

## Further developments

It is likely that in the future we will be able to test for alterations in more of the genes which commonly cause hearing impairment.

## For more information:

If you need more advice about any aspect of SNHL, you are welcome to contact:

### Clinical Genetics Departments

Northern Scotland (main base Aberdeen)  
Tel: 01224 552120 Fax: 01224 559390  
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)  
Tel: 01382 632035 Fax: 01382 645731  
(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)  
Tel: 0131 651 1012 Fax: 0131 651 1013  
(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)  
Tel: 0141 201 0808 Fax: 0141 201 0361  
(Glasgow, Argyll & Bute, Ayrshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

For support and advice, please contact:

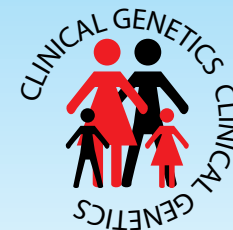
### The National Deaf Children's Society

15 Dufferin Street  
London  
EC1Y 8PD

Seen in clinic by.....

This leaflet was written by Guy's & St. Thomas' Clinical Genetics.  
Updated by Genetic Interest Group Scotland. Last updated Apr 2002.

# The Genetics of Sensorineural Hearing Loss (SNHL)



General information

## What is sensorineural hearing loss (SNHL)?

SNHL is deafness which is caused by problems that affect the inner ear (cochlea) or auditory nerve. There is another type of deafness - conductive deafness - that is caused by problems in the outer or middle parts of the ear.

## How common is SNHL?

About 1 in 1000 children is either born deaf or develops profound hearing loss in early childhood. A further 1 in 1000 becomes deaf before adulthood.

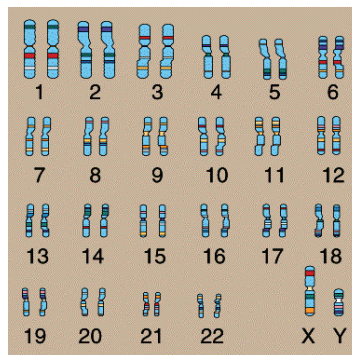
## What causes SNHL?

About one-third of cases of SNHL occur as a result of a baby being born prematurely or suffering from an infection such as mumps or meningitis. Of the remaining cases, the majority are caused by a genetic alteration.

## What are genes and chromosomes?

Our genes can be thought of as a set of instructions for building and maintaining our bodies. There are around 30,000 different genes altogether and we have 2 copies of each one. They are made of a chemical called DNA and contained inside larger structures called chromosomes which are found in every cell in the body.

Most people have 23 pairs of chromosomes (46 in total). One of each pair comes from the mother and the other from the father. Chromosomes are numbered 1 (the largest pair) to 22 (the smallest pair); the 23rd pair are called sex chromosomes because they determine whether a person is male or female.



## Which genes cause sensorineural hearing loss?

There are at least 80 different genes which are important for normal hearing. These genes are carried on many different chromosomes. An alteration in any one of these genes can cause deafness.

## How is SNHL inherited within families?

### Recessive Genes

Most cases of SNHL are caused by alterations in "recessive" genes.

We have two copies of every gene, one that we get from our mother and the other that we get from our father. Everyone carries a few genes which have an alteration (spelling mistake) in the DNA code.

Usually we never find out about these because we also have a working copy of the same gene. If, by chance, both parents carry one altered copy of the same deafness gene, then there is a 1 in 4 chance of both of them passing on the altered copy to each of their children. A child who receives two altered copies of the gene will have hearing impairment.

### Dominant Genes

Deafness can also be caused by alterations in "dominant" genes. A dominant gene is one in which an alteration in just one copy of the gene is enough

to cause problems. The second copy of the gene is unable to compensate. If neither parent is deaf, it is most likely that the alteration in the gene arose for the first time in the affected child. If there is no family history of deafness, we usually cannot tell whether a child's hearing impairment is due to an alteration in a recessive or a dominant gene.

## What is the chance of another child being born with a hearing impairment?

If there is no family history of hearing impairment, the chance of a couple who already have one child with hearing impairment having another deaf child is 1 in 8 (about 12 percent).

## What is the chance of someone with hearing impairment having a deaf child?

This depends on whether or not both parents are hearing impaired. If one partner has normal hearing, the chance is around 1 in 20 (5 percent). Couples where one or both partners have hearing impairment should be offered genetic counselling if they are concerned about the chance of having a hearing impaired child.

## Genetic Tests

At present we are only able to look for alterations in one of the many genes known to cause hearing impairment. This gene is called Connexin 26. We think that alterations in Connexin 26 account for 30 percent of cases of recessive deafness. We only manage to find an alteration in the Connexin 26 gene in about 1 in 10 children with severe or profound hearing impairment. If we do find an alteration in the Connexin 26 gene, the chance of that child's parents having another child with SNHL is 1 in 4 (25 percent).