Genetics and deafness

What is genetics?

We all get or inherit characteristics, like the colour of our eyes and hair, from our parents. Some types of deafness can also be inherited.

Genetics looks at how these characteristics are inherited by studying genes.

What are genes?

Genes determine the characteristics we inherit from our parents. Every cell in our body has two copies of about 30,000 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.

a pair of chromosomes

Picture supplied courtesy of the North East London Regional Cytogenetics Laboratory.

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 many thousands of proteins which the cell needs to do its usual job.

What happens when the DNA sequence of a gene changes?

Sometimes the DNA sequence of a gene can change – scientists refer to this change 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.

Types of gene mutation

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

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. Even in the same family the 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.

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 deaf person.

If you have this type of deafness the chance of this type of mutation causing deafness in your children is far less 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 deaf person whose deafness is due to a recessive mutation may have hearing parents and may have deaf and hearing brothers and sisters, and no previous family history of deafness even though the deafness is genetic in origin.

Are men and women affected equally by gene mutations?

One type of chromosome, the sex chromosome, is different between men and women.

- In women, the pair is made of two X chromosomes.
- In men the pair is made up of an X and a Y chromosome.
- The Y chromosome is always inherited from the father and determines the male sex.

This means that men and women are affected in different ways by the presence of a gene mutation:

- If a woman has a recessive gene on one of her X chromosomes (an X-linked gene) she will be hearing because on her other X chromosome she has a normal copy of the same gene. She is a carrier and can pass on the affected gene to her children (see how in the example later in this section)
- If a man has a recessive gene on his X chromosome he will be deaf because his Y chromosome is responsible only for determining the male sex.

A female carrier of the X-linked recessive deafness gene will therefore have an equal chance of having children as follows, assuming that the father of the children has a normal copy of the X chromosome.

- A hearing son – who has a normal copy of the X-linked gene from his mother and a Y chromosome from his father.
- A deaf son – who has a mutated X-linked gene from his mother and a Y chromosome from his father.
- A hearing daughter who is a carrier – she has a mutated X-linked gene from her mother and a normal X chromosome from her father.
- A hearing daughter who is not a carrier – she has a normal X-linked gene from her mother and a normal X chromosome from her father.

If the affected gene is located on any other chromosome types other than the X chromosome then both sons and daughters have equal chances of inheriting deafness.

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 – though both men and women can have this type of deafness.

How common is genetic deafness?

Approximately one in 1600 children is born moderately to profoundly deaf because of a genetic cause.

Why genetic information may be helpful

Genetic information about deafness may be helpful.

- To find out the chances of having a deaf child if you are deaf or deafness runs in your family.
- To try and 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, a type of deafness 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 genetic counselling?

If you would like genetic information about your deafness, you will need to have a consultation in a genetic clinic. There are 30 NHS centres offering genetic counselling for deafness around the country. In order 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?

The consultation is often known as genetic counselling. 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) to discuss the facts about 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.
- They may ask you about your family history to understand how deafness is inherited in your family though you do not need to have a family history of deafness to have inherited deafness (see **Types of gene mutation**).
- They 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.
- They may look at your audiological records to help with the diagnosis.
- They may arrange for you to have other tests, for example special scans. These will help them to find out about problems in parts of your ear, such as the inner ear, and to exclude any common syndromes.
- They may suggest that you have a genetic test to help with the diagnosis.
- They may tell you how your deafness is inherited or why there may be uncertainty.
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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 find out 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 deaf child.

If you are deaf, you will be 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 are not tested to see if they are carriers unless they are old enough to give informed consent – this means they have to understand what they are agreeing to have done.

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 then 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 then you won't need to go to the clinic again.

Are all deafness genes tested?

Although doctors know about more than 50 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 being offered at present is a test to screen the gene for the 'connexin 26' protein. This protein is needed for hearing. Other genetic tests that you may be offered are as follows:

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

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 and become faster and more efficient. This will make it possible to test for more genes than at present and so improve the diagnosis and the accuracy of information about your deafness and the chances of having deaf children.

The genes discovered so far mainly cause deafness in childhood or young adulthood. However, deafness in older people – hearing loss due to ageing – can have an inherited component too. In the future, researchers will probably 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 of being affected by factors such as loud noise than another, might make it possible to identify people at risk of losing their hearing. They may then be able to change their lifestyle to protect their hearing.

Researchers also hope to understand the role of proteins needed for hearing. With this knowledge researchers hope to develop new treatments for improving hearing.

The NHS and genetics

The Department of Health recognises the potential benefits offered by genetic testing. The department's white paper, published in June 2003, set out how they plan to invest in the provision of NHS genetic services. The plans aim to make sure that this new area of medicine can benefit everyone and that the NHS can cope with increased demand in the future.

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Deaf Connexions produces a range of information sheets covering all aspects of hearing loss and deafness. If you would like further information contact :

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