Fabry disease is a rare condition caused by certain gene variants (also known as mutations). People with Fabry disease have trouble breaking down and getting rid of certain fatty waste substances within their cells. The disease can affect cells in many parts of the body, including the kidneys, eyes, heart, skin and vascular system.1-3 The rate of hearing loss in the Fabry disease community is much higher than that in the general population and tends to start earlier and progress slowly.4 Hearing loss in Fabry disease is believed to be caused by an accumulation of these waste substances within the blood vessels of the inner ear. This narrows the blood vessels, reducing blood flow and damaging the auditory nerve.2,3,5
THE EAR

The human ear is designed to capture sound and convey 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 3 DIFFERENT PARTS:6

THE OUTER EAR includes the visible part called the pinna, which catches sound, and the ear canal.

THE MIDDLE EAR contains the eardrum and 3 tiny bones called ossicles (hammer, anvil and stirrup) that pass sound waves to the inner ear.

THE INNER EAR houses the semicircular canals, which help with balance, and the spiral-shaped cochlea, which converts sound waves into nerve impulses that are sent to the brain.

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

Hearing loss is quite common. According to the World Health Organization, more than 5% of the world’s population has disabling hearing loss.7 The rate of hearing loss in the Fabry disease community is much higher than that in the general population.8

The most common type of hearing loss associated with Fabry disease is called sensorineural hearing loss, which is caused by damage to the nerve fibers within the inner ear. This reduces the volume and the clarity of perceived sound. While this type of hearing loss is progressive and irreversible, there are many ways it can be treated—from hearing aids to cochlear implants.9-11
HOW CAN A PERSON TELL IF THEY HAVE HEARING LOSS?

People who have hearing loss may:

- Habitually turn up the volume on the phone, television or other media devices
- Hear a constant ringing in their ears (tinnitus)
- Have difficulty understanding or keeping up with conversations
- Need to be face-to-face with someone in order to carry on a conversation
- Frequently ask people to repeat themselves
- Often think people are mumbling
- Have trouble understanding others when there is background noise
- Feel that they are constantly straining to hear
- Avoid encountering new places, new people, groups or social situations

The symptoms and effects of hearing loss can be subtle and are not always easy to recognize. Anyone who experiences one or more of the symptoms listed above may have hearing loss and should consider discussing a full hearing evaluation with your health-care professional.
Hearing or audiology testing is simple, painless and noninvasive. It is typically performed by a health-care professional who specializes in hearing and usually involves:

• A thorough hearing health history

• Audiometry testing for tone and speech. These tests involve:
  — Listening for tones at different volumes and pitches through headphones
  — Repeating words heard at different loudness levels

• Tympanometry testing to gauge the movement of the eardrum. During this test a soft plug that changes sounds and pressure is placed in the ear

• Reviewing a graph depicting the test results (called an audiogram) with the health-care professional

Special pediatric audiology testing is designed specifically for children who may have hearing loss is also available.

Once a baseline is established, periodic audiology evaluations are recommended, as is the documentation of symptoms and their progression.
Hearing loss can negatively impact quality of life in numerous ways. Ordinary activities, such as social gatherings or attending live music performances, may present new difficulties. Even simply talking with friends, colleagues and family members may become challenging. Because of this, the treatment of hearing loss can enrich a person’s experiences across many aspects of life and enhance their ability to communicate with friends and loved ones.

If left untreated, hearing loss can have serious effects on overall health and well-being. In adults, untreated hearing loss may:

- Put a strain on interpersonal relationships
- Lead to withdrawal from social activities, contributing to loneliness and isolation
- Be linked to emotional issues, such as irritability, frustration, negativity and anger, and lead to psychological problems including depression, stress and anxiety
- Result in reduced alertness and awareness, increasing the risk to personal safety
- Contribute to mental decline including impaired memory, thinking and judgment
- Have a negative economic effect. People with untreated hearing loss are more likely to be unemployed—and those who find work tend to earn less, and have more difficulty functioning on the job than those with normal hearing. Their medical costs tend to be higher as well, which can also take its toll on finances

Hearing loss in children can have even more devastating effects and should be addressed early to ensure normal development.

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

- Impede the development of social skills
- Lead to social isolation and low self-esteem
- Interfere with the development of learning and attention skills needed to succeed in school and life
What types of treatments are available?

TREATMENT FOR TINNITUS

A variety of treatments are available for tinnitus, including medications, devices that use sound to help mask the tinnitus and various forms of therapy. Medical examination can help determine which treatment approach may be best for a person’s specific needs. If tinnitus is associated with hearing loss, hearing aids may be helpful.

HEARING AIDS

Hearing aid technology has advanced dramatically in recent years. These noninvasive devices are designed to amplify sound—and they come in so many different types and configurations, there should be one to fit every ear and budget! Most hearing aids are wireless and battery operated. Some of the main types of hearing aids that are available are described below.

- **Completely in the canal (CIC)** devices, which sit fully inside the ear canal, are the smallest and least visible type of hearing aid. Although their very small size offers some advantages, it also limits the features they can offer and can make them awkward to handle.
- **In-the-canal (ITC)** hearing aids are slightly larger than CIC devices and fit partly within and partly outside the ear canal. Their larger size allows them to offer some additional features than CIC devices.
- **In-the-ear (ITE)** devices fit in the bowl-shaped area of the outer ear. Many offer features that can’t be included in most smaller hearing aids, such as a volume control or special microphones that can help when there is background noise.
- **Behind-the-ear (BTE)** hearing aids have two components: a piece that sits behind the ear that holds the electronics, and an earpiece that goes inside the ear. They come in a range of different styles including receiver in the ear (RITE) and receiver in the canal (RIC) and can provide an even wider variety of features.

COCHLEAR IMPLANTS

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 actual sound, the cochlear implant provides a sense of sound by bypassing the damaged area of the inner ear. Cochlear implants require surgical implantation.
Hearing loss can be managed in many different ways, depending on its type and severity.

ASSISTIVE LISTENING DEVICE (ALD)28
ALDs provide amplification of sound or improved communication for specific hearing challenges. Some are designed to work alone and some work with hearing aids. ALD use includes:

- **Telephone**: A wide variety of options are available to enhance telephone communication for both mobile devices and landlines—from amplified phones to captioned telephone service from Bluetooth ear pieces to hearing aid–compatible phones

- **Television**: There are many devices that can help enhance sound perception without turning up the television’s actual volume—from wireless headsets with personal volume control to Bluetooth-enabled accessories for hearing aids

- **Alerts**: A number of alert systems are available that feature enhanced sound, visual cues, vibration or some combination of all of these to provide warnings or reminders. They include vibrating alarm clocks; doorbells, smoke alarms and carbon monoxide detectors that use increased volume and flashing lights to attract attention; and wearable technology, such as bracelets and smart watches and pagers that use vibration and light to alert the wearer to notifications, alarms and incoming calls

- **Wireless microphones**: These small, light, portable devices are designed to boost hearing in noisy environments, such as restaurants, meeting rooms or outside near busy roads. They are available in several convenient designs that can be clipped onto clothing or placed in a pocket

SPEECH/LANGUAGE THERAPY29
Speech and language therapy can help children with hearing loss improve communication skills, expressive language, speech skills and interaction skills.

The cost of testing and treatment will depend on the type of health care available, geographic location and insurance coverage.
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References


TIPS

Here are some strategies and workarounds to promote better communication with hearing loss:

- Talk to people about hearing loss. If they understand the situation, they will be more able to help enhance communication.
- Don’t be embarrassed to ask people to repeat what they have said whenever necessary.
- Try to sit face-to-face when speaking with someone—this can make it easier to decipher speech and pick up visual cues.
- When going out to a restaurant with a group, try to find a time and place that isn’t too crowded and noisy. Request a table in a quiet spot.
- Listening closely requires serious concentration and can use a lot of energy. Take a break in a peaceful place when needed.
- Connect with other people with hearing loss. They can be a great source of support and information.