

A guide for people
living with Fabry disease

Fabry disease and **HEARING LOSS**

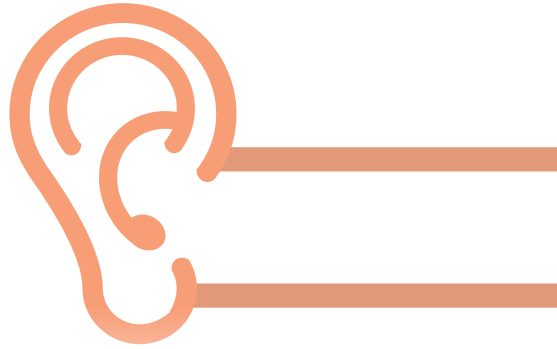


Fabry is a rare disease caused by genetic mutations.¹ People with Fabry have trouble breaking down and getting rid of certain sugary-fatty waste substances in the cells of their body.² The disease can affect many parts of the body, including the kidneys, eyes, heart, skin, and vascular system.³

The rate of hearing loss in the Fabry community is much higher than that in the general population and tends to start earlier and progress faster.⁴ Hearing loss in Fabry may be directly caused by an accumulation of these waste substances in the nerve cells of the ear; and indirectly by the accumulation in the blood vessels of the inner ear.^{5,6} This blocks and narrows the blood vessels, reducing blood flow and damaging the auditory nerve.⁵

YOUR EAR

The human ear is designed to capture sound and convey its signals to the brain.^{7,8} The ability to hear sound helps us understand our environment, communicate with others, learn, and develop.^{9,10}



THE EAR IS MADE UP OF 3 DIFFERENT PARTS:

THE OUTER EAR

includes the visible part called the pinna, which catches sound and sends it to the middle ear inside.^{7,8}

Each part of the ear is essential to good hearing and any impairment or damage along the way can affect your ability to hear.⁷

Hearing loss is quite common: According to the World Health Organization, over 5% of the world's population has disabling hearing loss.¹¹ The rate of hearing loss in the Fabry community is much higher than that in the general population and tends to start earlier and progress faster.⁴

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.^{4,12,13} This reduces both the volume and clarity of perceived sound, meaning sounds are softer and more difficult to understand.^{13,14} While this type of hearing loss is permanent, there are many ways it can be treated—including hearing aids and cochlear implants.^{5,13}



THE MIDDLE EAR

contains the eardrum and three tiny bones called ossicles that vibrate to the sound.^{7,8}

THE INNER EAR

houses the snail-shaped cochlea, which converts sound vibrations into signals that are sent to the brain.^{7,8} It also has semicircular canals to help with balance.⁷

HOW DO I KNOW IF I HAVE HEARING LOSS?

This symptom checklist may help you determine if your hearing may be impaired.

DO YOU:^{10,15,16}

- Find yourself constantly turning up the volume on your phone, television, or other devices, or being told your devices are too loud by others?
- Hear a constant roaring, ringing, or hissing in your ears (which is called tinnitus)?
- Have difficulty understanding or keeping up with conversations? Do you 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 understanding 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 provider.

How is hearing tested?



Hearing or audiology testing is easy, painless, and non-invasive. It is performed by a hearing healthcare professional and usually involves:¹⁷

- A hearing health and medical history
- A test 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 (e.g. repeating the word, pressing a button etc.)



- You might also be checked to see how well your eardrum is moving and the functioning of the middle ear:^{15,16}

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:^{15,16}

An audiogram is produced during an audiometry test

A tympanogram is produced during a tympanometry test

Special paediatric hearing tests are designed specifically for children with hearing loss.¹⁷

Once a baseline is established, periodic hearing evaluations are recommended, as is the documentation of your symptoms and their progression.⁵

Why should I seek treatment for hearing loss?



Hearing loss can have a wide-ranging impact on your day-to-day life and well-being, as well as affecting those around you.¹⁹⁻²¹

Getting treatment as soon as possible can help improve or fix the problems caused by hearing loss.^{22, 24-26, 28}

Hearing loss can cause or contribute to:

- Communication problems and strains on your relationships²¹⁻²³
- Exhaustion or fatigue, from having to concentrate to be able to hear and understand those around you^{19, 24}
- Withdrawal from social activities, contributing to loneliness and isolation, and reduced stimulation^{19, 25}
- Reduced work or school performance²⁶
- Emotional and psychological issues: including anger, frustration, low-self-esteem, stress, anxiety and depression^{23, 25}
- Physical issues: including headaches, tense muscles and increased blood pressure²³
- Safety issues from reduced alertness, awareness or functioning^{24, 27, 28}
- Financial issues: hearing loss typically negatively affects household income, and people with hearing loss are more likely to be unemployed and paid less than those with normal hearing^{26, 29}
- Hearing loss has also been linked with increased risk of dementia and cognitive decline, including problems with learning and memory.^{24, 30, 31}



Hearing loss in children^{9, 31}

The earlier hearing loss occurs, the greater the impact on a child's development. Likewise, the earlier hearing loss is addressed, the less serious the ultimate impact will be.²⁴

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

- Slow the development of social skills
- Lead to feeling alone and low self-esteem
- Interfere with the development of the learning and attention skills needed in school and life



What types of treatments are available?

TREATMENT FOR TINNITUS^{33,34}

There are a variety of treatments available for tinnitus—from medications to masking devices to various forms of therapy. Which one is right for you will be determined 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, most are digital and all are powered with a hearing aid battery.³⁵ The size, price, special features, and way they sit in the ear can vary a lot.³⁵ Finding the right one for you depends on your lifestyle, cosmetic concerns and your degree of hearing loss.³⁶

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

In-the-ear (ITE)³⁶

ITE aids are usually custom-made to fit snugly in your ear canal. Types include:

- Completely in the canal (CIC)— this style is one of the smallest, most discreet ITEs sitting deep in the ear canal
- In-the-canal (ITC) – this type is slightly larger than CIC and sits in the lower portion of the outer ear bowl. While more visible, their size allows the inclusion of more features, like volume control
- Low profile – this group is similar to the ITC but larger. Its fit ranges from filling half to almost the entire outer ear bowl. The larger sizes can be useful for people with dexterity issues, and can accommodate more features



Behind-the-ear (BTE)³⁶

BTE aids hook over the top of the outer ear with tubing that routes the sound into the ear canal. Examples of BTE aids include:

- Receiver in the ear (RITE) or Receiver in the canal (RIC) – with this type of hearing aid the speaker sits in the ear canal while the microphone and processor sit on top of the ear



COCHLEAR IMPLANTS³⁷

For those with severe or profound hearing loss, the surgical implantation of 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 components. 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.

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

ASSISTIVE LISTENING DEVICE (ALD)³⁸

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

- **Telephone:** A wide variety of options are available to enhance communication on the telephone—for both mobile and landline—from amplified phones to hearing aid-compatible phones and captioned telephone services
- **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. These include wirelessly connecting your hearing aid to the television, either directly or through a supporting accessory, and wireless headsets with personal volume control
- **Alerts:** So you don't miss important warnings, alerts, or appointments, there are a number of available systems that feature enhanced sound, visual cues, vibration, or some combination of these. They include vibrating alarm clocks that shake you awake in the morning; smoke alarms and carbon monoxide detectors that use increased volume and flashing lights to get your attention; and wearable technology, such as bracelets and fobs, that use vibration and light to alert you to incoming calls and alarms

SPEECH THERAPY^{9,39}

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 enhance 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 requesting 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 visual cues
- 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. Request 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.

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