

A guide for people
living with Fabry disease

Fabry disease and hearing loss



Fabry is a rare disease caused by genetic mutations. People with Fabry have trouble breaking down and getting rid of certain sugary-fatty waste substances in the cells of their body. The disease can affect many parts of the body, including the kidneys, eyes, heart, skin and blood vessels.

Hearing loss is more common in people with Fabry than it is in the general population, and tends to start earlier and progress faster than age-related hearing loss. This is thought to be because of a build up of waste substances in the nerve cells of the ear, or in the blood vessels of the inner ear. This blocks and narrows the blood vessels, reducing blood flow and damaging the auditory nerve (the nerve connecting the inner ear to the brain).

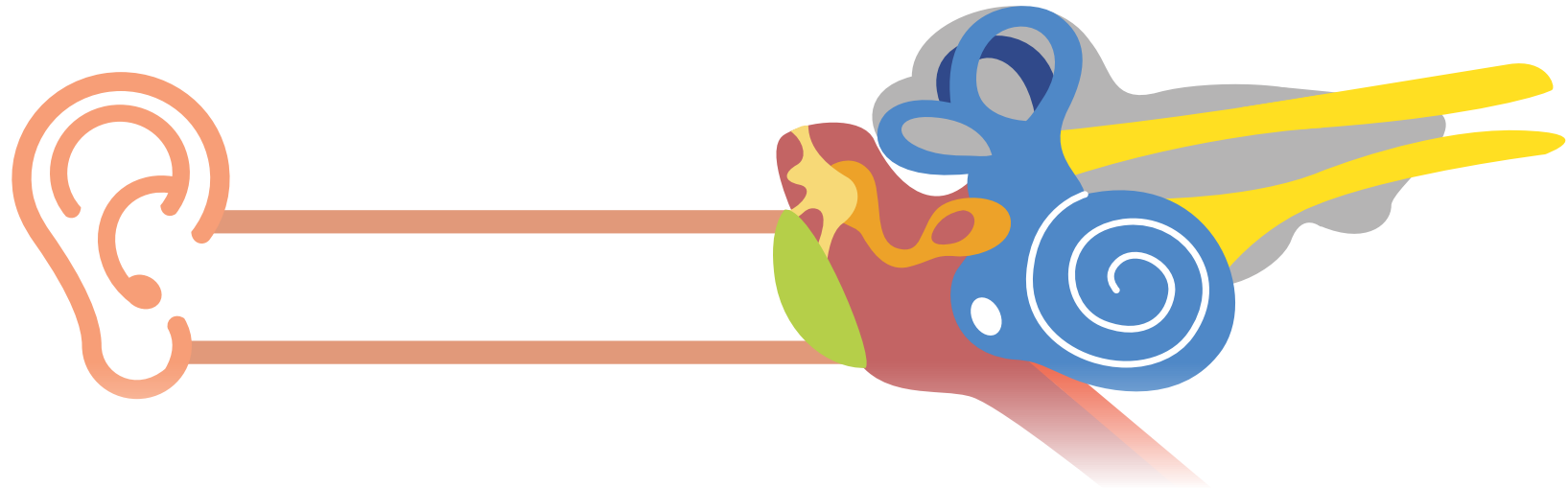
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This guide has been developed and funded by Amicus Therapeutics UK Ltd.



Your ear

The human ear is designed to capture sound and send its signals to the brain. The ability to hear sound helps us understand our environment, communicate with others, learn and develop.



The ear is made up of three main parts.

The outer ear

This is the visible part of the ear. It catches sound, which travels to the middle ear.

The middle ear

This part contains the eardrum and three tiny bones, called ossicles, that vibrate to sound.

The inner ear

This part contains the snail-shaped cochlea, which converts the sound vibrations from the ossicles into signals that are sent to the brain. The inner ear also has semicircular tubes to help with balance.

Each part of the ear is essential to good hearing, and a problem in or damage to any part can affect your ability to hear.

Hearing loss is quite common. According to the World Health Organization, over 5% of the world's population has disabling hearing loss.

The most common type of hearing loss associated with Fabry is caused by damage to the cochlea or nerve fibres within the inner ear. This type of hearing loss is called sensorineural hearing loss. With this type of hearing loss, sounds are softer and more difficult to distinguish. While this type of hearing loss is permanent, there are many ways it can be treated, including hearing aids and cochlear implants.

How do I know if I have hearing loss?

The checklist below may help you decide whether you have hearing loss.

DO YOU...

- Find yourself constantly turning up the volume on your phone, television or other devices, or being told your devices are too loud?
- Hear a constant roaring, ringing or hissing in your ears (which is called tinnitus)?
- Have difficulty understanding or keeping up with conversations, and often misunderstand what people say, especially in noisy places?
- Have difficulty hearing on the phone?
- Frequently ask people to repeat themselves?
- Often think people are mumbling?
- Have trouble hearing others when there is background noise or more than two people?
- Feel you are constantly straining to hear?
- Avoid social situations and crowded places because of hearing difficulties?

If you have answered yes to any of these questions, you may have hearing loss and should consider discussing a full hearing evaluation with your healthcare professional.

How is hearing tested?



Testing is easy, painless and non-invasive. It is performed by a hearing healthcare professional and usually involves:

- gathering information about your health and medical history, and
- testing to check what volumes and pitches you can hear for speech and noise in general.

This kind of check is called audiometry testing. It involves putting on headphones or earbuds and listening for words or noises at different pitches and volumes. When you can hear something you respond (for example, by repeating the word or pressing a button).



You might also be checked to see how well your eardrum is moving and have the functioning of the middle ear assessed.

This kind of check is called tympanometry testing. It involves putting a soft plug in your ear that will create pressure changes and generate sound.



The results of your tests can be shown on a graph and discussed with your healthcare professional.

The results of an audiometry test are shown in a graph called an audiogram.

The results of a tympanometry test are shown in a graph called a tympanogram.

Special hearing tests are designed specifically for children with hearing loss.

Regular hearing evaluations are recommended.

Why should I get treatment for hearing loss?



Hearing loss can have a wide-ranging effect on your day-to-day life and well-being, and can affect those around you.

Getting treatment as soon as possible can help slow down hearing loss.

Hearing loss can cause or contribute to the following.

- Communication problems and strains on your relationships.
- Exhaustion or fatigue from having to concentrate to be able to hear and understand those around you.
- Withdrawal from social activities, contributing to loneliness and isolation, and reduced stimulation.
- Reduced performance at work or school.
- Emotional and psychological issues, including anger, frustration, low self-esteem, stress, anxiety and depression.
- Physical issues, including headaches, tense muscles and increased blood pressure.
- Safety issues from reduced alertness or awareness.
- Financial issues. Hearing loss typically affects household income, and people with hearing loss are more likely to be unemployed and paid less than those with normal hearing.
- Hearing loss has also been linked with an increased risk of dementia, including problems with learning and memory.



Hearing loss in children

The earlier hearing loss happens, the greater the effect on a child's development. Likewise, the earlier hearing loss is treated, the better the chance of not having serious effects.

The ability to hear is vital to learning how to listen, understand and talk. It's fundamental to developing language and verbal communication skills. If left untreated, childhood hearing loss can have a lasting effect, even beyond speech development. It can:

- slow the development of social skills
- lead to loneliness and low self-esteem, and
- interfere with the development of the learning and attention skills that are needed in school and life.



What treatments are available?

There are many ways to manage hearing loss. Your treatment will depend on the type and severity of your hearing loss.

Treatment for tinnitus

Various treatments are available for tinnitus, from medication to devices and various forms of therapy. Which one is right for you will be discussed during a medical examination. If your tinnitus is associated with hearing loss, hearing aids may be helpful.

Hearing aids

Hearing aids work by amplifying sounds. The size, price and special features of hearing aids, and the way they sit in the ear, can vary a lot. Finding the right one for you depends on your lifestyle, concerns about appearance and the degree of hearing loss.

There are lots of choices, but hearing aids can be roughly grouped into two categories.

- **In the ear**

These are usually custom-made to fit snugly in your ear.

- **Behind the ear**

These hook over the top of the outer ear with tubing that routes the sound into the ear.

Cochlear implants

For those with severe hearing loss, surgically implanting a device that directly stimulates the auditory nerve may be a viable option. Rather than amplifying sound, the cochlear implant provides the sense of sound. It consists of two parts. The **external** piece contains a microphone, processor and transmitter. The **internal** part contains a receiver that converts signals from the transmitter to electrical pulses that are sent through the nerve to the brain, where they are perceived as sound.

Assistive listening device (ALD)

ALDs provide solutions for specific issues, like hearing the television or waking up to an alarm. Some are designed to work alone and some with hearing aids. Categories include the following.

- **Using the phone**

A wide variety of options are available to help with using the phone, from amplified phones to hearing-aid compatible phones and captioned telephone services (which allow you to listen to what the other party is saying while also read captions of what the other party is saying).

- **Watching television**

ALDs for the television allow you to set the volume of the television for you independently of the volume set for everyone else in the room. This may be achieved by a wireless connection from your hearing aid to the television, or a wireless headset with personal volume control.

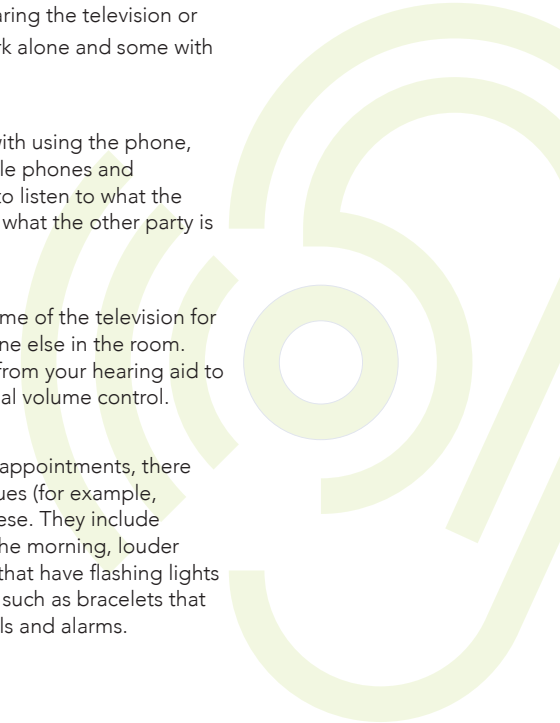
- **Alerts**

So you don't miss important warnings, alerts or appointments, there are systems that feature louder sounds, visual cues (for example, flashing lights), vibration or a combination of these. They include vibrating alarm clocks that shake you awake in the morning, louder smoke alarms and carbon-monoxide detectors that have flashing lights to get your attention, and wearable technology such as bracelets that vibrate and light up to alert you to incoming calls and alarms.

Speech therapy

Speech and language therapy can help children with hearing loss catch up on delays in speech development.

A member of your healthcare team can advise you on getting support for your hearing loss.



Tips

Here are some suggestions to help improve communication with hearing loss.

- Talk to people about your hearing loss. If they understand your situation, they can work with you to help with communication.
- Don't be embarrassed to ask for what you need in a given situation, whether it's having someone repeat what they have said or asking for a seat where you can see everyone's faces.
- Try to sit face-to-face when you are speaking with someone, so you can hear them more clearly and pick up on facial expressions.
- When going out to a restaurant with a group, try to find a time and place that isn't too crowded, noisy or dimly lit. Ask for a table in a quiet spot.
- Listening closely requires serious concentration and can use lots of energy. If you are tired or under the weather don't push yourself. Take a break in a peaceful place.
- Connect with other people with hearing loss. They can be a great source of support and information.

Support organisations

For hearing loss

- Action on Hearing Loss
- Hearing Link
- The Ear Foundation

For Fabry disease and rare diseases

- The MPS Society
- Rare Disease UK

Please contact your healthcare team for information, guidance and support.

Information in this guide came from the following sources.

A Ciceran, S De Maio, J Inborn Errors Metab Screening, 2016

DA Laney, and others, J Genet Couns, 2013

A Ortiz, and others, Mol Genet Metab. 2018

S Hegemann, and others, Eur J Clin Invest, 2006

Fabry disease audiology symptoms, National Fabry Disease Foundation. (www.fabrydisease.org/index.php/component/content/article?id=127)

A Keilmann, and others, Fabry Disease: Perspectives from 5 Years of FOS, 2006

How hearing works, Cochlear.com (www.cochlear.com/uk/en/home/diagnosis-and-treatment/diagnosing-hearing-loss/how-hearing-works)

Effects of hearing loss on development, American Speech-Language Hearing Association (www.asha.org/public/hearing/effects-of-hearing-loss-on-development)

RA Dobie & SB Van Hemel, National Research Council, Hearing loss: Determining eligibility for Social Security benefits, National Academy Press, 2005

Deafness and hearing loss (April 2021), World Health Organization (www.who.int/news-room/fact-sheets/detail/deafness-and-hearing-loss)

Hearing loss guide, Healthy Hearing (www.healthyhearing.com/help/hearing-loss)

Types of hearing loss, Healthy Hearing (www.healthyhearing.com/help/hearing-loss/types)

Types and causes of hearing loss, Cochlear.com (www.cochlear.com/uk/en/home/diagnosis-and-treatment/diagnosing-hearing-loss/types-and-causes-of-hearing-loss)

Signs of Hearing loss, Hearing Industries Association. (www.betterhearing.org/your-hearing-health/signs-of-hearing-loss)

Symptoms Hearing loss, NHS (www.nhs.uk/conditions/hearing-loss)

Hearing loss tests, Healthy hearing (www.healthyhearing.com/help/hearing-loss/tests)

What is tympanometry and how is it used? Healthy hearing, (www.healthyhearing.com/report/33583-What-is-tympanometry-and-how-is-it-used)

S Arlinger, Int J Audiol, 2003, O Nordvik, and others, BMC Ear Nose Throat Disord, 2018

The hidden healthcare cost behind untreated hearing loss (www.nehoustonhearing.com/this-just-in-the-hidden-healthcare-cost-behind-untreated-hearing-loss)

The impact of hearing loss on relationships, Healthy Hearing (www.healthyhearing.com/report/52619-The-impact-of-hearing-loss-on-relationships)

Consequences of hearing loss, Hear-it (www.hear-it.org/consequences-of-hearing-loss-2)

Impact of hearing loss, Cochlear.com (www.cochlear.com/uk/en/home/diagnosis-and-treatment/when-to-consider-implants-for-adults/hearing-loss-is-affecting-daily-life)

How hearing loss can impact mental health, Healthy Hearing (www.healthyhearing.com/report/52437-The-complex-link-between-depression-and-hearing-loss)

The impact of untreated hearing loss on household income. Better hearing institute (www.betterhearing.org/HIA/assets/File/public/marketrak/MarkTrak_VII_The_Impact_of_Untreated_Hearing_Loss_on_Household_Income.pdf)

FR Lin & L Ferrucci, Arch Intern Med, 2012

S Kochkin & CM Rogin, Hear Rev, 2000

D Jung & N Bhattacharyya, Ann Otol Rhinol Laryngol. 2012

A Wingfield & JE Peelle, Aging health, 2012

FR Lin, and others, JAMA Intern Med, 2013

NJ Laughan, and others, Deafness & Education International. 2017

Tinnitus, Mayo Clinic (www.mayoclinic.org/diseases-conditions/tinnitus/diagnosis/treatment/drc-20350162)

Tinnitus, NHS inform (www.nhsinform.scot/illnesses-and-conditions/ears-nose-and-throat/tinnitus)

Hearing aids: How to choose the right one, Mayo Clinic (www.mayoclinic.org/diseases-conditions/hearing-loss/in-depth/hearing-aids/art-20044116)

Hearing aid types and styles, Healthy Hearing (www.healthyhearing.com/help/hearing-aids/types)

Cochlear implants, Healthy Hearing (www.healthyhearing.com/help/hearing-aids/cochlear-implants)

Assistive listening devices guide (3rd edition), Healthy Hearing (www.healthyhearing.com/help/assistive-listening-devices)

Speech and language therapy, National Deaf Children's Society (www.ndcs.org.uk/information-and-support/language-and-communication/spoken-language/speech-and-language-therapy)