# Fact Sheet
# The inheritance of deafness



This fact sheet is designed to help parents understand about "genetic" and "inherited" deafness. Many parents who have one deaf child want to know about the chances of having another deaf child. It is natural for parents of a child to experience a period of sadness after they discover that their child has a minor or major permanent disability. Parents of a deaf child quickly realise how much more time and work is needed to look after this child compared to a normal hearing child.

## Could I have another deaf child?

The answer to this question is often yes. Before a doctor warns a family that the hearing loss in their child is probably genetically based (passed on by the parents and their families) he or she usually spends some time examining the child, checking the family history and doing tests to pick up other causes of deafness such as congenital rubella (German measles) before birth. It is uncommon to find unexpected environmental causes of deafness. If there are no particular reasons for the deafness it is usually genetic deafness. This means it was passed on through the family genes.

## Are there different types of genetic (hereditary) deafness?

There are a number of different types of hereditary deafness. It is probably most helpful to divide it into two categories:

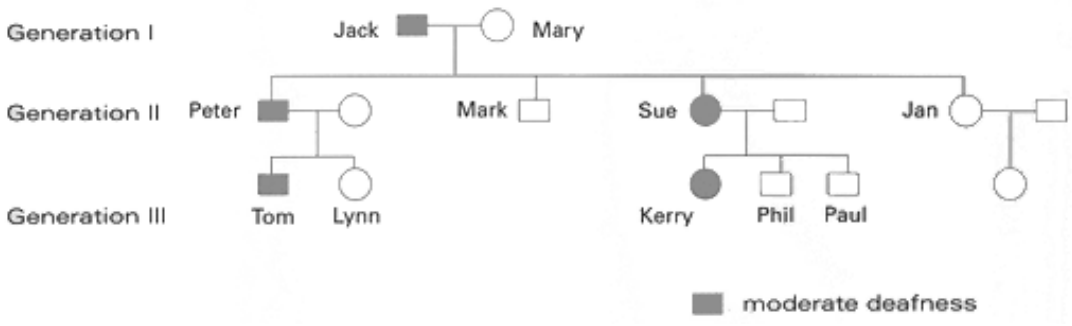
1. dominant deafness
2. recessive deafness.

## What is autosomal dominant deafness?

This is the hereditary type of deafness where parents can usually see it being handed on from one generation to the next.

Fig.1 is an example of a family where deafness has been inherited with a dominant pattern. Grandparent Jack is moderately deaf. Of his children, Peter and Sue are moderately deaf, and in the third generation, Tom and Kerry have moderate deafness. For a deaf person with dominant deafness there is usually a one-in-two chance he or she will hand this on to any child. This chance is the same for every pregnancy. Parents with dominant deafness usually have their children's hearing tested as young babies. Then hearing aids can be fitted early if needed and this helps children with language development. Without the hearing aids a child with deafness may not learn to speak.

Fig.1 Autosomal dominant deafness



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## What is recessive deafness?

In this type of genetic deafness there may be no family history of deaf relatives and yet one or more children are deaf for genetic reasons. There are two types of recessive deafness:

- autosomal recessive - affects boys and girls
- X-linked recessive - affects only boys.

### Autosomal recessive deafness

Fig. 2 is an example of autosomal recessive deafness. In this family Terry and Karen have good hearing as does Terry's brother and Karen's two sisters. However, Craig and June have severe deafness. Many parents find this surprising, but the explanation is as follows:
All the genetic information we pass on to our children is contained in complex deoxyribonucleic acid (DNA) strands packaged up as chromosomes which are in the nuclei of all human cells. The X and Y chromosomes, also known as sex chromosomes, decide if a baby is a boy or a girl. Autosomes are also chromosomes but they are different because they do not determine a child's sex. There are:

- 22 pairs of autosomes plus two X chromosomes in female cells, and
- 22 pairs of autosomes and one X and one Y chromosome in male cells.

Genes are arranged on the DNA strands in an orderly sequence. The individual genes are too small to be seen but the chromosomes can be seen under the microscope. Genes determine personal characteristics such as hair colour, eye colour, height and even whether we get certain disorders.

A baby starts when an egg cell from the mother and a sperm cell from the father join together to make an embryo that grows in the mother's womb. The egg cell contains:

- 22 single autosomes and one X chromosome (22 autosomes + X).

The sperm cell contains:

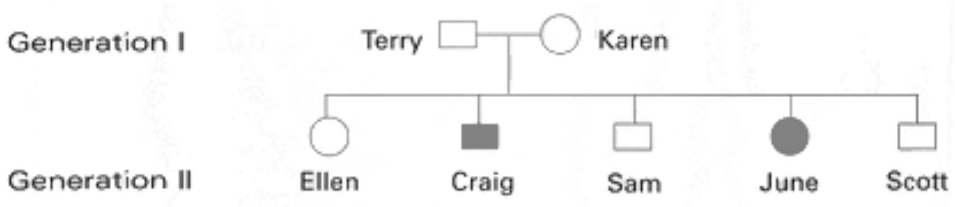
- 22 single autosomes and one X or one Y chromosome (22 autosomes + X) or (22 autosomes + Y).

Adding up the total chromosomes for the baby makes it either:

- 44 autosomes + X + Y for a male baby or
- 44 autosomes + two X for a female baby.

It is the father's sperm that determines the sex of a baby.

Fig. 2 – Autosomal recessive deafness



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Just as chromosomes come in pairs so do the genes. Each chromosome of a pair has the same gene in the same place. A dominant gene (H) always has an effect on the child but a recessive gene (h) only produces its effect if there is no dominant gene to over-ride it. It is easier to understand if you think of the dominant gene as bigger and louder so it blocks out the other (recessive) gene from having an effect. Many cases of deafness are caused by recessive genes. The child inherits one recessive gene (h) from each parent. Usually the parents have normal hearing as they have one dominant gene (H) for normal hearing. Each carrier parent has a profile Hh. This means they carry a bigger gene for hearing and a smaller gene for deafness, so the strong H (dominant) gene for normal hearing wins and the parent does not have deafness.

As you can see in Fig. 3 there are four possible ways in which genes can be handed on to a baby. This couple have a one-in-four chance of having a deaf child in every pregnancy. When a child is deaf but otherwise normal and a careful check reveals no other cause, the family is advised that the deafness could be autosomal recessive in origin.

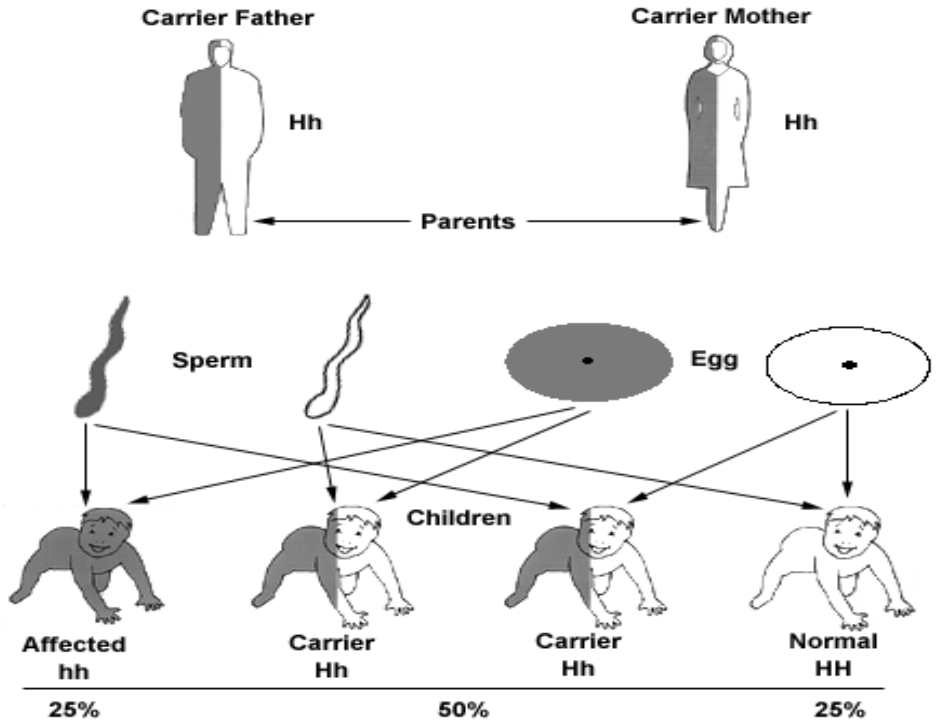
In some families who have a healthy deaf child we may suspect (but not be able to prove) that the child has an autosomal recessive form of hearing loss. When deafness appears "out of the blue" and we cannot establish a cause the recurrence risk for a future child to be born deaf is given as approximately one in six. The lower risk is because occasionally children are born deaf due to new mutations and also because there may possibly be unrecognised environmental causes of deafness is some children.

Unfortunately, a one-in-six figure is still quite a high risk, just as high as scoring a six each time you throw a dice.

### What is X-linked recessive deafness?

This type of recessive deafness is very uncommon and only affects boys. As with haemophilia, the gene for this type of deafness is on the X chromosome and is passed on to a son by a carrier mother. One-in-two (half) of her sons will be deaf.

Fig.3 – How autosomal recessive deafness is transmitted



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### What is Mitochondrial deafness?

This type of deafness also appears to be very uncommon .Children with this type of deafness often have extra problems such as in muscle or skin. It is caused by defects in genes in the mitochondria which are tiny organelles in the cytoplasm (not the nuclei) of human cells which are responsible for controlling the generation of energy in cells. In children with mitochondrial gene defects we often see transmission of problems from mother to all the children, in varying degrees.

## Should we have more children and when?

Parents have the right to make the decision about future children. Having a deaf child usually means a great deal more work and anxiety for parents, especially if they know that they have a high chance of having another deaf child in another pregnancy. Many parents wish to delay the decision to have another child until they are confident and relaxed about the speech, language and general progress of their deaf child. They want to have plenty of time to work with the new baby if he or she also turns out to be deaf.

## Can blood tests tell if you are a carrier for deafness?

The answer to this question is usually, but not alway, no. There are at least 50 different types of recessive deafness. However, there is one type of autosomal recessive deafness that is much commoner than the other types. Children with this type of deafness can usually be identified by a blood test (DNA test) which checks for errors in the connexin 26 gene. This test is available (when indicated) in some Genetics units and Children's Hospitals.

## What is the chance that my deaf child will have a deaf child later on?

If your son or daughter is deaf and the reason has not been definitely found, his or her chance of having a deaf child later on is fairly low (but it could still happen).

Some books have given a figure of risk about one-in-twenty, but it is not really known. However, if two people who are known to have deafness marry, the chance that they will have deaf children is significant.

## What is the chance that the child's brother or sister could have a deaf child later on?

Provided that it has been found that the sibling has normal hearing, his/her chance of bearing a deaf child is a low risk, although it is slightly higher than that of a person from a family with no history of deafness.

In families with X-linked recessive deafness, all the daughters of affected males will be carriers. There is a one-in-four chance that male grandchildren will be deaf.

## We have one deaf child. If we decide to have another child and it is also deaf, will the deafness be similar to that of our first deaf child?

Yes, often, but there is no guarantee. Occasionally, for example, in a family with some moderately deaf members there can be one who is severely affected. Remember one child (with hearing aids) can find a moderate degree of deafness a minor problem and another child with an almost identical audiogram (hearing test) may find it a much bigger problem.

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The answers to these questions are "2006 answers". The answers may be different, better and clearer in 2008 or 2010, as genetics is a science undergoing an explosion of knowledge and discovery at present. It will be wise for your deaf child and his siblings to ask the same questions again when they are young adults.

Most states have genetic education programs which provide additional information and resources concerning genetic disorders and the availability of genetic counselling clinics.

## Where further information can be obtained from in NSW

**Genetic Education Program**
PO Box 317
St Leonards NSW 2065
Tel: (02) 9926 7324 / (02) 9906 7529

Genetics Clinics are available in a number of metropolitan and rural centres. You can find the location of the one closest to you by ringing the Genetics Education Program at the above number. At present Genetics Clinics are held at:

### Sydney and Newcastle area

- **Camperdown, Royal Prince Alfred Hospital**
  Tel: (02) 9515 5080
  Fax: (02) 9515 7595
- **Kogarah, St George Hospital**
  Tel: (02) 9350 3635
  Fax: (02) 9350 3901
- **Liverpool Health Services**
  Tel: (02) 9828 4665
  Fax: (02) 9828 4650
- **Newcastle, Hunter Genetics**
  Tel: (02) 4985 3100
  Fax: (02) 4985 3105
- **Penrith, Nepean Hospital**
  Tel: (02) 4734 3362
  Fax: (02) 4734 2567
- **Randwick, Sydney Children's Hospital**
  Tel: (02) 9382 1704
  Fax: (02) 9382 1711
- **St Leonards, Royal North Shore Hospital**
  Tel: (02) 9926 6478
  Fax: (02) 9926 7880
- **Westmead, The Children's Hospital at Westmead**
  Tel: (02) 9845 3273
  Fax: (02) 9845 3204

### Country

- **Bathurst Community Health Centre**
  Tel: (02) 6339 5677
  Fax: (02) 6332 2039
- **Broken Hill Community Health Centre**
  Tel: (02) 8080 1554
  Fax: (02) 8080 1611
- **Canberra**
  Tel: (02) 6244 4042
  Fax: (02) 6282 2844
- **Coffs Harbour Health Campus**
  Tel: (02) 6656 7000
  Fax: (02) 6656 7687
- **Forster Community Health Centre**
  Tel: (02) 6555 6822
  Fax: (02) 6554 8874
- **Gosford, Central Coast Genetic Counselling Service**
  Tel: (02) 4337 0207
  Fax: (02) 4337 0217
- **Goulburn, Child Development Unit**
  Tel: (02) 4827 3951
  Fax: (02) 4827 3958
- **Kempsey**
  Tel: (02) 6588 2882
  Fax: (02) 6588 2800

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- **Mudgee Community Health Centre**
  Tel: (02) 9378 6236
  Fax: (02) 6372 7341
- **Muswellbrook Community Health Centre**
  Tel: (02) 6542 2050
  Fax: (02) 6542 2005
- **North Coast, Child and Family Health Centre**
  Tel: (02) 6625 0111
  Fax: (02) 6625 0102
- **Port Macquarie Community Health Centre**
  Tel: (02) 6588 2882
  Fax: (02) 6588 2800
- **Tamworth Community Health Centre**
  Tel: (02) 6766 2555
  Fax: (02) 6766 3967
- **Taree, Mid North Coast Area Health**
  Tel: (02) 6592 9703
  Fax: (02) 6592 9607
- **Wagga Wagga Base Hospital**
  Tel: (02) 6938 6666 (switch)
  Tel: (02) 6938 6443 (direct)
  Fax: (02) 6921 5632
- **Wollongong, Genetic Counselling Service**
  Tel: (02) 4253 4267
  Fax: (02) 4253 4257

## Remember

- You should look for the cause of your child's deafness by consulting your family doctor, medical specialists or geneticists.
- If you cannot find a reason for your child's deafness it may be caused genetically.
- Genetic hearing losses often occur again in other children.



***This document was reviewed on Thursday, 1 June 2006.***

the children's hospital at Westmead | SYDNEY CHILDREN'S HOSPITAL RANDWICK | Kaleidoscope HUNTER CHILDREN'S HEALTH NETWORK

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