FACTSHEET

The inheritance of hearing loss and deafness

This fact sheet is designed to help parents understand about "genetic" and "inherited" hearing loss. Many parents who have a child with a hearing loss want to know about the chances of having another child with a hearing loss.

Could I have another child with hearing loss?

The answer to this question is often yes. Before a doctor tells a family that the hearing loss in their child is inherited (passed on by the parents and their families) he or she usually spends some time examining the child, asking about the family medical history and doing tests to identify other causes of hearing loss. In about 50% of individuals who have hearing loss, it is due to a genetic cause. This means it was passed on through the family genes.

Are there different types of genetic (hereditary) hearing loss?

There are a number of different types of hereditary hearing loss. It is probably most helpful to divide it into:

1. Autosomal recessive hearing loss
2. Autosomal dominant hearing loss
3. Less common forms of inherited hearing loss including X-linked and mitochondrial hearing loss

What is autosomal recessive hearing loss?

In this type of genetic hearing loss there may be no family history of relatives with hearing loss. This is the most common form of inherited hearing loss. The chance of it is increased when the child's parents are closely related (known as consanguinity).

Autosomal recessive hearing loss

Fig. 1 is an example of autosomal recessive hearing loss. 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 hearing loss. 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 called genes which are packaged into chromosomes in all human cells. 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. There are 23 pairs of chromosome in each cell – 22 numbered pairs (called autosomes) that are the same in men and women and one pair of sex chromosomes - The X and Y chromosomes, which determine if a baby is a boy or a girl.

Unlike most cells in the body, the gametes (egg cells for women, sperm cells for men) contain a half set of chromosome and therefore genes. When the egg and sperm unite to form a pregnancy the resulting baby again has a pair of each chromosome number with one from each parent. Just as chromosomes come in pairs so do the genes. Each chromosome of a pair has the same gene in the same place. The baby inherits one copy of each gene from the mother and one copy from the father. It is the father's sperm cell which determines the sex of a baby – male if Y chromosome in sperm or female if X chromosome in the sperm.

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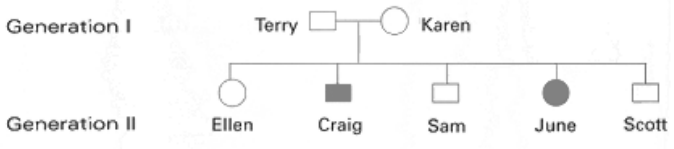
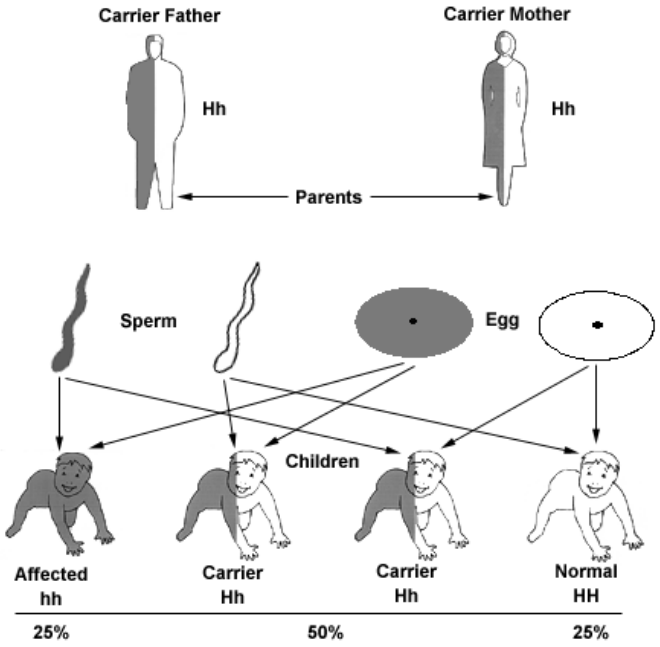
Fig. 1 – Autosomal recessive hearing loss



A dominant gene (H) has an effect on the child but a recessive gene (h) effect will be hidden unless it is present in both copies of the gene pair. Many cases of hearing loss 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 working copy of the gene (H) for normal hearing in addition to the hearing loss copy of the gene (h). Each carrier parent has a profile Hh.

As you can see in Fig. 2 In families with autosomal recessive hearing loss, there are four possible ways in which genes can be passed on to a baby. This couple have a one-in-four chance of having a child with hearing loss in each pregnancy.

Fig. 2 - How autosomal recessive hearing loss is transmitted



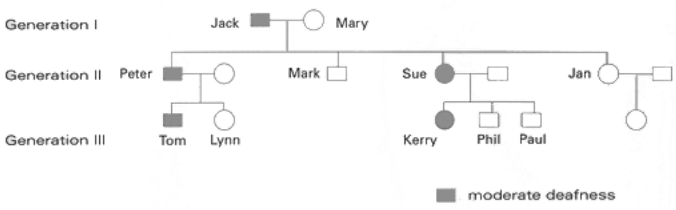
In some families who have a child with a hearing loss we may suspect but not be able to prove that the child has an autosomal recessive form of hearing loss. When a hearing loss appears “out of the blue” and we cannot establish a cause, the risk for a future child to have a hearing loss is given as approximately one in six. The lower risk is because occasionally children have a hearing loss due to new mutations (changes in the genes which starts with them) and also because there may possibly be an unrecognised environmental (non-genetic) cause of hearing loss.

What is autosomal dominant hearing loss?

In this type of genetic hearing loss, there may be more than one family member with hearing loss.

Fig.3 is an example of a family where hearing loss has been inherited with a dominant pattern. Grandparent Jack has a moderate hearing loss. Of his children, Peter and Sue have a moderate hearing loss, and or the grandchildren, Tom and Kerry have a moderate hearing loss. For a person with dominant hearing loss, there is usually a one-in-two (50%) chance he or she will hand this on to any child. This chance is the same for every pregnancy.

Fig. 3 - Autosomal dominant hearing loss



What is X-linked recessive hearing loss?

This type of recessive hearing loss is very uncommon and only affects boys. The gene for this type of hearing loss is on the X chromosome and is passed on to a son by a carrier mother. One-in-two (50%) of her sons will have hearing loss.

What is mitochondrial hearing loss?

This type of hearing loss also appears to be uncommon. Children with this type of hearing loss 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.

We have one child with hearing loss. If we decide to have another child and he or she also has hearing loss, will the hearing loss be similar to that of our first child?

Often the level of hearing loss is similar between family members but there is no guarantee. Occasionally, in a family with some members with moderate hearing loss there can be someone who has a severe hearing loss. Remember one child (with hearing aids) can find a moderate degree of hearing loss a minor problem and another child with an almost identical level of hearing may find it significant challenge.

Should we have more children and when?

This is a decision that will be different for each family. It can be helpful to speak with a genetic counsellor at a genetic

clinic. Genetic clinics in New South Wales are listed at the end of this factsheet.

Can blood tests tell if you are a carrier for hearing loss?

There are at least 50 different types of recessive hearing loss. However, there are 2 types of autosomal recessive hearing loss for which testing is readily available i.e. connexin 26 and connexin 30 hearing loss. These types of hearing loss can be identified using DNA obtained from a blood sample. This test may be offered in Genetic clinics and children's hospitals after genetic counselling. For these particular types of hearing loss, a carrier can also be identified by a blood test.

What is the chance that my child will have a child with hearing loss?

If your son or daughter has hearing loss and the reason has not been found, his or her chance of having a child with hearing loss is fairly low but it could still happen.

Some people estimate that the possibility is about one-in-twenty (5%), but it is not really known. If two people who have hearing loss have a child, the chance that they will have children with hearing loss is likely to be higher as both the parents may have deafness from genetic causes

What is the chance that the child's brother or sister could have a child with hearing loss?

Provided that it has been found that the brother or sister has normal hearing, his/her chance of having a child with hearing loss is low, although it is higher than that of a person from a family with no history of hearing loss. A genetic counsellor may be able to provide more precise information about this issue.

The answers to these questions are based on current knowledge. The answers may be different, better and clearer in the future, as genetics is a science undergoing an explosion of knowledge and discovery. It may be useful for your children 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 clinics.

Remember:

- You can look for the cause of your child's hearing loss by consulting your family doctor, paediatrician, ENT surgeon or geneticists.
- If the cause of your child's hearing loss cannot be found, it may be genetic.
- Genetic hearing losses can occur in future children.
- Genetic counselling is available from genetic clinics.

Where to get further information from in NSW

The Centre for Genetics Education

Royal North Shore Hospital Community Health Centre

Level 5, 2c Herbert Street
St Leonards NSW 2065

Tel: (02) 9462 9599

Fax: (02) 9906 7529

Email: contact@genetics.edu.au www.genetics.edu.au

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:

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

For information on regional centres, visit the Centre for Genetics Education program website: www.genetics.edu.au or call (02) 9462 9599