Genetic tests
Routine genetic testing looks for mutations in two of the many genes known to cause deafness. These genes are called Connexin 26 and Connexin 30.

Alterations in these two genes are the most common cause of recessive deafness.

If we do find an alteration in both copies of either the Connexin 26 or Connexin 30 gene, the chance of that child's parents having another child with SNHL is 25% (1 in 4).

Future developments
It is likely that in the future it will be possible to test for alterations in further genes which are known to cause deafness.

Please discuss this with your genetic counsellor or doctor in the Genetics Clinic.

Further information
National Deaf Children's Society (NDCS)
Helpline: 0808 800 8880
Minicom: 0808 800 8880
Mon to Thurs, 9.30am - 9.30pm
Fri 9.30am - 5pm
Email: helpline@ndcs.org.uk
www.ndcs.org.uk

Feedback
We appreciate and encourage feedback. If you need advice or are concerned about any aspect of your care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From a mobile or abroad:
0115 924 9924 ext. 65412 or 62301
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS,
Freepost NEA 14614,
Nottingham NG7 1BR

www.nuh.nhs.uk

If you require a full list of references for this leaflet please email patientinformation@nuh.nhs.uk or phone 0115 924 9924 ext. 67184.

The Trust endeavours to ensure that the information given here is accurate and impartial.

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Elizabeth King, Clinical Genetics © August 2018. All rights reserved. Nottingham University Hospitals NHS Trust. Review August 2020. Ref: 1034/v3/0818/AH.
What is sensorineural hearing loss (SNHL)?
SNHL is deafness caused by problems that affect the inner ear (cochlea) or the auditory nerve. There is another type of deafness - conductive deafness - that is caused by problems in the outer or middle parts of the ear.

About one in 1,000 children is either born deaf or develops profound hearing loss in early childhood. A further one in 1,000 individuals becomes deaf before adulthood.

What causes SNHL?
About a quarter of SNHL cases occur as a result of a baby being born prematurely or suffering from an infection such as mumps or meningitis. Half of the SNHL cases presenting in childhood are due to a genetic cause.

What are genes and chromosomes?
Genes are instructions for specific functions in the body and also make each of us a unique individual. There are many thousands of different genes, each carrying a different instruction. If a gene is altered (or 'mutated'), it may not work correctly.

Genes are present on tiny structures called chromosomes. Humans have 46 chromosomes in each cell. All chromosomes come in pairs; we inherit one copy from each parent. Therefore we have two copies of every gene; and inherit one copy from each parent.

Which genes cause SNHL?
There are many different genes which are important for normal hearing. An alteration in any one of these genes can cause deafness.

SNHL can sometimes be a part of a wider genetic condition or ‘syndrome’ in an individual who usually has other medical and/or learning problems. The information in this leaflet relates only to isolated or ‘non-syndromic’ SNHL.

How is SNHL inherited within families?
Recessive Inheritance
About 80% of cases of early onset SNHL are caused by alterations being inherited in a ‘recessive’ pattern. 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 an altered copy of the same deafness gene, then there is a one in four chance of both of them passing on the altered copy to each of their children. A child who has both altered copies of the gene will have hearing impairment.

Dominant Inheritance
Deafness can be inherited in a ‘dominant’ inheritance pattern. In this situation an alteration in just one copy of the gene is enough to cause problems. The second, healthy copy of the gene is not able to compensate.

X-linked Inheritance
There are some genes on the X chromosome that are known to cause hearing loss in boys. Girls have two X chromosomes and if they have an altered copy of the deafness gene on one X chromosome, they may not have hearing loss, as the other copy can compensate. However, they can have a son with X-linked deafness.

If there is no family history of deafness, we cannot predict accurately whether a child’s hearing impairment is due to an alteration in a recessive, dominant or X-linked 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 with normal hearing who already have one child with hearing impairment having another deaf child is 18% (about 1 in 5).

What is the chance of someone with a 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 10% (1 in 10). Couples where one or both partners have hearing impairment should be offered genetic counselling if they want to discuss the chance of having a hearing impaired child.