

This fact sheet describes different types of deafness and hearing loss and includes the symptoms, causes, and any treatment or testing that is available.

In summary

- Hearing loss that is in the range of severe or profound is called **deafness**.
- Hearing loss may occur for a number of reasons including an environmental cause alone, a combination of environmental and genetic causes, or genetic causes alone.
- Over 100 genes are known to cause hearing loss either by itself or in combination with other symptoms (a genetic syndrome) and testing is available in certain cases to clarify the cause of an individual's deafness and inform risk for other family members and future generations.

WHAT ARE THE TYPES OF DEAFNESS AND HEARING LOSS?

The ear is a complex organ that allows us to hear, being made up of three main parts: the **outer**, **middle** and **inner ear** (Figure 63.1). 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**. The eardrum and ossicles carry the vibrations to the fluid that fills the inner ear. The vibrations move through fluid in the snail-shaped organ called the **cochlea** stimulating tiny hairs within. When the hairs vibrate they send a signal to the **auditory nerve** which passes information to the brain where sound is processed.

Types of hearing loss and deafness are categorised according to the part of the ear that is affected, with **conductive hearing loss** referring to structural changes in the ear that affect hearing such as blockages of the ear canal or problems with the ossicles, and **sensorineural hearing loss** referring to problems with the inner ear including the hair cells that respond to vibrations, the auditory nerve, or the processing centre in the brain.

Hearing is measured in decibels (dB), which is a measure of sound from 0db being near total silence, a whisper being 15db and normal speech being around 60db.

Severity of hearing difficulty can be categorised by the decibel level that a person can hear:

- Mild (26-40 dB)
- Moderate (41-55 dB)
- Moderately severe (56-70 dB)
- Severe (71-90 dB)
- Profound (90 dB).

A number of terms are used to describe individuals with different levels of hearing difficulty:

- 'Hearing impairment' and 'hearing loss' are often used interchangeably to refer to hearing shown to be below levels for normal hearing by hearing tests (audiometry) and refer to changes in hearing over time. It is common for hearing to decline with increasing age, with more than 1 in 10 Australian's reporting long-term hearing problems.
- 'deafness' (starts with a small 'd') is a commonly used term that implies hearing levels in the severe-to-profound range determined by audiometry. This may be due to hearing loss over time that leads to deafness, or may be present from birth or early in life.
- 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 **AU**Stralian Sign **LAN**guage, and belong to a distinct cultural group.

Hearing loss or deafness may be present before an individual learns to speak, termed **pre-lingual hearing loss**, or may occur later in life after development of normal speech, termed **post-lingual hearing loss**. Typically both ears are affected (**bilateral hearing loss**), but sometimes only one ear is affected (**unilateral hearing loss**).

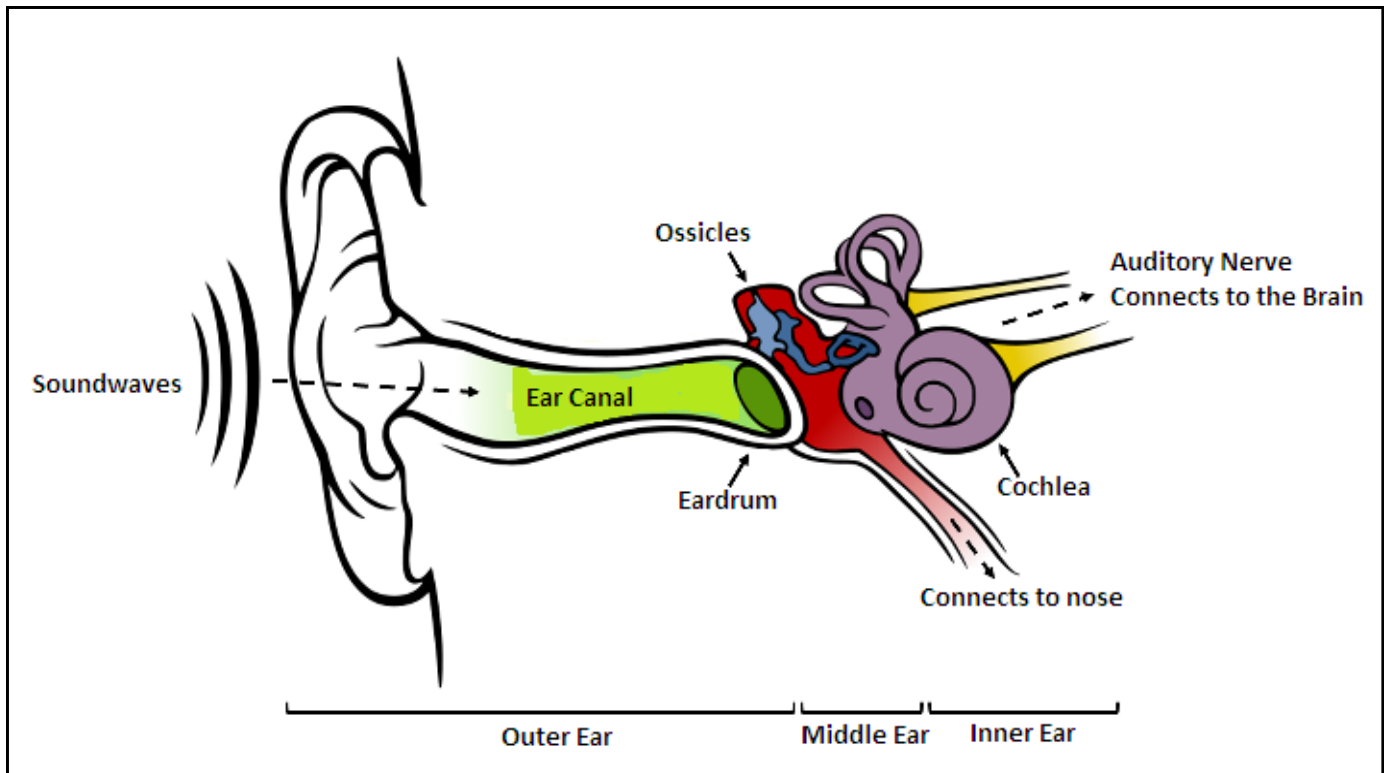


Figure 63.1: The ear and its internal structures

WHAT CAUSES HEARING LOSS AND DEAFNESS?

Our body is made up of millions of cells, and in each cell there are instructions, called genes, that make all the necessary structural components and chemicals for the body to function. These genes are packaged onto little long strands known as chromosomes.

We all have 46 chromosomes arranged into 23 pairs. One copy of each pair is inherited from our mother and the other from our father. The first 22 chromosome pairs are numbered and are known as autosomal chromosomes. The 23rd pair is made up of the sex chromosomes called X and Y. Males have an X and a Y chromosome and females have two copies of the X chromosome.

Since all our chromosomes come in pairs, all our genes also come in pairs. Sometimes, a gene may have a variation in the instruction that causes the gene to no longer function properly. This variation is called a **mutation** or **pathogenic variant**, and means that the product produced by the gene, called a protein, is impaired or even absent.

Gene mutations may be inherited from a parent, or occur for the first time in an individual. Once you have a gene mutation however, it may be passed on to future generations. This is referred to as genetic inheritance.

Hearing loss and deafness may be caused by factors in the environment, a combination of genetic and environmental factors, or genetic factors alone.

In about 50% of cases, the cause is exposure to an environmental factor including:

- Exposure to loud noises such as music or industrial machinery (industrial deafness)
- 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 increasingly being recognised that some individuals may be more susceptible to hearing damage from environmental factors. This suggests that both genetic and environmental factors are important for these individuals.

Deafness that is caused by genetic factors alone accounts for the other 50% of cases that do not have any environmental explanation. **Genetic deafness** occurs without any other symptoms (deafness only) in about 70% of cases and this is known as **non-syndromic genetic deafness**, while 30% occur as part of a group of symptoms that are known to cause a particular syndrome and this is known as **syndromic genetic deafness**. (See figure 63.2)

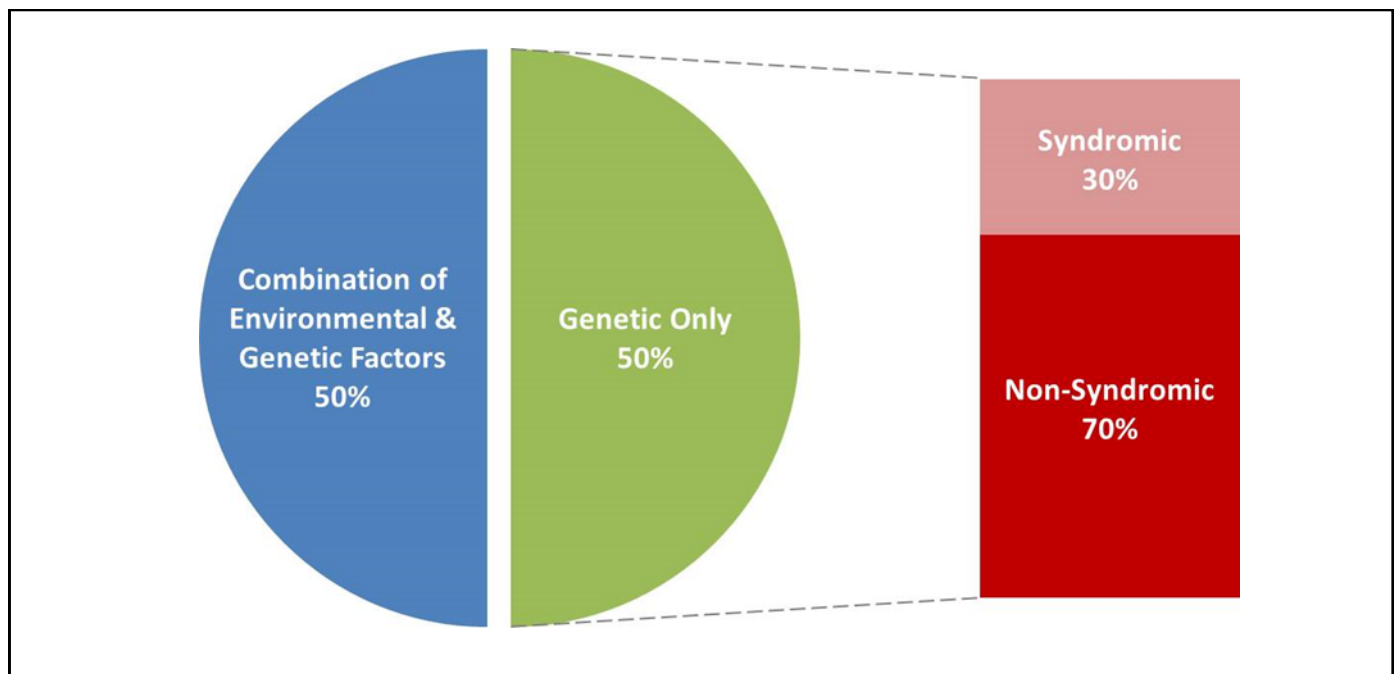


Figure 63.2: Causes of hearing loss and deafness

Both syndromic and non-syndromic genetic deafness may be due to a single faulty gene. **Syndromic deafness** involves genes important for many different functions in the body in addition to hearing. There are around 450 different syndromes that involve hearing loss as one of many symptoms. Non-syndromic deafness on the other hand involves genes whose main role is in hearing alone. There are more than 100 known genes for non-syndromic deafness.

The most well-known gene involved in non-syndromic genetic deafness is **GJB2**, which contains the instructions for the protein called **connexin-26**.

This protein is important in the inner ear, and while we still don't know exactly what its role is, the outcome of a mutation that produces abnormal connexin-26 or stops its production altogether disrupts the flow of information about sound through the inner ear to the brain and results in **sensorineural** (due to problems with the inner ear) **pre-lingual** (onset before speaking) **non-syndromic deafness**. Research is on-going to better understand genes like **GJB2** and to identify the many different genes involved in hearing loss and deafness that we don't yet know about.

HOW IS DEAFNESS INHERITED?

Genetic deafness may be inherited in different ways depending on whether it occurs by itself (non-syndromic), as part of a group of symptoms (syndromic), and which of the many known genes is involved. For **GJB2**, the mode of inheritance is known as **autosomal recessive**, where both parents are unaffected carriers of a mutation and have one copy of their gene that is faulty (has the mutation) and one that is working. Their offspring will be affected only if they inherit a faulty copy from both parents, meaning that they have no working copy of the gene (Figure 63. 3).

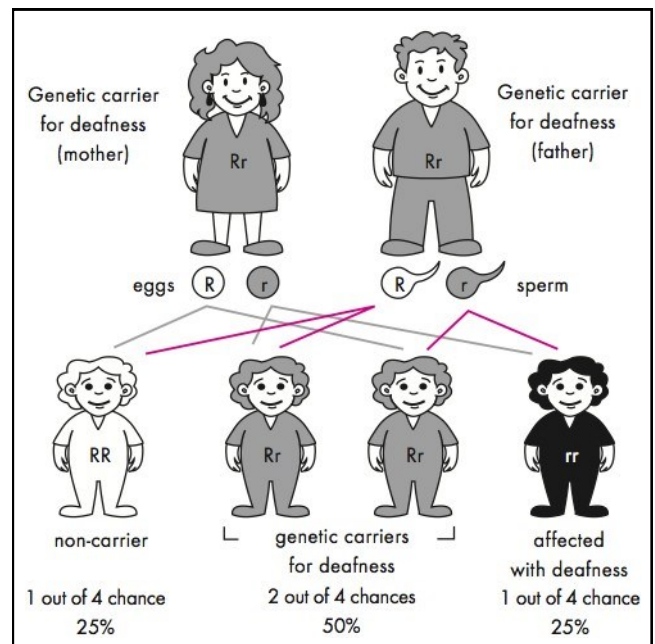


Figure 63.3: Autosomal recessive inheritance where both parents are carriers of the faulty gene. The faulty gene copy is represented by 'r'; the working copy by 'R'.

For every pregnancy where a couple are both genetic carriers of a **GJB2** mutation there is:

- 1 chance in 4 (25% chance) that they will have a child who inherits both copies of the recessive gene mutation from his/her parents. In this case, no working gene product will be produced and their child will be affected.
- 1 chance in 4 (25% chance) that their child will inherit both copies of the working gene and will be unaffected and not a genetic carrier.
- 1 chance in 2 (2 chances in 4 or 50% chance) that their child will inherit the recessive gene mutation and the working copy of the gene from the parents and he/she will be an unaffected genetic carrier, just like the parents.

Many other patterns of inheritance occur for hearing loss and deafness therefore it is important to discuss your family history in detail with a health professional to inform an accurate risk prediction for family members and future generations.

IS THERE ANY TESTING OR TREATMENT AVAILABLE FOR DEAFNESS?

Clinical Testing

Deafness is usually diagnosed using audiometry, which measures how well an individual hears. Other tests are used to determine the cause of deafness, including structural changes to the ear or environmental factors. Genetic counselling may help to clarify whether an individual's deafness has been inherited.

Genetic Testing

Genetic testing is available to try and identify the faulty gene running in a family. As mentioned, over 100 genes are known to cause deafness so deciding which to test will depend on the specific type of hearing loss or deafness in the affected family member as well as family history. This can be discussed with your treating health professional who can determine if this type of testing will be informative for your family.

The first step is a **mutation search** in an affected family member to try and identify the faulty gene. If this is identified genetic testing can be offered to other family members who are at-risk which is called **predictive testing**. If a mutation cannot be found though, no further genetic testing can be offered in the family.

Prenatal testing and PGD

For couples where the familial mutation is identified testing may be available during a pregnancy to determine whether or not the baby has inherited the mutation. It may also be possible to undergo pre-implantation genetic diagnosis (PGD) on an embryo created using in vitro fertilisation (IVF) to ensure that a child does not inherit the mutation. These options are best discussed and considered before pregnancy, when possible, in order to ensure all possible risks, benefits and outcomes can be explored.

Treatment Options

Conductive deafness can sometimes be corrected by surgery or other interventions, while in general sensorineural hearing loss is permanent and may be helped by hearing aids or cochlear implants. Individuals with severe or profound deafness are able to access interventions to help them communicate including learning to lip-read or use sign language.