



Fabry Disease: A Guide for the Newly Diagnosed^{©2006}

1. *I've just been diagnosed with Fabry Disease! Where do I go for more information on the disease, treatment, and medical care for Fabry Disease?*

The best way to obtain the most accurate, current, clear, and comprehensive information is to be seen at a lysosomal storage disease center (LSDC). LSDCs are genetic centers that specialize in the treatment of patients with lysosomal conditions such as Fabry disease. At most centers you will see a medical geneticist, genetic counselor, and nurse who work as a team to answer your questions, discuss testing, identify your at-risk family members, and develop a comprehensive evaluation and treatment plan for you. The LSDC will work with your current doctors to organize the treatment, tests, and specialists you need. There is at least one lysosomal storage disease center in every region. A partial list of treatment centers can be found online at the Fabry Support and Information Group's website at <http://www.fabry.org/FSIG.nsf/Pages/Links> and/or the Emory LSDC's website at http://www.genetics.emory.edu/LSDC/lscd_websites.php. Please feel free to call the Emory Lysosomal Storage Disease Center at 800-200-1524 to locate a center in your state.

2. *What causes Fabry Disease?*

Fabry Disease is an inherited condition caused by an absence or dysfunction of a specific chemical known as α -galactosidase A. This chemical is an enzyme, which is a protein that promotes a specific chemical reaction. α -galactosidase A is normally produced by the body and helps break down a substance called globotriacylceramide, also known as GL-3. When a person lacks α -galactosidase A, GL-3 builds up in the tissues of the body.

3. *What are some common symptoms of Fabry Disease?*

The build-up of GL-3 in tissues and blood vessels over time causes problems with the skin, kidneys, stomach, intestines, heart, brain, and nerves. Although symptoms can vary from one person to another, the most common symptoms are described below.

Common symptoms in childhood include: a purplish-pink skin rash, decreased sweating, fatigue, diarrhea, headaches, frequent overheating, protein in the urine, and burning or tingling pain in their hands or feet. Children can also have episodes of severe pain which are usually triggered by illness, overheating, or stressful situations. (see the "Fabry Disease in Children" fact sheet for more information)

As people with Fabry disease move into the teenage and adult years without treatment, the GL-3 continues to build up, causing increased health problems including: hearing loss, leaky heart valves, enlargement of the lower heart chambers, depression, heart attacks, kidney disease, and strokes.

Fabry disease symptoms affect both men and women. Not every person with Fabry disease will have all of the same symptoms of disease progression, but without treatment, the disease always gets worse over time.

4. *Is there a treatment for Fabry disease?*

Currently there is one Federal Food and Drug Administration (FDA) approved treatment for Fabry disease in the United States called enzyme replacement therapy (ERT). The goal of ERT is to replace the enzyme, α -galactosidase A, missing or not working in patients with Fabry so that their body can breakdown GL-3 and remove it from the tissues. Patients getting ERT usually visit an infusion center every other week and receive the enzyme by an intravenous infusion. When began early in the course of the disease, replacing the enzyme helps slow the progression of the disease, reduces complications, and may even prevent long term complications. (For more information on ERT see "Enzyme Replacement Therapy for Fabry Disease" booklet)

5. *Even though I've been diagnosed with Fabry Disease, I don't feel sick, do I need to be evaluated?*

YES! Fabry disease is a progressive, life-threatening condition that can get worse quickly. Several symptoms of Fabry disease may not cause you to feel bad until they are severe.

6. What other kind of specialists will I need to see?

Patients affected by Fabry Disease should visit their primary care physician and LSDC at least once a year for a thorough evaluation, physical exam, and laboratory tests. These tests help monitor disease progression and overall health. Many individuals affected by Fabry disease will also have a nephrologist to monitor kidney functions, a cardiologist to monitor their heart, a neurologist to monitor their brain function and pain, and a psychologist to monitor depression or anxiety. Other specialists may be needed to address gastrointestinal issues or other Fabry related symptoms.

7. What tests are used to monitor Fabry disease?

Although different lysosomal storage disease centers may vary slightly in their test and time schedule, current recommended studies to evaluate Fabry related symptoms include yearly:

- CBC, platelet count, serum creatinine + BUN, GL3, thyroid studies, and a basic metabolic chemistry panel
- Routine Urinalysis
- 24 hour Urine with creatinine, glomerular filtration rate, and protein clearance
- Random urine measuring total protein and creatinine levels.
- EKG
- Echocardiogram
- Brain MRI or Head CT
- Hearing test
- Eye exams
- Pulmonary function tests
- Depression/Anxiety assessment

8. How can I determine who else in my family has Fabry disease?

When you visit an LSDC or genetic center, the genetic counselor or geneticist will take a detailed family history of your immediate and extended family for several generations. This information will help identify anybody who might be at risk for Fabry Disease so he or she can get testing and treatment.

The identification of at risk family members is determined by looking at the way in which people are related. Fabry follows an X-linked pattern of inheritance, meaning that the non-working gene that causes Fabry disease is located on the X-chromosome. Women have two X chromosomes and men only have one X chromosome. If a female has Fabry Disease, there is a 50% chance that her daughters will have Fabry disease and a 50% chance her sons will have Fabry disease. If a male has Fabry disease, all of his daughters and none of his sons will be affected.

Testing of Fabry disease is done through a simple blood test or tests. Since men and women have different numbers of X chromosomes, the type of testing required for diagnosis of Fabry disease is different in men and women. Men can be diagnosed with Fabry disease by measuring their level of functioning enzyme alone. Women need to have enzyme and DNA analysis to find out if they have Fabry Disease.

9. Where do I go to participate in Fabry disease research?

Research on Fabry disease is performed at almost every LSDC across the world. Large national studies may have picked a few sites across the country to run their tests. You can learn more about current Fabry studies by searching the NIH's listing of research at www.clinicaltrials.gov, talking with your LSDC, or joining the discussion at FSIG www.fabry.org.

10. Where can I find more information about Fabry Disease?

There are many resources that can provide more information and support for patients with Fabry Disease.

- The Fabry Support and Information: <http://www.fabry.org/>
- The National Fabry Disease Foundation: <http://www.thenfdf.org/>
- The Fabry Community: <http://www.fabrycommunity.com/>
- The Emory LSDC can be reached at 800-200-1524 or 404-778-8565 during work hours, but the website has information and several fact sheets about Fabry Disease under the Lysosomal Storage Disease Heading: <http://www.genetics.emory.edu/LSDC/lscd>