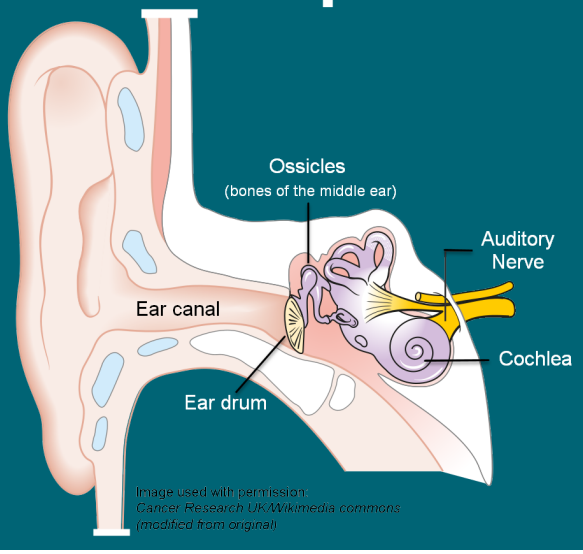
What to Expect at a Genetics Visit for Hearing Loss



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In this information sheet:

Why was I referred for a genetics visit?

Anyone with hearing loss could get referred for a genetics visit if their doctor suspects a genetic cause. A genetics referral is recommended for newborns and infants with hearing loss. Hearing loss that happens early in life often has a genetic cause. A genetics visit may also be useful for a person diagnosed at a later age, especially when there is no clear reason for hearing loss. However, gradual hearing loss as you get older is very common and does not need a genetics visit.

What can a genetics visit tell me about hearing loss?

A genetics visit can help:

- Find out why a person has hearing loss.
- Predict how hearing may change over time.
- Estimate the chance for other health conditions (such as vision, heart, or kidney problems).
- Estimate the chance for hearing loss in other family members (including future children).

What happens at a genetic visit?

A genetic counselor talks with you by phone or video visit. You are asked about the family medical history and the birth history of the person with hearing loss. Time is spent reviewing possible causes of hearing loss and answering your questions. Genetic testing may be offered. If a physical exam would help with a diagnosis, you will be scheduled to meet a medical geneticist (a doctor who specializes in genetic conditions).

About Hearing Loss

Hearing loss is when your ability to hear sound is less than usual. It can affect one ear or both ears. The amount of hearing loss is labeled as mild, moderate, severe, or profound.

Types of hearing loss

The types of hearing loss describe which part of the ear is involved.

Sensorineural hearing loss is when the inner ear (cochlea) or the nerve from the ear to the brain (auditory nerve) does not recognize sound in the usual way.

Conductive hearing loss is when sound does not pass easily from the outer ear to the ear drum and tiny bones of the middle ear (ossicles). This may be due to changes in the way the ear is formed or caused by a temporary problem, like an ear infection or fluid in the ear.

Mixed hearing loss happens when there is a combination of both conductive hearing loss and sensorineural hearing loss.

Auditory neuropathy is when the ear detects sound normally, but has trouble sending it to the brain.

What to Expect at a Genetic Visit for Hearing Loss

Genetic causes of hearing loss

Genetic hearing loss is caused by a change in a gene that affects hearing. There are over 400 genes that can lead to hearing loss.

Non-syndromic hearing loss: Some genes only affect hearing. This is called non-syndromic hearing loss. Seven out of 10 people with genetic hearing loss have a non-syndromic type. The most common type of non-syndromic hearing loss is due to changes in a gene called GJB2 (or connexin 26).

Syndromic hearing loss: There are over 300 genes that can cause hearing loss and also affect other parts of the body. This is called syndromic hearing loss because hearing loss is part of a genetic syndrome. Most of these conditions are rare.

Could there be a genetic cause with no hearing loss in the family?

There could be a genetic cause even if no one else in the family has hearing loss. More than half of all early hearing loss is due to a genetic cause and usually there is no family history of deafness or hearing loss. The most common genetic cause of hearing loss is due to recessive inheritance. This happens when two hearing parents silently carry a change in a gene that causes hearing loss.

What are genetic tests?

Genetic tests are medical tests that can find out whether or not certain genes are working correctly. Most genetic testing is done by taking a blood sample from the arm or collecting a saliva sample.

What genetic testing is done for hearing loss?

Genetic testing is usually done on a panel of over 200 genes that cause hearing loss. This means one blood sample is tested for many genetic causes of hearing loss at the same time.

Can the cause always be found by genetic testing?

There are many different causes of hearing loss, and a specific cause is not always found. Genetic testing does not test for all types of genetic hearing loss. There are also non-genetic causes of hearing loss, such as premature birth, infections, and exposure to certain medicines.

What other tests are recommended for a person with hearing loss?

Other tests may be recommended for a person with hearing loss based on their medical history or a genetic testing result.

Testing could include:

- Eye exam by an ophthalmologist (eye doctor specialist)
- Electrocardiogram (EKG) - a test that monitors the heartbeat
- Urine studies (urinalysis with microscopy)
- Thyroid studies

Some people with hearing loss may have imaging done to look at the inner ear and the auditory nerve. An otolaryngologist (ear doctor) can discuss the risks and benefits of imaging studies.