

THE FABRY DISEASE GLOSSARY

When you read about Fabry disease, you may find there are difficult medical words that are difficult to understand. In this glossary, you will find a list of medical words related to Fabry disease and what they mean. If you would like a printed version of this list, you can print it by clicking the printer button below this document.



Word	How to say it	Meaning
Acroparesthesia	ak-roh-par-uhs-thee-zhuh	Aburning, tingling, or pricking sensation or numbness in the hands or feet. A common symptom in people with Fabry disease. ^{1,2}
Alpha-galactosidase A (alpha-GAL)	al-fuh-gal-ac-toos-i-deys A	An enzyme (protein) that removes a type of fat called globotriaosylceramide (Gb3) inside the body. ³ In people with Fabry disease, this enzyme is missing or does not work well, leading to a build-up of this fat in blood vessels and organs. ^{2,4}
Anhidrosis	an-hi-droh-sis	Not being able to sweat. A common symptom in people with Fabry disease, more often in boys and men. ⁵
Angiokeratomas	an-jee-oh-ker-uh-toh-muhz	Small, red or dark-blue spots on the skin. The most common skin symptom in people with Fabry disease. ⁵
Arrhythmia	uh-rith-mee-uh	Having an irregular heartbeat. ⁶ A common symptom in people with Fabry disease. ⁷
Cardiologist	kahr-dee-ol-uh-jist	A doctor who specialises in the heart and heart conditions, including the diagnosis, management and treatment of them. ⁸
Chaperone therapy	shap-uh-rohnther-uh-pee	A medication for people with Fabry disease that helps correct faults in the enzyme, alpha-Gal, so that it can break down extra fat (Gb3). ⁹
Chronic kidney disease	kron-ikkid-nee dih-zeez	A condition where the kidneys are damaged and can't filter blood as good as they should. ¹⁰
Corneal opacity	kawr-nee-uhloh-pas-i-tee	A condition where a hazy cream-coloured circle develops on the cornea (the transparent part of the eyeball that covers the iris and pupil and lets light in). ^{11,12} This usually happens when the cornea has been damaged and can lead to a loss of vision. ¹³ However, it does not usually affect the vision of people with Fabry disease. ⁷
Enzyme	en-zahym	A complex protein that is produced by living cells and is reponsible for speeding up certain chemical reactions in our body. Without enzymes, most metabolic reactions would take much longer and would not be fast enough to sustain life. ¹⁴
Enzyme assay	en-zahymas-ey	A test that measures the activity of a particular enzyme. ¹⁵ An assay for alpha-galactosidase A (alpha-Gal) is used to check whether a person has Fabry disease. In boys and men, low activity of alpha-Gal means they have Fabry disease. In girls and women, a genetic test is done to check for Fabry disease. This is because girls and women with Fabry disease may have normal alpha-Gal activity. ⁵
Enzyme replacement therapy	en-zahymri-pleys-muhnt ther-uh-pee	A medication that provides new healthy enzymes to a person that is missing that enzyme or whose enzymes do not work well. ¹⁶ Enzyme replacement therapy for Fabry disease contains the enzyme alpha-Gal. ⁵
GalactosidaseAlpha (GLA) gene mutation	guh-lak-toh-si-deys al-fuh myoo-tey-shuhn	A gene mutation is a permanent change in the structure of a gene that may change how it works. ¹⁷ The GLA gene makes possible the production of an enzyme called alpha-galactosidase A (alpha-Gal). ⁷ A mutation in the GLA gene means that it has changed and may not work as it should. People with Fabry disease have a GLA gene mutation. ²
Gastrointestinal symptom	gas-troh-in-tes-tuh-nlsimp-tuhm	Any symptom that happens in your gastrointestinal system, from your mouth to your stomach and intestines. ¹⁸ Examples of gastrointestinal symptoms include diarrhea, abdominal cramping, flatulence, a feeling of fullness earlier than normal, nausea and vomiting. ^{19,20} Gastrointestinal symptoms are common in people with Fabry disease. ¹⁹
Genetic counselor	juh-net-ik koun-suh-ler	A healthcare professional who provides information to patients and families about heritable disease. ^{21,22}
Genotype	jee-noh-taip	The set of genes that each person, animal or organism has. ²³
Globotriaosylceramide (Gb3)	gloh-boh-trahy-uh-oh-ser-ahm-ahyd	A type of fat in cells of the body that helps cells divide and communicate. ³ To keep a healthy level of this fat, the body sends an enzyme called alpha-GAL to remove this fat. ³ However, in people with Fabry disease, alpha-GAL is missing or does not work as well, so this fat builds up in the body. ²
Glycosphingolipid	glahy-kuh-sfing-guh-lip-id	A type of fat inside the cells of the body that helps cells divide and communicate.Globotriaosylceramide (Gb3) is an example of a glycosphingolipid. ³ Gb3 is the type of fat that builds up in the cells and organs in people with Fabry disease. ²
Heterozygote	heh-tuh-row-zai-got	A person, animal, or organism that has two different forms of a gene (part of a cell that contains DNA information). ^{24,25}
Hypohidrosis	hi-poh-hi-droh-sis	Sweating less than normal. A symptom that people with Fabry disease may experience, more common in boys and men. ⁵
Ischemic stroke	ih-skee-mik strohk	A condition where the blood flow to the brain is blocked. An ischemic stroke is a medical emergency and needs immediate medical attention. ²⁶
Left ventricular hypertrophy	left ven-trik-yuh-ler hahy-pur-truh-fee	A condition where the left ventricle (cavity) of the heart grows bigger, or the wall grows thicker, than it should. ²⁷ Heart conditions, such as left ventricular hypertrophy, are common in people with Fabry disease. ⁷
Lyso Gb3 (globotriaosylsphingosine)	gloh-boh-trahy-uh-oh-sil-sfing-goh-seen	A different form of globotriaosylceramide (Gb3), a type of fat that builds up in people with Fabry disease. ⁵ Lyso Gb3 is thought to play a role in Fabry disease because it speeds up the storage of Gb3. ²⁸
Lysosomal storage disorder	lahy-suh-soh-muhl stohr-ij dis-awr-der	A genetic condition where a person is missing an enzyme (protein), which leads to an abnormal build-up of certain substances in the body. Seventy lysosomal storage disorders have been discovered, and more are likely to be discovered in the future. Fabry disease is a lysosomal storage disorder. ²⁹
Medical geneticist	med-i-kuhl juh-net-uh-sist	A doctor who specialises in genetic conditions and how to manage and treat them. They can also provide counseling to patients with genetic conditions. ³⁰
Multidisciplinary team	muhl-tee-dis-uh-pluh-ner-ee teem	A group of healthcare professionals, such as a cardiologist, medical geneticist, nephrologist, neurologist, psychologist, from multiple areas of expertise that works together on a patient's care and treatment. ³¹
Nephrologist	neh-frah-leh-jist	A doctor who specialises in kidneys and kidney conditions. ³² Your kidneys are organs that filter your blood and remove waste and extra water. ³³
Nervous system	nur-vuhs sis-tuhm	The network of nerves and cells that carry the signals between the brain and the rest of the body. It enables thinking, language, feeling, learning, memory. The nervous system has 2 parts: ³⁴ 1. The central nervous system: this is made up of the brain and spinal cord 2. The peripheral nervous system: the network of nerves that extend throughout the body. ³⁵
Neurologist	noo-rol-uh-jist	A doctor who specialises in brain and nerves and how to treat conditions that affect them. ³⁶
Neuropathic pain	noo-rop-uh-thick peyn	A pain that is caused when parts of the nervous system to do with the touch sensation are damaged. ³⁷ A very common symptom in people with Fabry disease that is probably caused by a build-up of certain fats in the nerves. ³⁸
Phenotype	Phee-noh-taip	The physical characteristics of something living that can be seen. ³⁹
Psychologist	sahy-kol-uh-jist	A trained healthcare professional who uses talking therapies to treat mental health problems and help people cope with emotional, social and physical problems. ⁴⁰ If you need support to help cope with your Fabry diagnosis or symptoms, your doctor may refer you to a psychologist.
Tinnitus	tin-i-tuhs	Hearing noises that do not come from an outside source. They can sound like ringing, hissing, whistling, humming, buzzing, chirping, or clicking sounds. ⁴¹ In people with Fabry disease, doctors think tinnitus is caused by blocking or narrowing of the blood vessels in the inner ear caused by a build-up of Gb3. ⁴²
Transient ischemic attack (TIA)	tran-zee-uhnt ich-skee-mik uh-tak	A condition where the blood flow to the brain is blocked for a short period of time (usually less than 5 minutes). ⁴³ This type of stroke is also known as a 'mini-stroke'. ⁴⁴ A TIA is a medical emergency and needs immediate medical attention. ⁴³

ARE THERE OTHER MEDICAL WORDS YOU DO NOT (COMPLETELY) UNDERSTAND?

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Word	Meaning

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