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ACTION ON HEARING LOSS

Genetics and deafness

THE FACTS

Genetics and deafness

This factsheet is part of our **Ears and ear problems range**. It is written for parents of children who are deaf and parents-to-be who are deaf. We use the term 'people who are deaf' to refer to people who are deaf, deafened or have any form of hearing loss.

Read this factsheet to find out:

- What are genes?
- What is genetics?
- How can a DNA sequence affect hearing?
- What types of gene mutation cause deafness?
- Why is genetic information helpful?
- What is a genetic test?
- What are the benefits of genetic research into deafness?
- Where can I get further information?

If you would like this factsheet on audio tape, in Braille or in large print, please contact our helpline – see front page for contact details.

What are genes?

Genes determine the characteristics we inherit from our parents. Every cell in our body has two copies of about 25,000 different genes – one copy is inherited from your mother and one from your father. They are mostly grouped together in structures called chromosomes. You can see chromosomes under a microscope, but not genes.

Genes are made from a chemical called deoxyribonucleic acid (DNA). DNA is built from four different chemical 'building blocks'. The order in which these building blocks are strung together is unique to every gene and is known as the 'DNA sequence'. The DNA sequence of a single gene tells the cell how to make one of the many thousands of proteins that the cell needs to do its usual job.

What is genetics?

We all get or inherit characteristics such as the colour of our eyes and hair from our parents. Genetics looks at how these characteristics are inherited by studying genes.

Some types of deafness can also be inherited. About one in 1,600 children is born moderately to profoundly deaf because of a genetic cause.

How can a DNA sequence affect hearing?

Sometimes the DNA sequence of a gene can change – scientists refer to this as a 'mutation'. These changes can mean that the gene does not function correctly. For example, if the gene mutation interferes with the instructions for making a protein needed for hearing, then it could cause deafness.

What types of gene mutation cause deafness?

The chances of developing deafness caused by a mutated gene depends on whether the mutation is **dominant** or **recessive**.

Dominant gene mutation

A **dominant gene mutation** causes deafness when only one copy of the gene is affected. The affected gene can come from the mother or the father.

The chance of passing on this mutation to your children is one in two. However, dominant genes do not always have the same effect on everyone. In the same family, a gene can cause profound deafness in one person and mild deafness in another. Sometimes, it may not affect a person's hearing at all, or the effect of the gene may be so mild that doctors cannot see it.

A dominant gene mutation may have been in the family for generations. Alternatively, it can sometimes appear for the first time in a family without a history of deafness.

Recessive gene mutation

A **recessive gene mutation** causes deafness only when both copies of the gene are affected, so both the mother and father must have passed on an affected gene to the person who is deaf.

If you have this type of deafness, the chance of this type of mutation causing deafness in your children is far lower because both you and your partner must have an affected gene.

Someone who has a recessive mutation and a normal copy of the same gene will be hearing because the recessive gene 'recedes' into the background. They are called 'carriers' and can pass on the affected gene to their children. Most carriers never know they are carriers unless they have a genetic test.

Recessive mutations are the most common cause of inherited deafness. A person whose deafness is due to a recessive mutation may have parents who are hearing. They may

also have brothers and sisters who are both hearing and deaf, and no family history of deafness – even though the deafness is genetic in origin.

What are mitochondrial genes?

Most of our genes are located on chromosomes, but some are found in structures called mitochondria in the cells of our bodies. Mitochondria are inherited from our mothers only. This means that if deafness is caused by a mutation in a mitochondrial gene, only women can pass on deafness to their children, although both men and women can have this type of deafness.

Why is genetic information helpful?

Genetic information about deafness may be helpful:

- to find out the chances of having a child who is deaf if you are deaf or if deafness runs in your family
- to try to identify the cause of your deafness
- to look for medical conditions that might be associated with your deafness. About 30% of deafness in young children is associated with other medical conditions or 'syndromes' – this type of deafness is called 'syndromic deafness'. These additional conditions are usually of no concern, but it may be important to identify some. For example, someone with Usher syndrome has deafness from birth and loses their sight gradually.

How can I get this information?

If you would like information about genetics and deafness, you will need to have a consultation in a genetic clinic. This consultation is often known as 'genetic counselling'. There are about 30 NHS centres offering genetic counselling for deafness around the country. To get an appointment at one of them, your GP, paediatrician, ENT consultant or audiological physician will need to refer you. It does not cost you anything to get genetic counselling on the NHS.

What does the consultation involve?

You will see a member of the genetics team – this may be a clinical geneticist (a doctor), a clinical nurse specialist in genetics or a genetic counsellor (someone with a science background and knowledge of genetics) – who will discuss the facts of your deafness. You will usually have one or two appointments, with each session usually lasting up to an hour. Other people in your family can go with you if they want to.

You will see someone who is sympathetic and trained to talk to you with sensitivity. During the consultation they:

- Will try to explain any facts (such as the different ways in which deafness can be inherited) as clearly as possible and tell you about any genetic tests. They will make sure that you understand what is being said so that you can make up your own mind.
- May ask you about your family history to understand how deafness is inherited in your family. Remember that you don't need to have a family history of deafness to have inherited deafness (see *What types of gene mutation cause deafness?* on page 3).
- May ask you about any relevant medical history to find out if your deafness has an environmental cause or if it is syndromic – you may need a medical examination. Syndromic deafness is when a person has deafness and other medical conditions (see page 4).
- May look at your audiological records to help with the diagnosis.
- May arrange for you to have other tests, such as special scans, which will help them find out about problems in parts of your ear, such as the inner ear, and to exclude any common syndromes.
- May suggest you have a genetic test to help with the diagnosis.
- May tell you how your deafness is inherited or why there may be uncertainty.

What is a genetic test?

You may be offered a genetic test as part of your consultation. You will need to give a blood sample, which will be tested to see if you have a mutation in a gene needed for hearing.

If the gene mutation that causes deafness in your family can be identified, it may be possible to:

- Get a more accurate diagnosis of the cause of your deafness. This may help reveal whether your deafness is likely to get worse.
- Help predict whether you are likely to develop other health or medical problems.
- Get a more accurate prediction of the chances of having a child who is deaf.

If you are deaf, you will be the first member of your family to be tested. If a genetic mutation is not found then other family members will not be tested because a cause has not been established for your deafness. If a mutation is found, other interested family members will be offered testing.

Children aged 16 and younger are not tested to see if they are carriers unless they are old enough to give informed consent – this means they have to understand the test to which they are agreeing. The term for this in medical law is ‘Gillick competence’.

Children can be tested if there is real concern that they may be deaf and if genetic testing can confirm the cause of the deafness.

What happens after a genetic consultation?

If you are not offered a genetic test, you will get a letter explaining what was discussed during your consultation. You can contact the genetic clinic if you don’t understand anything in the letter.

If you had a genetic test, you may have to wait months for the results of the test. This is because clinics test blood samples in batches and may wait a while before they have enough samples to test. You will get a letter in the post telling you about the results. If the test results show that your deafness is due to a genetic mutation, you will be invited to go back to the genetic clinic to discuss the results. If the test shows that your deafness does not have a genetic cause, you won’t need to go to the clinic again.

Are all deafness genes tested?

Although doctors currently know about more than 80 genes for hearing at the moment, only a few are routinely tested for. This is because many genes are too large to be tested or they cause deafness in only a few people, and accurate testing has not yet been developed for these genes.

The main genetic test offered at present is a test to screen a gene called *GJB2* for mutations that alter the instructions for how to make the protein connexin 26. This protein is needed for hearing. Other genetic tests that you may be offered are as follows:

- If deafness within your family seems to be inherited from your mother, you may be tested for specific changes in mitochondrial DNA (see *What are mitochondrial genes?* on page 4).
- If doctors think you have a condition known as Pendred syndrome, of which deafness is a part, you may be tested for changes in the *SLC26A4* gene.

What are the benefits of genetic research into deafness?

In the future, many more genes needed for hearing may be discovered. The technology to find mutations in these genes will improve, becoming faster and more efficient. This will

make it possible to test for more genes than at present, and so improve the diagnosis and accuracy of information about your deafness and the chances of having children who are deaf.

The genes discovered so far mainly cause deafness in childhood or young adulthood. However, deafness in older people – hearing loss due to ageing or presbycusis – can have an inherited component too. Researchers are beginning to discover which genes contribute to hearing loss due to ageing. This knowledge, together with a better understanding of which genes make one person more at risk than another from factors such as loud noise, might make it possible to identify people at risk of losing their hearing. They may then be able to change their lifestyle to protect it.

Knowledge of the genes needed for hearing will also help researchers to understand more about how the ear works. It is hoped that this will lead to new treatments for protecting and improving hearing.

Where can I get further information?

British Society for Human Genetics (BSHG)

An independent body representing UK human genetics professionals with information for patients and a directory of clinical genetics centres.

Clinical Genetics Unit, Birmingham Women's Hospital, Birmingham B15 2TG

Telephone 0121 627 2634 Fax 0121 623 6971

bshg@bshg.org.uk www.bshg.org.uk

Genetic Alliance UK

A national alliance of organisations that supports children, families and individuals affected by genetic disorders. Their website has information about clinical genetic centres.

Unit 4d, Leroy House, 436 Essex Road, London N1 3QP

Telephone 020 7704 3141 Fax 020 7359 1447

mail@geneticalliance.org.uk www.geneticalliance.org.uk

National Deaf Children's Society (NDCS)

Supports all deaf children, young people and their families in overcoming the challenges of childhood deafness.

15 Dufferin Street, London EC1Y 8UR

Freephone helpline and textphone 0808 800 8880 Fax 020 7251 5020

helpline@ndcs.org.uk www.ndcs.org.uk

Further information from Action on Hearing Loss

Our helpline offers a wide range of information on many aspects of hearing loss. You can contact us for further copies of this factsheet and our full range of factsheets and leaflets – see the cover page for contact details.

Action on Hearing Loss Library

Our library has specialist publications ranging from academic journals to books for children. It is the largest library in Europe on deafness and hearing loss.
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Tel/ textphone 020 7915 1553 Fax 020 7915 1443
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