Important points

- Hearing loss due to either
  - Conductive hearing loss - structural problems within the ear affecting hearing
  - Sensorineural hearing loss - problems with the tiny sensory hair cells in the cochlea, or the auditory nerve, or in the processing of sound in the brain
- Onset can be
  - Before speech is usually present (pre-lingual hearing loss). All hearing loss that is present at birth (congenital) is pre-lingual, but not all pre-lingual hearing loss is congenital
  - After the usual time of speech development (post-lingual hearing loss)
- In about 50% of individuals who are deaf, the cause is a combination of genetic and environmental influences (such as infection, noise, age)
- In about 50% of individuals who are deaf, their genetic make-up alone appears to account for deafness. ‘Genetic deafness’ may be due to changes in a single gene, or a combination of genes
  - In about 1/3 of cases or 30% of the time, it occurs as part of a genetic condition (syndromic deafness) where deafness is just one of a number of symptoms. There are over 400 different syndromes involving hearing loss, so each of the syndromes associated with the hearing loss is individually relatively rare
  - In about 2/3 of cases or 70% of the time, deafness occurs by itself (non-syndromic)
- Over 100 different genes have been identified that when faulty directly cause or are associated with non-syndromic genetic deafness. There are still many other genes not yet identified
  - The pattern of inheritance depends on the type of change causing the gene to be faulty, and its chromosomal location
- Within the pre-lingual non-syndromic hearing loss group that is genetic, 75%-80% of cases follow a pattern of autosomal recessive inheritance in families
  - Where both parents are unaffected carriers of the faulty gene involved, there is a 25% chance in every pregnancy that their child will be affected
  - The most common gene involved in this group is called connexin 26 (GJB2) but there are many others
- Genetic counselling may help to clarify whether an individual’s deafness has been inherited, or is due to environmental factors
  - Genetic testing is available for connexin 26-associated deafness and for a few other forms of genetic deafness

A number of different terms are used to describe a condition in which people have difficulty either perceiving or understanding sounds (auditory information).

- ‘Hearing impairment’ and ‘hearing loss’ are often used interchangeably to refer to hearing shown by hearing tests (audiometry) to be below levels for normal hearing
- ‘Deafness’ (starts with a small ‘d’). A commonly used term that implies hearing levels in the severe-to-profound range determined by audiology
- Deaf culture (always starts with a capital ‘D’) is used to describe members of the Deaf community. In Australia, members of the Deaf community communicate using sign language called Auslan, which stands for AUStralian Sign LANGUAGE, and belong to a distinct cultural group

Hearing loss is a common condition in the community

- More than 1 in every 10 Australians report long-term hearing problems
- About 1 in 1000 babies are born with a significant degree of hearing loss

The chance of a person developing hearing loss increases with age

  - Some people develop hearing loss during childhood and others later in life
  - By the time a person is 65 years old there is about a 1 in 3 chance they will have some kind of problem with their hearing

The hearing system

The ear has three main parts: the outer, middle, and inner ear (see Figure 60.1).

Sound waves enter through the outer ear and reach the middle ear, where they cause the eardrum to vibrate.

The vibrations are transmitted through three tiny bones in the middle ear called the ossicles. These three bones are named the malleus, incus, and stapes (and are also known as the hammer, anvil, and stirrup respectively). The ear drum and ossicles carry the vibrations to the inner ear. The stirrup transmits the vibrations through the oval window and into the fluid that fills the inner ear. The vibrations move through fluid in the snail-shaped organ called the cochlea. Tiny hair cells in the cochlea pass information to the auditory nerve that sends signals to the brain where sound is processed.
What is deafness?

Hearing is measured in decibels (dB). The severity of the hearing loss is graded as:
- Mild (26-40 dB)
- Moderate (41-55 dB)
- Moderately severe (56-70 dB)
- Severe (71-90 dB)
- Profound (90 dB+)

Deafness may:
- Remain the same throughout an individual’s lifetime (stable hearing loss)
- Change from time to time (fluctuating hearing loss)
- Become more severe over time (progressive hearing loss)

Onset of hearing loss can be:
- Before speech is usually present (pre-lingual hearing loss). All hearing loss present at birth (congenital) is pre-lingual, but not all pre-lingual hearing loss is congenital
- After the usual time of development of normal speech (post-lingual hearing loss)

Deafness commonly affects both ears (bilateral hearing loss) but sometimes it only affects one ear (unilateral hearing loss). People with deafness may be able to hear some sounds clearly, and other sounds not as clearly. For others, sound may seem too loud, too soft, muffled or difficult to understand.

Those with a mild or moderate hearing loss may not have any trouble with hearing in quiet surroundings but experience difficulty following conversations in noisy environments. Some severe or profoundly deaf people cannot hear sounds and communicate by lip-reading and/or sign language. Some people with deafness, especially those who have had a high degree of hearing loss from a young age, may access specific interventions to help them learn to speak.

What are the different types of deafness?

Deafness can be categorised according to which part of the ear is affected.

Conductive hearing loss

Sometimes deafness is due to structural changes in the ear which prevent sound from travelling to the auditory nerve, including blockages in the ear canal and problems with the tiny bones (ossicles) in the ear.

For example, otosclerosis is caused by a problem in the growth of the bones of the middle ear. It usually affects the last bone in the chain, the stapes and prevents structures within the ear from working properly thus causing hearing loss. For some individuals with otosclerosis, the hearing loss may become severe.

Conductive deafness can sometimes be corrected by surgery or other interventions.

Sensorineural hearing loss

Also known as nerve deafness is much more common than conductive deafness. Most commonly, this type of hearing loss is due to a problem with the tiny sensory hair cells in the cochlea, the auditory nerve or the processing centre in the brain.

Generally sensorineural hearing loss is permanent, but hearing aids or cochlear implants can help.

What causes deafness?

Deafness may be caused by factors in the environment, a combination of genetic and environmental factors or genetic factors alone (see Figure 60.2).

Environmental factors and environmental/genetic interactions

In about 50% of cases of deafness, the cause is either exposure to an environmental factor alone or a combination of both environmental and genetic susceptibility including:
- Exposure to loud noises such as music or industrial machinery (industrial deafness). This is sensorineural hearing loss in which the tiny sensory hair cells in the ear are permanently damaged
- Exposure to infections such as rubella and cytomegalovirus (CMV) virus during pregnancy may cause a baby to be born with deafness and bacterial meningitis can cause deafness during childhood
- Use of drugs which are ‘toxic’ to the ear (ototoxic drugs) which can cause deafness in adults and children
- Head injuries
- Ageing-deafness which commonly occurs in older people is known as presbycusis, and is often considered a natural part of the ageing process

It is now thought that an individual’s genes play a part in increasing susceptibility to industrial deafness, ototoxic drugs or presbycusis. That is, the deafness has been caused by a combination of both environmental and genetic factors (see Genetics Fact Sheet 11 for further information about environmental and genetic interactions).

Genetic factors

The individual’s genetic make-up alone appears to account for deafness in around half of all those affected by the condition (genetic deafness). If an environmental cause cannot be found for an individual’s deafness, then the deafness is usually genetic.

‘Genetic deafness’ may be due to variations in a single gene, or to the interaction of information produced by a number of different genes.
- Usually occurs by itself, without other symptoms (non-syndromic)
- In about 1/3 of cases or 30% of the time, it occurs as part of a genetic condition (syndromic) where deafness is just one of a number of symptoms (see Figure 60.2)

Syndromic genetic deafness

A clinical genetics consultation can help to identify whether or not syndromic hearing loss is present. There are over 450 different syndromes involving hearing loss, so each of the syndromes associated with the hearing loss is individually relatively rare.

Just one of the many types of syndromic hearing loss is Waardenburg syndrome.
- People with Waardenburg syndrome inherit sensorineural hearing loss with changes in the pigmentation (colour) of the skin, hair and eyes. They may have a white forelock of hair, eyes with multi-coloured irises, or two differently coloured eyes
- Waardenburg syndrome follows a pattern of autosomal dominant inheritance in families and affects about 1 in 4000 people (see Genetics Fact Sheet 9 for a discussion of autosomal dominant inheritance)
Non-syndromic genetic deafness

The cells of the body contain the genes or set of instructions for the cell to make all the necessary proteins (chemicals) for our bodies to grow and work normally (see Genetics Fact Sheet 1).

If a gene is changed so that it does not work properly, the gene is described as being faulty (ie. there is a gene mutation present). The information contained in the product of the faulty gene is impaired, or is not produced in the right amounts (see Genetics Fact Sheets 4 & 5).

Over 100 different genes have been identified as directly causative, or associated with non-syndromic genetic deafness. There are still many other genes not yet identified. For example, no genes have yet been identified for familial otosclerosis, although seven possible locations for causative genes have been described.

The pattern of inheritance of non-syndromic deafness in families depends upon which faulty gene is involved, whether the faulty gene is located on a chromosome in the nucleus of the cell or located in the mitochondrial DNA, and whether one or both copies of the gene need to be faulty for the hearing loss to occur.

More than 50% of pre-lingual deafness is genetic. As shown in Figure 60.2, within the pre-lingual non-syndromic hearing loss group that is genetic.

1. In about 1% of non-syndromic genetic deafness, hearing loss is due to faulty genes that are located on the X chromosome, so that males are more likely to be affected than females, or deafness is the mitochondrial See Genetics Fact Sheets 10 and 12 for a discussion of X-linked recessive inheritance and mitochondrial pattern of inheritance, respectively

2. About 20% to 25% of cases follow a pattern of autosomal dominant inheritance in families (see Genetics Fact Sheet 9) and occur when

1. The faulty gene copy is located on an autosome (one of the numbered chromosomes)

2. The effect of the change in the faulty gene copy is ‘dominant’ over the information in the working copy of the gene on the partner chromosome (see Genetics Fact Sheets 1, 4 & 5)

In these cases several family members are likely to be affected, although the severity of hearing loss may vary between different family members.

- When a parent has deafness or hearing loss due to an autosomal dominant faulty gene copy, there is a 1 chance in 2 (ie. 2 chances in 4) or 50% chance in every pregnancy that their child will inherit the copy of the faulty gene. The child has a high chance of being affected by hearing loss at some time in their life

- A single gene responsible for the majority of cases of autosomal dominant non-syndromic hearing loss, has not been identified

More than 3/4 or 75% to 80% of non-syndromic genetic deafness cases follow a pattern of autosomal recessive inheritance (see Genetics Fact Sheet 8) and occurs when:

1. The faulty gene copy is located on an autosome (one of the numbered chromosomes)

2. The effect of the change in the gene is ‘recessive’ or hidden by the presence of the working copy of the gene (see Genetics Fact Sheets 1, 4 & 5)

With autosomal recessive inheritance there is often no family history of deafness, so that parents often do not suspect that the cause of deafness in their child is genetic

- About 50% of the time, sensorineural pre-lingual deafness that follows a pattern of autosomal recessive inheritance in families is caused by changes in the gene called GJB2 which contains the information for the body to produce the protein called connexin 26 – see figures 60.3 and 60.4
The autosomal recessive pattern of inheritance in families with the faulty connexin 26 protein

We all have two copies of the GJB2 gene (often referred to as the Cx26 gene), which contains the information for the body to produce the protein called connexin 26. This protein has an important role in enabling sound to travel through the ear. When the connexin 26 protein is faulty, problems occur with how the sound is transported, causing deafness.

An individual who has one faulty Cx26 gene copy and a working Cx26 copy of the gene on its partner chromosome 13, is a carrier of the Cx26 faulty gene.

- Around 3% of people in the general population are carriers of the Cx26 faulty gene (genetic carriers for Cx26 deafness)
- Genetic carriers for Cx26-associated deafness have normal hearing

As described above, the inheritance of the Cx26 gene is most often autosomal recessive (see Genetics Fact Sheet 8) and is influenced by two factors:

1. The faulty gene copy is located on chromosome 13, an autosome (one of the numbered chromosomes)
2. The effect of the change in the gene is ‘recessive’ or hidden by the presence of the working copy of the gene (see Genetics Fact Sheets 1, 4 & 5)

The inheritance of the faulty Cx26 gene in these families is represented in Figures 60.3 and 60.4 where the faulty gene copy is represented by ‘r’ and the working copy by ‘R’. There are four possibilities, in every pregnancy, for the combinations of gene copies passed from the parents.

As shown in Figure 60.3, if a couple who are both carriers of the faulty Cx26 gene have a baby, in every pregnancy there is

- 1 chance in 4 (25% chance), that they will have a child with a high chance of being affected
- 3 chances in 4 that their child will not have genetic hearing loss although there is a 50% chance that the child will be an unaffected genetic carrier for Cx26-associated deafness just like the parents

As shown in Figure 60.4, if only one parent is a carrier of the faulty Cx26 gene, in every pregnancy there is

- No chance that the couple will have a baby affected with deafness caused by a faulty Cx26 gene copy
- A 1 chance in 2 (ie. 2 chances in 4) or 50% chance that the baby will be an unaffected genetic carrier for Cx26-associated, just like his/her parents

Although faulty copies of the Cx26 gene have been associated with an autosomal dominant pattern of inheritance in some families, this is much rarer.

What if there is a family history of deafness?

Genetic counselling may help to clarify whether an individual’s deafness has been inherited, or is due to environmental factors (see Genetics Fact Sheet 3).

A clinical geneticist will usually spend time examining the individual with deafness, checking the family history and doing tests to identify whether their deafness is part of a syndrome, or due to environmental causes.

Genetic counselling can help by discussing the implications of deafness for that individual and their family, to identify sources of support, and to estimate the chance of deafness occurring again in future children and other family members.

Just as hearing parents can have deaf children, deaf parents can have hearing children, even when both parents’ deafness is genetic. This is because there are over 100 different genes associated with deafness, many of which are recessive.
Is testing available for deafness?

**An individual**

In some states and territories in Australia such as NSW, all newborn babies are screened for hearing loss soon after birth.

There is still a lot of research to be done into the genetic causes of deafness, and for the vast majority of genes associated with deafness genetic testing is not yet available.

In Australia genetic testing is available for *connexin 26*-associated deafness and for a few other forms of genetic deafness where the gene involved has been identified.

**Family members**

Genetic testing may be available to other family members if a deafness-causing gene has already been identified in the individual with deafness.

**During pregnancy**

Generally, there is no testing for deafness available during a pregnancy.

In some families who have already had a child with deafness in which the faulty gene causing the problem has been identified, prenatal testing (testing in pregnancy) or preimplantation genetic diagnosis (PGD) or testing of embryos before a pregnancy, may indicate whether or not a baby is also likely to have deafness, but will not be able to predict the severity of the deafness.

For more information about prenatal testing and options for PGD, please refer to Genetics Fact Sheets 17, 17C and 18.

Genetic counselling may assist in making an informed decision about these options with the most up-to-date information (see Genetics Fact Sheet 3).

**Other Genetics Fact Sheets referred to in this Fact Sheet:** 1, 3, 4, 5, 8, 9, 10, 11, 17, 17C, 18

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