What is Fabry Disease?

Fabry disease (FD) is an X-linked genetic disease caused by a deficiency of the enzyme α-galactosidase A (α-Gal A) in the body. The enzyme α-Gal A's function is to break down a fatty substance called globotriaosylceramide (or GL3). When α-Gal A is absent, GL3 builds up in the blood vessel walls throughout the body. As the abnormal storage of GL3 increases with time, the body's blood vessels become narrowed, leading to decreased blood flow and undernourishment of the tissues. This abnormal process occurs in various cell types and in blood vessels throughout the body, particularly those blood vessels in the skin, kidneys, heart, brain and nervous system. The early symptoms of Fabry disease, which usually begin in childhood, include decreased sweating, heat intolerance, proteinuria, a reddish-purple skin rash (angiokeratoma), severe pains in the hands and feet, hearing loss, chronic fatigue, depression, anxiety, and gastrointestinal issues such as chronic diarrhea. Fabry disease symptoms affect both women and men. Since the disease is progressive, untreated Fabry disease results in many severe health problems such as kidney failure, heart problems including enlargement of the left side of the heart (left ventricular hypertrophy) and valve disease, and cerebrovascular problems such as a stroke. Not every person with Fabry disease will have all the same symptoms of disease progression; however, without treatment the disease always gets worse over time. (1)

How do you know if you are depressed?

Everybody has times when they feel sad or low. However, in some individuals the “blues” can be more severe and last longer than usual. This is called depression. Depression is an illness that involves the body, mood, and thoughts. It affects the way a person eats and sleeps, the way one feels about oneself, and the way one thinks about things. Depression is not a sign of personal weakness or a condition that can be willed or wished away. People with a depressive illness cannot merely "pull themselves together" and get better. Depression often interferes with normal functioning and causes pain and suffering not only to those who have a disorder, but also to those who care about them. Signs that suggest depression include:

- Persistent sad, anxious, or "empty" mood
- Feelings of hopelessness, pessimism
- Feelings of guilt, worthlessness, helplessness
- Loss of interest or pleasure in hobbies and activities that were once enjoyed, including sex
- Decreased energy, fatigue, being "slowed down"
- Difficulty concentrating, remembering, making decisions
- Insomnia, early-morning awakening, or oversleeping
• Appetite and/or weight loss or overeating and weight gain
• Thoughts of death or suicide; suicide attempts
• Restlessness, irritability, and anger
• Persistent physical symptoms that do not respond to treatment, such as headaches, digestive disorders, and chronic pain.\(^{(4)}\)

Not everyone who is depressed experiences every symptom listed above. Some people experience a few symptoms, some many. Severity of symptoms varies with individuals and also varies over time. Men affected by depression may appear to be irritable, angry, and discouraged rather than sad. A depressed child may pretend illness, refuse to go to school, get into trouble at school, cling to a parent, or worry that the parent may die. Many individuals with Fabry disease are affected by depression.\(^{(3,7)}\) Depression is a symptom of Fabry disease and is treatable.

**What signs indicate that you are anxious?**
Many people feel anxious before exams, presentations, or first dates. Individuals affected by anxiety disorders have constant, unremitting worry and fears about everyday activities. Signs and symptoms of an anxiety disorder include:

• Trembling or feeling shaky
• Muscle tension, aches or pains
• Restlessness
• Shortness of breath or feeling smothered
• Increased heart rate
• Sweating or cold/clammy hands
• Dry mouth
• Lightheadedness
• Hot flashes or chills
• "Lump in throat' sensation
• Nausea, diarrhea or abdominal distress
• Feeling keyed up or unable to relax
• Easily startled
• Poor concentration
• Trouble falling asleep or staying asleep
• Irritability

Anxiety disorders also often involve anticipating the worst, even though there is little reason to expect things to go badly.\(^{(5)}\) Individuals with Fabry disease are often affected by anxiety.

**What are panic attacks?**
Panic attacks are repeated episodes of intense fear that strike often and without warning. Symptoms of Panic attacks include any of the anxiety symptoms listed above plus:
• fear of dying
• fear of going crazy
• chest pains
• feeling like you are not yourself or that things are not really happening to you
• numbness/tingling sensations
• feeling like you are choking

Panic attacks can occur at any time, even during sleep. An attack generally peaks within 10 minutes, but some symptoms may last much longer. You may genuinely believe you're having a heart attack or losing your mind, or on the verge of death. Panic attacks can be part of Fabry disease, and should be addressed, not ignored. 

How is depression treated?
Appropriate treatment can help most people who suffer from depression. Current treatments include medications and talk therapies with therapists that ease the impact of depression. The first step in determining a treatment path is a psychological evaluation by a psychiatrist and/or a psychologist. A diagnostic evaluation will include a complete history of symptoms such as: when they started, how long they have lasted, how severe they are, whether the patient had them before and, if so, whether the symptoms were treated and what treatment was given. In individuals affected by Fabry disease, studies have shown that treatment with enzyme replacement therapy (ERT) does improve emotional as well as physical health.
accurately reflect the actual circumstances. Negative thinking fades as treatment begins to take effect. The Emory LSDC has several psychologists and psychiatrists skilled in treating Fabry related depression, anxiety, and panic attacks. Please call us at 404-778-8565 or 800-200-1524 if you would like a referral for any of these conditions. We want to help you get the treatment you need.

**Where can I find out more information on Fabry disease, depression, and anxiety?**

*The National Institutes of Mental Health website on Depression, Anxiety, and Panic Disorders/Attacks*


[http://www.nimh.nih.gov/HealthInformation/panicmenu.cfm](http://www.nimh.nih.gov/HealthInformation/panicmenu.cfm)

*Fabry Support & Information Group (FSIG)*

108 NE 2nd Street, Suite C, P.O. Box 510, Concordia, MO 64020

Phone: (660) 463-1355

Internet: [http://www.fabry.org/](http://www.fabry.org/)

*The Emory Lysosomal Storage Disease Center*

2165 North Decatur Road, Decatur, GA 30033

Phone: 404-778-8565 or 800-200-1524

Internet: [http://www.genetics.emory.edu/genservices/lsdc.php](http://www.genetics.emory.edu/genservices/lsdc.php)

**References:**


