italy (63). Out of more than 5000 such women, with neither experience nor family history of deafness who were offered the testing, 55% chose to go ahead. As yet, only carriers have been identified, however it is only time before foetuses likely to be affected with deafness will be identified. It is not known whether parents would choose to end the pregnancy or not. It is very likely that such a population screening programme for deafness will be rigorously rejected by members of the worldwide Deaf community.

Attitudes towards genetic testing for deafness from deaf people and their families

The author and colleagues documented the views of 87 Deaf participants ascertained from delegates attending a conference on Deaf issues for Deaf people (55,56). This study showed that there was a small group of Deaf participants who said they would be interested in PND for deafness and also preferred to have deaf children. There was the theoretical possibility that they may choose to have a termination for a hearing foetus. The same authors conducted a much larger study documenting the views of 644 deaf individuals, 143 hard-of-hearing individuals and 527 hearing individuals with either a deaf parent or a child (39,40). From this study, 49% hearing participants with a family history of deafness, 39% hard-of-hearing and deafened participants, and 21% deaf participants said they would all be interested in having PND for deafness. From those interested in prenatal testing for deafness, 16% hearing, 11% hard-of-hearing and deafened, and 5% deaf participants said they would do so because they would have a termination if it were to be shown that the foetus was deaf. For the participants who said they would choose this option, it is possible that they may have had such a negative experience of living with a hearing loss in themselves or their family, perhaps observing that deafness created isolation or even discrimination, that they did not want to take the risk of passing on deafness to their children.

Aside from this, the majority of all groups who said they would use PND did so only for preparation for the baby (e.g., so they could learn BSL) rather than because they wanted to have a termination of a deaf foetus. This could be seen as reassuring to members of the Deaf community in that most would not wish to end the pregnancy if the test indicated the baby was likely to be deaf.

Other research has produced similar results; Brunger et al. looked at 96 hearing parents of deaf children ascertained in a hospital setting. There, 96% of the sample had positive views towards genetic testing and 87% said they were interested in having PND for deafness with the intention of using this just for preparation rather than acting on it via a termination (64). Martinez et al. gathered the views of 133 hearing students and 89 deaf and hard-of-hearing students from a U.S. university. They showed that 64% hearing participants and 44% deaf participants said they would be interested in having PND for deafness with no data on opinions about termination (20). Stern et al. used the same study questionnaire and similar groups of participants as the Middleton et al. research described above. They gathered the views of 135 deaf, 166 hard-of-hearing and deafened, and 37 hearing individuals from a number of different sources, including support groups as well as medical and educational settings. The results were classified into those who identified with the Deaf community, those who identified primarily with the Hearing World (including hearing and some deaf participants), and those who identified with both communities. The data showed that 23% participants who identified more with Deaf community were interested in prenatal diagnosis for deafness, compared to 47% of participants who identified more with the hearing world. With regards to attitudes towards termination, approximately 8% of participants who identified with the hearing world said they would consider having this if the foetus was deaf but none of those who identified with the Deaf

community said they would (21). Finally, a study by Dagan et al. (65) looked at the views of 139 hearing parents of deaf children from Israeli Jewish families. 49% said they would consider having prenatal diagnosis for deafness with 17% saying they would consider having a termination for deafness (65).

Therefore the results from this selection of studies follow approximately the same patterns, with some participants interested in prenatal diagnosis for deafness, but much less interested in TOP for deafness.

Within work by the author, a very small number of deaf participants, three (2%) did say they would consider having prenatal diagnosis with selective termination of a hearing foetus, since they preferred to have deaf children (39,40). This reaction is somewhat extreme, and it is difficult to say whether, in reality, anyone would choose such a course of action. However, what this does demonstrate is the extent of the feelings of Deaf cultural solidarity that some Deaf people have, and also the fact that deafness is not automatically perceived as a disability. Indeed to be hearing in this instance would be a disadvantage. This fits in with previous literature already documented that shows some deaf parents prefer to have deaf children (14,18,39,40,55,56).

Summary profile of parents interested in prenatal testing for deafness

The author has also documented the attitudes of parents of deaf children towards many different aspects relating to the deafness in the family (11). It is possible from this work to create a profile of the type of person who may choose to have prenatal genetic testing for deafness. In summary, deaf parents of deaf children who were interested in prenatal diagnosis for deafness (because they wanted to avoid passing deafness on) were more likely to prefer to have hearing children, see their deaf children as disadvantaged, feel an actual burden of having a child who is deaf, and want a cure for deafness in their child. Hearing parents interested in prenatal diagnosis for deafness were more likely to consider termination for deafness as acceptable, to find communication with their child less than perfect (ranging from successful to poor), and to find the experience of obtaining education for their child difficult or complicated.

Looking at these collective results, it is possible to infer that many factors influence interest in PND for deafness. If such factors were modified then interest in PND for deafness might decline. For example, if parents were able to see their child as advantaged or less of a burden, or if they felt that communication with their child was easier or there were more straightforward processes to obtaining appropriate education, then they may be less interested in PND for deafness.

Conclusions

This chapter has reviewed some of the literature documenting the attitudes of deaf individuals and their families towards various issues surrounding genetic testing for deafness. This has been considered within the context of Deaf culture and the varying perceptions of the impact of deafness. Deafness does not appear to be a condition that most people (deaf and hearing) feel is "serious" enough to warrant prenatal testing nor selective TOP. However, inevitably there are people who would consider using the technology in this way. Such people tend to perceive deafness as a burden or disadvantage, and one they were inclined to view as a struggle to live with. Despite the negative picture created about deafness, many other people view deafness positively. Culturally Deaf participants are particularly optimistic about their situation and feel that being deaf is not a disability and also not something that genetic technology needs to interfere with. This shows that deafness is not a condition that is clearly detrimental.

If consideration is ever given to large-scale "management" of deafness, for example, by population carrier screening, genetic testing added onto the Newborn Hearing Screening Programme or mass-scale availability of prenatal testing for deafness, involvement in policy decision making surrounding this must include input from deaf, culturally Deaf, hard-of-hearing adults as well as parents of deaf children. All such people are directly affected by such programmes and have valuable insight to offer about the potential impact of this.

Appropriate and effective clinical services for deaf people can be developed as long as health professionals take the time to learn about the diversity of cultural attitudes held by different people affected by deafness (66). Genetic counselling services require a specialist knowledge of deafness, Deaf culture, and the role that genetics has played within history (67). It is also imperative that communication and language differences are embraced. Training in Deaf Awareness would be valuable for any health professional wanting to work in this area.

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