HOW DO I TALK TO MY FAMILY ABOUT Fabry?
This booklet will help you talk to your family about Fabry. Every person with Fabry is different, and every family is different. So this booklet includes several different approaches to speaking with your family. Choose the approach that works best for you.

**What is family?**
There is no one-size-fits-all family. Your family is the group of people who are important to you and your well-being. They might be related to you biologically, legally (through adoption or marriage), or emotionally (through affection, dependence, obligation, or cooperation).
Why should I talk to my family about Fabry?

Fabry is a rare condition (relative to other chronic health conditions). Most people do not know what it is and may never have heard of it. You may find that even some doctors and other healthcare professionals do not know much about it.

It is important to help both your family and your healthcare providers understand what Fabry is, what it means for you, and how your diagnosis might impact your family. Some topics to share are major signs and symptoms and how the condition is passed down in families.

Sharing your story will help your relatives understand their family health history and possibly even their own personal history. Sharing the knowledge of your symptoms and the reason, or diagnosis, for them (i.e., Fabry) may help relatives make sense of their own physical experience, especially if they have symptoms medical professionals have been unable to explain.

In turn, it is helpful for you to know your whole family health history, including Fabry and other conditions that might run in your family. Putting the whole picture together will help you and your relatives:

- Identify risks due to shared genes.
- Talk to each other about health (including quality of life).
- Summarize health information to give to healthcare providers, as well as colleagues, teachers, and others.
What is Fabry?

Fabry is an inherited condition that causes deposits to build up in cells over time. Our bodies contain thousands of active substances called enzymes. For example, your stomach has enzymes that help break down the food you eat. Some of these enzymes are stored in structures called lysosomes, which help the body break down proteins, carbohydrates, and fatty substances.

In people without Fabry, the enzyme alpha-galactosidase A (pronounced al-fa-ga-lak-toe-sy-daze A), or alpha-GAL, breaks down a fatty acid called globotriaosylceramide (pronounced glow-bow-try-oh-sill-ser-ah-mide), or GL-3. People with Fabry do not have enough of the alpha-GAL enzyme. As a result, lysosomes fill up with GL-3 and no longer work normally. This is why Fabry disease is called a “lysosomal storage disorder”.

The build-up of GL-3 in the walls of blood vessels and other tissues can cause damage to major organ systems. The heart, kidneys, and brain may not function properly, causing problems that could be life-threatening.

How is Fabry passed on?

Fabry is an X-linked condition. This means that the gene that makes alpha-GAL is found on the X chromosome, one of the sex chromosomes. Males have one X and one Y, and females have two Xs. Males pass an X chromosome to their daughters and a Y chromosome to their sons. Women pass an X chromosome to all of their children, daughters and sons. Therefore, men with Fabry cannot pass it to their sons but will pass it to their daughters, and women with Fabry can pass it to both their sons and daughters.
My name is Alex, and I have Fabry. No one on my dad’s side has Fabry, but lots of relatives on my mom’s side do.

My grandfather, Joe, had it. He was diagnosed in his early 50s and died soon after. We think his two brothers also had it (they both passed away in their 40s). Grandpa Joe passed the gene change to my mother and all her sisters, Sarah, Maggie, and Jane.

My mom passed the gene change to me, and Aunt Sarah passed it to my cousin, Katie. Aunt Maggie didn’t have any children, and Aunt Jane passed the Fabry gene change to two of her children, Nick and Megan.
Who is at risk?

- Fabry occurs in both men and women, but it often affects males more severely than females.
- Fabry affects people of all races and ethnicities, all over the world.
- Women with Fabry have a 50% chance of passing it to their children – both daughters and son – during each pregnancy.
- Men with Fabry will pass it down to all of their daughters and none of their sons.

Major signs and symptoms

- Fatigue
- Burning pain in the extremities
- Impaired sweating
- Starburst pattern on the cornea (corneal whoring)
- Skin rash, or angiokeratomas
- Hearing problems
- Gastrointestinal problems
- Kidney problems
- Heart problems
- Stroke
The signs and symptoms of Fabry may appear in affected individuals at any age, and often appear later in females than in males. Family members with Fabry – even siblings – may have very different symptoms.

**Diagnosis**

It is important to know if you have Fabry disease, because Fabry can cause many health problems, including pain, heart problems, and kidney problems. Because Fabry disease is progressive, early diagnosis and access to appropriate medical care is especially important. However, Fabry is not very well known, and many people endure months and years of misdiagnosis before receiving a definitive diagnosis of Fabry. Many people experience symptoms of Fabry early in childhood – perhaps as early as four years old. Unfortunately, these early signs and symptoms are sometimes misunderstood, or even ignored, by parents, teachers, and other caregivers. This can lead to delays in seeking medical care as well as possible feelings of anger, self-doubt, depression, or even hopelessness at not being taken seriously (for more about these psychological reactions and others, see page 12).
Testing

Males with Fabry can be diagnosed through a simple test that indicates whether or not the enzyme alpha-GAL is present in the blood. Females, on the other hand, may have Fabry even if they have nearly normal levels of alpha-GAL. Therefore, genetic testing, which analyzes DNA, is needed to determine whether a female has Fabry. Genetic testing for Fabry involves a blood draw, and there are some psychological considerations. See page 10 for more on emotional and psychological issues.

If you are considering being tested for Fabry disease, genetic counselors can provide support before, during, and after your test.

Genetic counselors are specialized healthcare professionals with experience in medical genetics and counseling. They are trained to help individuals as they consider being tested, when the results are received, and in the weeks and months afterward.

Before testing, counselors can provide balanced information so you can make an informed decision about testing. They can also help both individuals and family members to be psychologically prepared to cope with a positive test.

If test results are positive, counselors can help the individual and the family adjust to the test results, and can assist them with understanding whatever specialist follow-up is necessary.
What is family health history?

Family health history is a collection of information about health conditions that run in your family, as well as the eating habits, activities, and environments that your family shares. Your family’s health is one part of the entire history of your family. Knowing about health conditions in your family can help you make healthy choices.

How can family health history affect my health?

You inherit many things from your parents and grandparents. They pass on culture and values through photos, recipes, stories, spiritual practices, and music. You also inherit some parts of how you look—for example, your height and the color of your eyes. Small structures in cells called genes carry information for these characteristics and guide your body through growth and development. Changes in genes may also lead to a risk in the family for developing certain health conditions, such as Fabry.

Sandra’s Story

Forty-four people in my family have Fabry. To me, knowledge is everything; I know the heartache and turmoil growing up without knowing why my family members were so sick.
Tell me more about my genes

Genes are the instructions inside each of your cells. Since everyone has slightly different genes, everyone has a different set of instructions. Genes are one reason why you are unique!

- A person has two copies of each gene, one from the mother and one from the father.
- Genes carry instructions that tell your cells how to work and grow.
- A change in a gene is called a mutation.
- Genes are inside cells. Every part of your body is made up of billions of cells working together.
- Genes are arranged in structures called chromosomes. Most people have 23 pairs of chromosomes. Copies of the chromosomes are found in each cell. There are 22 numbered pairs of chromosomes. The last pair consists of the sex chromosomes, called the X and Y chromosomes. Males have one X and one Y. Females have two Xs.
- Chromosomes are made up of DNA. DNA is the special code that spells out the instructions in your genes.

But everyone in my family handles the information differently. Some share a lot, and others aren’t interested in knowing about it. They are scared to be involved. Some want to learn more, and others think we are hypochondriacs, or crazy. However, we are still a close family; we stick together and support one another. We just pick and choose what we talk about.
How are individuals affected?
Receiving a diagnosis of a genetic condition can be a life-changing event for individuals and families. Fortunately, much has been learned about Fabry, such as what causes it and how its symptoms and psychological effects can be managed.

How are males and females affected differently?
Fabry was once thought to affect only males. Females were called “carriers” because it was believed that they could carry the gene change without displaying any symptoms. It is now recognized that all females with the gene change have Fabry. Women experience a wide range of symptoms that differ from person to person.

Why this variation? When a male inherits an X chromosome with the gene change, he produces little or no alpha-GAL, and he develops the symptoms of Fabry. Females have two X chromosomes, so even if one has the Fabry gene change, she has a second gene that could produce alpha-GAL. However, in each cell, only one X is “active” or “turned on”. Which X is active in any given cell is determined randomly through a process.

Mike’s story
I used to be a total workaholic and never got to see my kids. When I stopped working, I finally had time to get to know them, and we became very close. My wife has taken care of me through all of the pain and suffering. She is there for me, and she has a lot to do with me still being alive. I’ve known my best friend since we were teenagers, and we’d do anything for each other. He’s like my brother.
called X inactivation. Each organ in a woman’s body has its own X inactivation pattern.

Statistically, the X with the gene change should be active in 50% of a female’s cells. However, like flipping a coin, 50-50 odds do not always result in a perfect 50-50 split. In a female with the Fabry gene change, the X inactivation pattern may result in 60% of the normal Xs functioning in one organ, but only 40% in another. Put simply, the more normal X’s that are shut off, the more likely she is to have symptoms. Therefore, the severity and type of Fabry symptoms vary from woman to woman.

Unfortunately, some women are still told, “You are just a carrier,” and are not recommended for appropriate medical care. It is not true that women are merely carriers of Fabry. What is true is that a female’s personal X inactivation pattern can cause her to have some Fabry symptoms but not others. Symptoms vary in females more than they do in males, ranging from mild to severe, and often start later in life. However, all women with the gene change do have Fabry.

This disease involves a lot of time and patience with the people who have it. It helps a lot to have a support system. Many people with Fabry don’t have one because people don’t understand this disease.
Quality of life: Psychological and emotional issues

Many people with Fabry experience feelings of depression, hopelessness, isolation, and denial of their symptoms. Both males and females with Fabry have reported lower quality of life than the general population, especially in adulthood. As a person with a rare disease that has no “visible” symptoms on the outside, it can be hard for people with Fabry disease to explain their difficulties to other people, including employers, teachers, and friends, among others. It may even be hard to convince medical professionals to take them seriously, due to the “vague” nature of their symptoms, the difficulty diagnosing Fabry using many standard medical tests, and the lack of widespread knowledge of the disease. People with Fabry may have faced years of being accused of being “lazy” or “making it up” by a variety of people in their life before they are finally accurately diagnosed.

There are also emotional issues associated with testing for Fabry and dealing with the results of these tests. For some it may come as a relief to finally have an explanation for their puzzling symptoms.

Nelson Family story

There is a lot more information available about Fabry now than when we were diagnosed a decade ago. Back then we believed everything that was told to us, but now we know what to question. Doctors come and go, so we do our own research.
We're stronger, and it's not as scary as it used to be. Power and education are what it comes down to. We always have to pay attention. Every year we go talk to the girls' schools, to new nurses and teachers. When there are field trips outside we have to make sure there’s a plan because the girls don’t sweat. Sarah has had a different gym teacher each year, so she has to explain. It gets frustrating answering the same questions over and over. But if you keep a positive mindset, it makes everything a lot better.
Living with Fabry

For others, learning that they may be likely to develop symptoms themselves or pass a serious disease on to their children can be a difficult experience. Some people who test positive for the Fabry gene may become angry or fearful, while their close relative who tests negative for the gene may feel simultaneously relieved and guilty. All of these emotions are normal. In such cases, it may be helpful to talk with someone else from the Fabry community—someone in a similar situation with similar concerns and fears. It is important to reach out to get the support you need as well as an idea of what you can do to take control of your disease.
Just as our physical health affects us emotionally, so too can our psychological health affect us physically. This can happen either directly (for example, anxiety can contribute to feelings of chest pain or “butterflies” in the stomach) or indirectly (such as when feelings of depression result in our not taking proper care of ourselves or following medical advice).

It is important to pay attention to our emotional and psychological health as well as our physical health. This can include building a supportive network of friends and family, as well as talking with other people with Fabry through support networks and organizations. Actively learning more about Fabry and exploring how to manage your symptoms with genetic counselors and physicians can help alleviate feelings of uncertainty and confusion. Counseling with a therapist or health psychologist may be very helpful or even recommended in some circumstances. All of these things can help people with Fabry take back control of their life and their disease at a time when they may feel things are out of control.

Finally, don’t be afraid to ask for or accept support! There are a lot of different ways people in your life can help. Types of support can include practical support (help with transportation to doctor’s appointments or mowing the yard), emotional support (someone you trust and can turn to with worries or for reassurance), and appraisal support (someone who can help you figure out what is happening and how to cope with it). Any and all of these can contribute to not only our psychological health but that of our loved ones as well.
How are families affected?

Fabry is an inherited condition. This means it is passed on through generations. (Turn to page 3 for a chart showing how Fabry is passed down.) If you or someone in your family has Fabry, other family members, adults or children, may also have Fabry and not know it. This includes siblings, parents, and children as well as more distant relatives such as cousins. Genetic counseling for families with Fabry is important to help determine who should be tested for Fabry. As a simple example, if it is determined that you inherited Fabry from your mother, then your father’s side of the family would not need to be tested.

Knowing your family health history can help you make smart choices for yourself and your family. For example, you can:

• Make sure you and your family members are tested for Fabry.
• Get the right information before making important reproductive choices.
• See your doctor for an evaluation to determine how you may be affected.

Kathy’s story

My father was the first one to be diagnosed with Fabry. Men in my family all died young, but everyone had a hard time accepting the fact that there was anything genetically wrong with them. Many would not accept that there was even such a thing as Fabry.
Adoption
Individuals with Fabry who are adopted might grow up without knowing anyone else with Fabry. They don’t have any examples of what life is like with Fabry. This can make the person feel alone and isolated. Turn to page 30 for organizations that offer information and support.

Denial
Fabry is a rare disease, and some people are reluctant to admit that such a condition could be part of their family history. This led to a culture of silence in many families. It was easier to ignore the condition than to acknowledge something that could kill you.

We were told that it didn’t affect the girls, so we shouldn’t worry. But looking back, I understand that I do have Fabry. It was in high school that I started noticing it. Because of shortness of breath I could never run or square dance with my cousins. I always blamed it on being out of shape. When my son, Jack, started the Fabry Support and Information Group, we realized the number of females that are affected. It has been a focus to get the medical profession to acknowledge that women are not just carriers.
Many people in families with Fabry disease have already seen close relatives become affected with the disorder. Learning that you or a family members carry the gene change can lead to feelings of depression and hopelessness. Despair often leads to denial that the individual may be affected and can cause someone to delay seeking medical care. (Turn to page 26 for what to do if your family does not want to talk about Fabry.) Guilt at the idea that you may have passed Fabry along to your children can also contribute to denial and delay the testing and diagnosis of children, which can be particularly detrimental.

Cultural Differences

Different cultures have different perceptions about health and disease. In some cultures, discovering that there is genetic disease in the family may be considered God’s will. In other cultures, it might be seen as diminishing the family’s worth and can often be a source of guilt, shame and embarrassment. Because of these reactions, family members can be reluctant to talk about their diagnosis, even within the family, and may be slow to respond to their health issues and/or seek advice from a doctor or healthcare provider.

Shane’s story

Fabry has been rough on me. My mom’s family is from India, where it’s common practice not to talk about your afflictions. You can talk about small things like a headache or cold, but if it’s something that affects your life and affects other people’s lives, it’s taboo.
I have symptoms almost every day—most often burning in my hands and gastrointestinal problems. My mom and I talk about it openly, but if I’m in pain at family gatherings, I can’t explain myself.

Fabry has also given me a lot of opportunities. I have been fortunate to travel to conferences and meet people I never would have met. I plan to be a genetic counselor. I have dealt with genetic counselors all my life, so I know the great things they do to help people. My disease pointed me in the direction of where I want to go.
Why should I talk to my family about Fabry?

You share a lot with your family—including what can make you sick. Family members can have genes, habits, diet, and environment in common, so they share risk for the same health problems.

Most common diseases (heart disease, diabetes, etc.) are known as “multifactorial” conditions. They are caused by a combination of genes, lifestyle, and environment. In these cases, people with similar genes may not develop the same illness if they make different choices or live in a different environment.

On the other hand, some conditions are caused by a specific change in the DNA of a single gene, called a mutation. Many of these conditions, like Fabry, are rare. In these cases, genes play a much bigger role than lifestyle and environmental influences. These conditions usually develop when an individual is born with a specific gene mutation(s).

It is important to discuss all types of health conditions—common, rare, mild, fatal, acute, and chronic—with your family and doctors. Knowing about the conditions that run in your family can help you make informed decisions about your health.
How do I talk to my family about Fabry?

Fabry disease may cause different symptoms than common conditions like heart disease, diabetes, and cancer, but the steps you take to talk to your family about it are similar.

Talk to your family

Your relatives are the best source of information about your family. This means you are a source of information for them in return. Events like birthday parties, weddings, reunions, religious gatherings, holiday dinners, and funerals provide an opportunity to talk to family members about their lives. Share your purpose. Explain that the information you are sharing can help not only you but the whole family to receive better healthcare. You can ask family members questions about their health and tell them what you know about yours.

Use what you have

Be sure to have within easy reach your test results, letters from your doctor, or other information you have about your diagnosis, which can be shared with interested family members to help them understand your disease. It may help them understand their chances of having Fabry as well or the chance of children inheriting Fabry from their parents. Websites and brochures that have been useful for you may also be useful for your family members. This booklet can be passed along to help explain family health history and Fabry.

Plan an individual conversation

After you have brought up Fabry, you may want to talk with certain family members in more detail. Some people respond better to one-on-one talks rather than conversations in a large group setting. This will give you the chance to address any questions they have and ask questions of your own.
Talking about Fabry

These individual talks should take place when both you and your family member feel up to it: you should not feel rushed or tired. Give the conversation the energy and attention it deserves. Also make sure your family member does not feel threatened or pressured to talk about everything right then. Let him or her know you are open to questions and that it is not a one-time conversation; the discussion can continue in the future.

Bring support

If you have a friend or family member who is a medical professional or who understands Fabry and has been a help to you throughout your diagnosis, it could be useful to have him or her there for support during the conversation.

Send a letter

Some people may be more comfortable sharing and receiving health information in a letter or email. You may wish to send your family a message with an update on your health information and information about Fabry. Your doctor or genetic counselor may be able to help you write this letter and identify who in the family would benefit from being informed. A sample letter is on the next page to get you started.
Dear Family,

Many of you know that ______ has not been feeling well and that we have been involved in a medical work-up to try to find out what is wrong. We appreciate everyone's concern and support and want to share with you the results of our search.

These results are important for you to consider since they involve a genetic condition (Fabry disease). Some of you are unlikely to be impacted by the disease, but it may affect other family members, so it is something of which you should be aware.

______ has been diagnosed with Fabry disease. Fabry is a serious disease that can involve severe complications with a number of organs in the body.

The Fabry mutation is carried on the X chromosome. (Females have two X chromosomes, males have an X and a Y chromosome.) This means that both sons and daughters of a woman who has the Fabry mutation have a 50% chance of inheriting the mutation. Sons of a man who has Fabry and a woman who does not have Fabry will not inherit the Fabry mutation because they receive the Y chromosome from their father and the X chromosome from their mother. (The Fabry mutation can only occur on the X chromosome.) However, all daughters of a man who has Fabry will inherit the mutation.

That means in our extended family the following people are at risk of having inherited the Fabry mutation: ____________________________________________.

We recommend that everyone listed above get tested for the Fabry mutation. I will be glad to discuss this further with any of you who wish to do so. I'm sorry to have to share this news, but I believe it is important for you to be aware of the situation to make an informed decision about your health.

One note of caution is that you may find medical literature that says that females do not suffer from Fabry symptoms. This is no longer considered correct by the medical profession; females, as well as males, should be evaluated if they believe they could have inherited the Fabry mutation.

Our thoughts are with each of you.

Warmest regards,
How do I prepare for the talk?

Know the facts
Know the facts about Fabry so you can answer any questions your family members have or refer them to other resources. Use your own experiences and information you learned from your doctor and others. Remember, Fabry varies from person to person, so talk to your healthcare provider for medical advice. (Turn to page 2 for more details on Fabry that you can share with family.)

Know how it might impact your family
For each family member you talk to, understand what your diagnosis might mean for him or her. Because Fabry runs in families, it is possible that some of your relatives have Fabry themselves. (Turn to page 3 for a chart showing how Fabry is passed down.)

Get ready for different reactions
Be sensitive to a person’s wishes not to talk about certain topics. Some relatives might not be comfortable hearing about Fabry or other health conditions. Make it clear that you are available to talk whenever they are ready and that you will respect their wish not to talk in the meantime.

Conversation starters
There is not one right way to talk to your family about health and Fabry. After you tell your family your story, you might want to let them know what symptoms people can have that may be signs of Fabry.
Health problems caused by Fabry:

- Fatigue
- Burning pain in the extremities
- Impaired sweating
- Starburst pattern on the cornea
- Skin rash
- Hearing problems
- Gastrointestinal problems
- Kidney problems
- Heart problems
- Stroke

Questions about family to help build a family health history

- Do you know if your parents or grandparents took medicine on a regular basis? If so, what kinds and for what?
- What kinds of things have people in your family died from?
- Has anyone had problems in pregnancy or childbirth? What kinds of problems?
- Are there any diseases that run in our family?
- Is there anything else you would like to tell me about your life or health concerns in our family?
Talking about Fabry

**What should I do if my family does not want to talk about Fabry?**

Some family members might not understand why it is important to know about Fabry. Others might be nervous about receiving a diagnosis of Fabry themselves. It is normal to feel anxious or uncomfortable if something is unfamiliar. We are all at different emotional places at different times and will process the information differently.

You may wish to start the conversation by mentioning more common conditions that run in your family, like high blood pressure or asthma. Then move on to Fabry. Make it clear that although there is no way to lower your risk for Fabry, you can still lead your life normally, with hobbies, a career, and a family.

Try to get family members involved, even if they express initial reluctance or denial. If they are not willing to take steps to learn more on their own behalf, they might be willing to do it for their children or the rest of their family.
If family members remain unwilling to talk about it, respect their wishes. Denial can be a way for some people to cope. Just because someone does not seem to be listening to you doesn’t mean they do not hear what you are saying. Give them the time and space to think about what you have said and become more comfortable with the idea of Fabry.

You may find that some family members won’t know how to respond to you about Fabry right now, but they may be able to have the discussion in the future. If this happens, they will be better prepared and able to respond if they have already heard some of the messages about Fabry or have been directed toward information sources they can explore privately on their own in the meantime.
Talking about Fabry

Let them know you are available to talk when they are ready and if they have questions, and give them some resources for when they are ready to learn more. (Refer to page 30 for resources.)

When family members do not want to talk about Fabry, you may feel hurt, upset or alone. Try to understand that it is not unusual for family members to react to and cope with the news differently. It does not mean they don’t care. When this happens, it is all the more important to seek support from doctors, friends, other family members or other people you know with Fabry.

Finally, even if a family member decides not to get Fabry testing done, for whatever reason, they can still be encouraged to have their doctor closely monitor their heart and kidney function. These are among the organ systems most often affected by the disease.

Cheryl’s Story

I’m adopted and never knew anyone with fabry. My biological mom told me that my biological father had Fabry when I was a teenager. I had a lot of fevers as a child, and I thought everyone’s hands and feet burned when they had a fever.
I only knew what I had heard about Fabry, but I became the advocate for the family. I keep doing more research because I’m worried about my grandson. We don’t know what to expect for him now that he’s a teenager.

Everything is a learning experience. I have to manage my energy levels and know when to slow down. My family helps out around the house, but Fabry affects how much quality time I can spend with them. When I feel that I am becoming fatigued, or the pain crisis is starting, I rest more. I just try to be on top of it, so it doesn’t get on top of me!
Resources & Glossary

Resources
Learn More and Get Involved
Fabry Support & Information Group: www.fabry.org
National Fabry Disease Foundation: www.thenfdf.org
Fabry Community: www.fabrycommunity.com

Diagnostic Testing and Registry
GeneTests: www.genetests.org
Fabry Registry: www.fabryregistry.com

More Information
Genetic Alliance: www.geneticalliance.org
National Organization for Rare Disorders: www.rarediseases.org
National Society of Genetic Counselors: www.nsgc.org
Genzyme Medical Information: 800.745.4447, option 2

Glossary
Alpha-galactosidase A (alpha-GAL)—An enzyme that is missing, ineffective, or present in inadequate amounts in people with Fabry disease. It is normally found in the lysosomes.

Angiokeratomas—Dark-red or blue skin lesions, which may be flat or slightly raised, that are usually found in the area below the waist and above the knees in patients with Fabry disease.
Chromosome—A threadlike strand of DNA and protein in every plant and animal cell. Chromosomes carry the genes that define a person’s bodily makeup (for example: hair and eye color, disease expression).

Cornea—The thin transparent outer covering of the eyeball.

Corneal—Of or relating to the cornea.

DNA—An abbreviation for deoxyribonucleic acid, the storehouse of all hereditary characteristics. All chromosomes are made up of genes, and genes are made of DNA.

Enzyme—A protein produced by the body that acts to chemically change other substances. Enzymes are involved in breaking down or chemically altering substances so that the body can use or excrete them. Enzymes are typically named by adding an “ase” to the end of a word that describes them.

Fabry disease—A genetic disorder caused by a deficiency of the enzyme alpha-galactosidase A.
**Gene**—A piece of DNA that codes for a particular substance. Each gene occupies a specific location on a chromosome, which defines a person’s bodily makeup and function.

**Genetic**—Affecting or affected by genes (genetic disorder).

**GL-3**—An abbreviation for globotriaosylceramide.

**Globotriaosylceramide**—One type of glycosphingolipid compound which accumulates in the blood vessel walls of people with Fabry disease as a result of a deficiency in alpha-galactosidase A.
**Lysosome**—A small structure in most cells acting as the chemical plant of the cell. Lysosomes contain and make various enzymes that digest or break down substances. In Fabry disease, Gl-3 builds up in the lysosomes.

**Lysosomal storage disease**—A disease resulting from the storage or accumulation of material in the lysosomes. Fabry disease is a lysosomal storage disorder.

**Protein**—The building blocks of cells involved in all essential life functions (for example: cell growth, energy production, metabolism).

**Whorling**—A starburst pattern commonly found on the cornea of people with Fabry disease.

**X chromosome**—The sex chromosome associated with female characteristics. All females typically have two X chromosomes while males typically have only one X chromosome.

**X-linked disease**—A genetic disease whereby the defective gene is carried on the X chromosome. Fabry disease is an X-linked disease.