

Position statement	
Title	Genetics, Stem Cell Therapies and Deafness
Approved by	Board of Trustees
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Executive director owner	Brian Gale
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1. Policy

Introduction

1. 1. NDCS welcomes all medical discoveries that contribute to our greater and more positive understanding of deafness, empower individuals by giving them choices and enhance the potential of deaf children and young people and their families to lead lives which are as rich and varied as possible.
- 1.2. NDCS views as desirable research into gene-based therapy and stem cell therapies that might allow parents the choice of reducing the impact of deafness on their deaf child's social and educational development. We recognise that if any form of gene-based therapy or stem cell therapy for deafness does become available in the future the resulting improvement to hearing could possibly be more permanent than that afforded by cochlear implants or hearing aids. If this were to occur, it would pose different ethical dimensions to the choices involved; for most parents and deaf young people this will make the decision to use such an intervention method more complex.

There are many factors that may result in deafness and only about half of the causes of deafness can be attributed to genetic factors. NDCS welcomes the rapid advances in our understanding of the genetics of deafness, but stresses that the wider implications of all genetic research should be treated with the utmost sensitivity and care.

- 1.3. NDCS believes that the prospect of future medical developments in deafness should not detract from the support that deaf children presently need. NDCS will continue to campaign for health, education and social care services to provide the support that deaf children need to achieve their potential.

Genetic Counselling and Testing

- 1.4. Genetic testing and counselling services should be offered to deaf children and young people and their parents where it is wanted and considered to be appropriate in providing beneficial knowledge to either the child or the family.
- 1.5. NDCS urges the development of services that are fully accessible to deaf parents and young people who have sign language as their first language.
- 1.6. NDCS urges the development of research and services that:
 - enable the early identification of disabilities or health conditions that may, at some point in a deaf person's life, co-exist with deafness so that appropriate monitoring, intervention plans and services can be put in place
 - identify the vulnerability of a child to medication that, if administered, will create hearing loss or exacerbate the degree of hearing loss.

Genetic Screening

- 1.7. NDCS does not support whole-population screening for genetic conditions, with the consequent risk of moving towards a society in which difference is no longer accepted or tolerated.

Prenatal Genetic Testing

- 1.8. NDCS acknowledges that some parents, whether deaf or hearing, choose to undergo prenatal genetic testing in order to prepare for the child's future, while others may consider a termination of the pregnancy. It is not the role of NDCS to express an opinion about the way in which an individual chooses to use the information, but fundamental messages that NDCS will continue to promote include:
 - a) Deaf children and people make a positive contribution to society and can have happy and rewarding lives.
 - b) If appropriate support is provided to the family and child and positive attitudes are fostered, there is no reason why deaf children should not achieve at the same level as hearing children.

In Vitro Fertilisation and Preimplantation Genetic Diagnosis

- 1.9. It is not the role of NDCS to express an opinion on the way society decides to make use of preimplantation genetic diagnosis. (See section supporting section paragraphs 2.17 and 2.18)

Treatment of deafness with stem cells

- 1.10. There are significant research developments into how stem cells can be used to treat deafness. NDCS views as desirable medical research that might allow parents the choice of reducing the impact of deafness on their deaf child's social and educational development.

NDCS's understanding is that a number of obstacles need to be overcome before cell stems can be used safely and it will take a number of years to overcome these obstacles. It is also likely that cell stems will not bring the same level of benefit in all deaf people. It is not the role of NDCS to express an opinion about the way in which stem cells are used.

The fundamental messages that NDCS will continue to promote include:

- a) Deaf children and people make a positive contribution to society and can have happy and rewarding lives;
- b) If appropriate support is provided to the family and child and positive attitudes are fostered, there is no reason why deaf children should not achieve at the same level as hearing children.

Conclusion

- 1.11. In conclusion, NDCS recognises the rights of families with deaf children and potential parents from families who have a history of deafness to take advantage of the various genetic technologies and to use the results of such tests in a way that suits the individual family.

If asked for advice, NDCS will endeavour to ensure that the family receives balanced and clear information about deafness and genetic technologies in order to enable them to make an informed choice.

NDCS will continue to campaign for a society in which childhood deafness is not stigmatised and deaf children are viewed as individuals who have all the opportunities that are available to hearing people to develop and maximise their skills and abilities.

2. Supporting information for the position statement

Note: Factual changes to this section will be made from time to time to keep up with recent developments in genetics without requiring Trustee Board approval unless they necessitate changes to policy.

Introduction

- 2.1. Just as children inherit features, such as hair or eye colour, from their parents, sometimes deafness is inherited. Most people have 23 pairs of chromosomes in each of their cells. These are microscopic structures that contain genes. One of each pair of chromosomes is inherited from the mother and the other from the father. Sometimes there is an alteration or change in a particular gene that results in a different characteristic, for example deafness. The change in a gene can be called a 'fault' or 'mutation'.

- 2.2. Genes can work in different ways. Sometimes a child only needs to inherit one faulty gene from either their mother or their father to have that particular characteristic (*dominant inheritance*). Sometimes the child will need to have the same faulty gene passed on from both parents in order to have the condition (*recessive inheritance*). There are other different patterns of genetic inheritance that are less common.
- 2.3. Some people are known as *carriers*. This is where they have the altered gene but do not have the characteristic. So a person who is a carrier for deafness would carry the altered gene, but would not have deafness themselves.
- 2.4. Sometimes a gene mutation is a one-off that simply occurred by chance. This means that, although the deafness has a genetic cause, it has happened for the first time in that person and has not been inherited. However, it could be passed on to future generations.

NDCS vision and values

- 2.5. Our vision is a world without barriers for every deaf child. We believe that:
- Deaf children can do anything other children can do, given early diagnosis and the right support from the start.
 - Deaf children should be involved in decisions which affect them at as an early age as possible.
 - Families are the most important influence on deaf children and young people, and need clear, balanced information to make informed choices.
 - Effective language and communication skills lie at the heart of deaf children and young people's social, emotional and intellectual development.
 - Deaf children should be valued by society and have the same opportunities as any other child.
- 2.6. NDCS's youth strategy also emphasises that deaf children and young people have the right to information and to fully participate in decisions that affect their lives.
- 2.7. NDCS is committed to empowering parents of deaf children, and deaf children and young people themselves, to make informed choices by providing impartial advice and information on all aspects of childhood deafness. The field of genetics is wide and fast growing and NDCS feels it is important to clarify its position regarding different areas of genetics and genetic research.
- 2.8. NDCS's attitude to genetic developments is based on:
- a) A belief that deafness should not be regarded as an illness or a life-threatening or life limiting condition. The social model of disability on which NDCS draws on its response to issues relating to genetics asserts that deafness in itself is not a disability, but the way society responds to it does disable people.

- b) The NDCS policy on inclusion which states that: NDCS believes that inclusive practice arises from a philosophy which views diversity of strengths, abilities and needs as natural and desirable, bringing to any community the opportunity to respond in ways which lead to learning and growth for the whole community, and giving each and every member a valued role.

Genetic counselling

- 2.9. Genetic counselling provides information and support for individuals and families who are or have a chance of being affected by a range of inherited conditions, including deafness. Genetic counselling provides families with information about the cause of the condition, how it might affect the child and family in the future and, if the family wish to know, how likely they are to have another child with the same condition. Genetic counselling enables families to make informed decisions about their child, their child's future and may help them when planning for their family.
- 2.10. Some families find it helpful to know whether deafness and any other associated medical condition were inherited. Other families prefer to wait until their children are grown up and able to decide for themselves.
- 2.11. NDCS believes that families should be offered the chance to discuss their situation with their local clinical genetics (genetic counselling) team and should be referred by the doctor in charge of their child's audiological care.

Genetic testing

- 2.12. Families with a deaf child may be offered a genetic test. This may happen shortly after a baby has been identified as being deaf following newborn hearing screening. Alternatively, families with older children and young people themselves may be offered a test.
- 2.13. Genetic testing involves the child and possibly other family members having a blood test. The blood sample is used to look for a gene or genes known to be involved with deafness. Families will also be asked about any family history of deafness and this information can be a very important part of identifying a genetic cause.
- 2.14. It has been established that permanent deafness in about 50 per cent of children is due to a genetic cause. However, not all the genes related to deafness have yet been identified and only one (called *Connexin 26*) is routinely tested for in the genetics clinic. Some others can be requested specially (for susceptibility to ototoxic medications, some syndromes and so on, for example).

An altered *Connexin 26* gene affects the functioning of the hair cells within the cochlea and causes sensory-neural deafness. This means that the sound cannot be transmitted to the auditory nerve and onwards to the brain. Children who have deafness due to *Connexin 26* do not usually have any associated health conditions.

In about 30 per cent of children with genetic deafness, the deafness is part of a more complicated picture, known as a *syndrome*. A syndrome is simply a medical term meaning a collection of signs or symptoms that doctors recognise as being linked or having the same cause. Again, some of these syndromes can be confirmed with a genetic test, but many cannot. Genetic testing for a specific syndromic cause of deafness is usually only offered if the child has other signs or symptoms that would fit with that condition. For example, in Ushers syndrome progressive blindness is part of the syndrome in addition to the deafness.

Improved knowledge of genetics and improvements in genetic diagnosis could enable earlier identification of other disabilities or health conditions that coexist with deafness and develop later in life (for example: visual disorders such as Usher syndrome, keratitis-ichthyosis-deafness (KID) syndrome, Alstrom syndrome; kidney dysfunctions such as in Alport syndrome; heart conditions as part of Jervell and Lange-Nielsen-syndrome). This will help families and professionals plan for educational and health care needs, including audiology care. Having a certain syndrome, for example, may mean that a hearing loss will worsen over time and therefore more frequent hearing checks may be needed.

NDCS therefore supports the development of genetic diagnosis and would want the NHS to ensure such a service was widely available and accessible to deaf children, young people and their families.

Genetic vulnerability to medicines

- 2.15. Some children have a rare genetic increased susceptibility to deafness caused by the administration of certain medications, most frequently given in response to life-threatening conditions. It is possible to have a genetic test to identify whether they have this gene. If they do, the information can help inform the family and their doctor prior to the treatment.

Genetic Screening

- 2.16. Genetic screening of individuals is used to determine whether they carry a gene for a particular disease or condition that may affect them in the future or may be passed on to their children. The term 'genetic screening' is used to mean a test which is offered to a whole population or group within the population rather than specifically offered to an individual because of their personal or family history of a condition ('genetic testing').

Prenatal Genetic Diagnosis

- 2.17. If a family has a history of deafness and a known genetic cause has been identified, i.e. the particular gene has been confirmed through testing, an individual or couple may choose to have prenatal genetic testing in a future pregnancy. This type of testing may identify whether the foetus has inherited the familial deafness gene but is usually unable to predict exactly the level of deafness the child will have.

In-vitro Fertilisation and Preimplantation Genetic Diagnosis (PGD)

- 2.18. Preimplantation genetic diagnosis involves using in-vitro fertilization (IVF) techniques to produce embryos, but with the additional step that one or two cells from each embryo are removed and tested to see whether or not that embryo has inherited a particular gene or not. This allows selection of which embryos will be placed in the uterus. It is available for couples who have a chance of passing on a specific genetic condition. As with prenatal genetic diagnosis, a genetic cause (i.e. a particular gene) must already be identified in the family for PGD to be an option. PGD is possible and licensed by the Human Fertilisation and Embryology Authority (HFEA) for a range of genetic conditions. This does include several genetic causes of deafness. Most of these, though not all, are syndromes where deafness is one part of the condition and there are implications for health. NDCS acknowledges that some families with a history of deafness may wish to use this type of technology in planning their family.
- 2.19. Section 14 of the *Human Fertilisation and Embryology Act 2008* means that, where families are undergoing preimplantation genetic diagnosis, an embryo with an 'abnormality' that will develop into a 'serious physical or mental disability' must not be deliberately selected. The Act does not require families to undergo screening for such abnormalities (i.e. if a family are undergoing PGD for one condition at their request, there is no requirement for their embryos to be tested for other conditions within the family), but where a screening has taken place, such embryos cannot then be 'preferred' if other healthy embryos are available.

Treatment of deafness with stem cells

- 2.20. Considerable advances have been made in understanding how stem-cell treatment might eventually reduce the level of deafness in some deaf people. Some researchers have reported success in using stem cells to reduce deafness in animals. Medical research into methods of turning human embryonic stem cells into ear cells needed for hearing is ongoing.
- 2.21. **Definition of syndromes mentioned in this statement**

Alport Syndrome: The second most common inherited cause of kidney failure. Hearing loss and, occasionally, visual problems are associated with Alport Syndrome.

Alstrom Syndrome: A rare progressive genetic disorder characterised by obesity, deafness and visual problems in childhood and diabetes and kidney failure in adults.

Keratitis-ichthyosis-deafness syndrome: An inherited disorder in which affected persons have:

- a) Keratitis – gradual destruction of the cornea of the eye sometimes leading to blindness.
- b) Ichthyosis – localised areas of disfiguring reddish thickened skin.
- c) Deafness at birth.

Jervell and Lange-Nielsen-syndrome: A rare syndrome that causes deafness and a disruption of the heart's normal rhythm. Beginning in childhood, the irregular heartbeats increase the risk of fainting and sudden death.

Usher syndrome (sometimes referred to as "Usher's syndrome"): A relatively rare genetic disorder that is associated with a mutation in any one of 10 genes resulting in a combination of hearing loss and visual impairment and is a leading cause of deafblindness.

3. References

1. Guidelines for Aetiological Investigation, British Association of Audiological Physicians (BAAP) and British Association of Community Doctors in Audiology (BACDA), available from www.nhsp.info.
2. Making Babies: reproductive decisions and genetic technologies, Human Genetics Commission, January 2006.