Gastrointestinal problems

Many people with Fabry disease experience stomach problems, which may range from mild to severe. These disturbances may include pain after eating a meal, diarrhea, and nausea.

Heart problems

Heart problems are also common to Fabry disease and may become progressively worse with age. This is because GL-3 accumulation becomes greater over time and may cause increasing damage to the tissues and to the blood vessels supplying the heart. Heart problems may include:

Enlarged heart
Malfunctioning heart valves
Irregular heartbeat
Heart attack
Heart failure

Kidney problems

After years of GL-3 buildup, problems with the kidneys can develop, and kidney function can be compromised. Kidney damage can become so severe that the kidneys may lose some of their functionality (renal insufficiency) or may fail (renal failure). Thus, GL-3 accumulation in the kidneys may represent a major health risk for those with Fabry disease, and can be present in the absence of Fabry symptoms. However, kidney problems are not unique to Fabry disease. Often it is other signs and symptoms (like Fabry pain and angiokeratomas) that may lead a doctor to suspect Fabry disease.

Problems with the nervous system

Significant GL-3 accumulation can thicken small blood vessels in the brain. As a result, people may experience a number of symptoms including:

Weakness
Head pain
Numbness
Dizziness
Stroke

Psychological and social issues

Living with difficult physical symptoms is only one of the challenges people with Fabry disease may face. They may experience fear, depression, isolation, or guilt about passing the disease along. Family members may be affected as well.

To help people cope with these feelings, there are support groups specific to Fabry disease as well as groups for rare diseases generally. For help in locating a support group or to read about how patients cope with the disease, visit the web at FabryCommunity.com

The Kidney & Urology Foundation of America, Inc. is a national, not-for-profit organization dedicated to helping people avoid the debilitating effects of kidney and urologic diseases – disorders affecting as many as 70 million Americans today.

By funding research, medical conferences, and interdisciplinary hospital initiatives, the Kidney & Urology Foundation provides the leadership necessary to promote a better understanding of kidney and urologic diseases.

We are committed to making research and resources accessible to patients and their families, as well as to caregivers, and we measure our success by our ability to respond meaningfully and personally to the public and to people in need.

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KIDNEY UROLOGY Foundation of America, Inc.™

No. 5
Printed
Sept. 2008

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Fabry Disease Information for Patients and their Families

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Fabry Disease Overview

Fabry disease is a rare hereditary disorder caused by a faulty gene in the body. It affects more males than females: It is estimated that 1 in 40,000 males has Fabry disease, whereas the estimated prevalence in the general population is 1 in 117,000 people.

Hereditary (or genetic) disorders are those that are passed down from parents to their children through the genes. One or both parents may carry an abnormal gene that, when passed along to their children, can result in disease. Since the Fabry disease gene is located on the X chromosome, the disease primarily affects males (although some females can also experience symptoms).

When people inherit the abnormal gene that causes Fabry disease, their bodies cannot produce enough of an important enzyme called alpha-galactosidase A (pronounced al-fa-ga-lak-toe-si-daze a) or alpha-GAL. Alpha-GAL is needed to clear certain cells in the body of a fatty substance called globotriaosylceramide (pronounced glow-bow-tri-oh-syl-ser-a-mide) or GL-3.

Alpha-GAL helps clear GL-3 from the cells by breaking it down into particles small enough to leave the cell and enter into the blood stream. Once these particles enter the blood stream, they are either eliminated or reused to build other substances.

Without enough alpha-GAL, GL-3 cannot be broken down into smaller particles. Therefore, GL-3 cannot leave the cells and instead accumulates inside. Over time, this buildup of GL-3 causes damage to the cells.

The most commonly affected cells are found in blood vessels and tissues of the kidney, heart, skin, and brain. The buildup of GL-3 in these cells can eventually lead to life threatening problems.

Because Fabry disease is rare and causes a wide variety of symptoms, it can be mistaken for other diseases. Therefore, people may have the disease for a long time before it is accurately diagnosed. This is a concern because the longer a person has Fabry disease, the more damage is likely to occur in the body's organs and tissues and the more serious the person's condition may become. The earlier Fabry disease is diagnosed, the earlier doctors can start treatment to manage symptoms and try to prevent further health problems.

One way to increase the likelihood of early diagnosis is to learn about the disease and who is at risk of developing it. For example, Fabry disease is inherited and people whose family members have the disease are more likely to have the disorder than others without a family history. It may also help to learn about the symptoms of Fabry disease, as well as how and when these symptoms may arise.

Fabry Disease Diagnosis

The earlier Fabry disease is diagnosed, the earlier health care providers can begin treatments to help manage symptoms and disease-related complications. However, because Fabry disease is uncommon and its symptoms are not well recognized or may be mistaken for those of other illnesses, diagnosis can be challenging.

The following symptoms are common to Fabry disease and may prompt physicians to suspect and test for Fabry disease: burning pain in the hands and feet, temperature intolerance, a purple-red rash in the midriff area (angiokerotoma), corneal whorling, and episodes of intense pain (Fabry crisis).

If you or a family member experiences any or all of these symptoms, consult with your doctor. After an examination, he or she can determine whether to run a test for Fabry disease. This test, called an enzyme assay, measures the amount of alpha-GAL enzyme activity in the blood.

Because Fabry disease is an X-linked genetic disorder, it is more common in males than in females. However, females can have mild to moderate (or sometimes even severe) disease or can be asymptomatic carriers of the Fabry gene. Generally, enzyme assay testing is not useful for diagnosing Fabry disease in females. Instead, genetic testing can be done to either diagnose the disease in females or identify female carriers.

For more information about tests for Fabry disease, contact the Kidney & Urology Foundation of America at 800.633.6628 and we will be happy to direct you.

Signs & Symptoms

Because Fabry disease is rare, its symptoms are not always associated with the disease. There are, however, a number of signs and symptoms that are commonly experienced by people with Fabry disease. These may include:

- Pain
- Exercise intolerance
- Corneal whorlin
- Heart problems
- Nervous system problems
- Impaired sweating
- Skin rashes (angiokerotomas)
- Gastrointestinal problems
- Kidney problems
- Psychological and social issues

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Most of these symptoms can be attributed to GL-3 accumulation and the resulting damage to various parts of the body. Many of these symptoms are also common to other diseases, which may lead to misdiagnosis. In addition, people with Fabry disease may have all or only a few of the typical signs and symptoms. They may also experience different symptoms at different times throughout their lives. That's why it's important to discuss your symptoms and any risk factors that you may have (for example, family members with known or suspected Fabry disease) with your doctor.

Pain

Pain is one of the more common symptoms of Fabry disease and is often one of the first symptoms people experience. There are two major types of pain associated with Fabry disease:

1. Ongoing burning, tingling pain, and discomfort. This type of pain is called acroparesthesia, and mainly affects the hands and feet.
2. Occasional episodes of intense, burning pain. These usually start in the hands and feet and often spread to other parts of the body. These are called "Fabry crises" and can be debilitating. Fabry crises may last anywhere from minutes to several days.

Pain can be brought on by changes in weather, exposure to hot temperatures, stress, exercise, and/or fatigue.

Impaired sweating

Many people with Fabry disease sweat less than people without the disease. They may either perspire very little (hypohidrosis) or not at all (anhidrosis). This can cause overheating, frequent fevers, and sensitivity to weather extremes. Impaired sweating is generally caused by damage to the nerves and sweat glands.

Exercise intolerance

Some people with Fabry disease are unable to tolerate physical exertion, and may tire or become overheated even after mild activity. Physical exertion may also trigger episodes of pain. For these reasons, people with Fabry disease may need to modify their physical activities and/or avoid certain activities all together.

Skin rashes

One of the most visible signs of Fabry disease is a reddish-purplish rash called angiokeratoma. This rash is characteristic of the disease and may lead doctors to suspect Fabry disease. Angiokeratomas are generally located between the navel and the knees (doctors call this "bathing trunk distribution"), and sometimes in areas where the skin stretches, like elbows or knees. Angiokeratomas usually appear during adolescence and can become larger and more numerous with age.

Corneal whorling

Corneal whorling is a starburst pattern on the cornea of the eye. The starburst, or whorling, does not affect vision and can only be seen through a process called slit-lamp ophthalmoscopy, which is often part of a routine eye exam. Corneal whorling is caused by GL-3 deposits in the blood vessels of the eye and is another symptom that may lead a doctor to suspect the presence of Fabry disease.