**What is Fabry disease?** Fabry disease is a genetic lysosomal storage disorder that affects about 1 in 40,000 men and an unknown number of their female family members. Signs of Fabry disease include:

- severe pain in the hands and feet
- the inability to sweat
- fatigue
- a pinkish-purple skin rash typically in the bathing trunk area
- frequent diarrhea and constipation
- abdominal pain
- kidney failure
- heart problems
- hearing loss
- strokes,
- early death.

The condition is caused by lack of an enzyme called α-galactosidase which breaks down a fatty substance in the body called globotriaosylceramide or GL3. In families affected by Fabry disease, men who inherit the Fabry gene always develop related health problems, while not all women with the gene have symptoms. The difference in expression between men and women is related to the way in which the condition is inherited. Fabry follows an X-linked pattern of inheritance, meaning that the gene for Fabry disease is located on the X-chromosome. Men, who have only one X chromosome, are unable to produce the enzyme if they inherit the Fabry gene. Women, on the other hand, have two X chromosomes. If they inherit one Fabry gene (or are a heterozygote), they have a second gene on their other X chromosome which is able to produce enzyme. Heterozygotes’ α-galactosidase levels may vary from normal to very low. In the past, it was believed that women who were heterozygotes were carriers and would not have Fabry related health problems because they had a normal second copy of the gene. However, women can and do have Fabry related health problems. In some cases, woman can have health problems to the same degree as their male counterparts. Since women who carry one copy of a nonworking gene (also known as “heterozygotes”) do have symptoms of Fabry disease, it is important that they discuss Fabry disease with their doctor and obtain appropriate referrals to monitor their health.

**How do I determine if I have Fabry disease?** A diagnosis of Fabry disease in a woman has three parts. The first part is a genetics appointment and evaluation, the second part is a simple blood test that measures the levels of the α-galactosidase enzyme levels in your blood, the third part is a genetic test that determines if you carry the nonworking gene on one of your X chromosomes this test is run on the same blood drawn for the enzyme test.

**As a woman, can I have health problems related to Fabry disease?** Yes, women who are carriers for Fabry disease may experience any of the above listed symptoms of Fabry disease.
Are there certain tests suggested to check my health? Yes, it is important that you have heart and kidney studies to monitor these organs every year. Other studies, such as brain MRIs and hearing tests are also recommended, but on a less frequent basis. The Lysosomal Storage Disease Center at Emory University or a center near you that specializes in Fabry disease can monitor you and other family members and make recommendations to your primary care physician.

What is the treatment for women with Fabry disease? The best treatment for a woman affected by Fabry disease depends on her individual health problems and needs. Enzyme replacement therapy is available to help reduce the amount of fatty GL3 build-up in your body’s tissues and prevent irreversible damage to organs. Dilantin or Tegretol is often prescribed for Fabry related pains. Reglan is often used to help alleviate the stomach and intestinal issues associated with Fabry disease. At different points in a woman’s life, she may choose different treatments.

Will it hurt my baby if I become pregnant during treatment for Fabry disease? The answer to this question depends on the treatment a patient is undergoing. BEFORE becoming pregnant any woman wishing to consider pregnancy should discuss their medications with their doctor. No human studies have been done to establish the safety and efficacy of enzyme replacement therapy for Fabry disease during pregnancy. This drug should be used during pregnancy only if clearly needed. If a woman who is taking Dilantin or Tegretol to control her hand and foot pain wishes to become pregnant, she should discuss her medications with her doctor before becoming pregnant. Taking Dilantin and Tegretol during pregnancy increases the risk for birth defects and developmental delays in the baby. If you become pregnant unexpectedly, call your doctor as soon as you learn of your pregnancy to discuss medication use.

Who else in my family could be affected by Fabry disease? Fabry disease is an X-linked inherited disorder. Men with Fabry disease pass the nonworking gene on to all their daughters and none of their sons. Heterozygote females, whether symptomatic or not, have a 50% chance of passing the nonworking gene to either their sons or daughters. It is very important to discuss Fabry disease with any family member who could be affected by Fabry disease. If you would like a copy of a letter explaining Fabry disease to your family, and/or testing information please feel free to contact the Emory Lysosomal Storage Disease Center at (404) 778-8518 or (800) 200-1524.

When should I test my children to see if they have Fabry disease? Since the FDA’s approval of the enzyme replacement therapy treatment, many families are now choosing to test their children at an earlier age. Early diagnosis means early intervention. A team member of the Lysosomal Storage Disease Center at Emory University can discuss the decision to test your children with you further.

Where can I call to learn more about Fabry disease? A Lysosomal Storage Disease Center near you can be a great resource. The Emory Lysosomal Storage Disease Center can be reached at (404) 778-8518 or (800) 200-1534. Genzyme Medical Information (800-200-1534) can provide accurate information on enzyme replacement therapy for Fabry disease. Websites for additional information:

- [www.Fabry.org](http://www.Fabry.org) (Fabry Support and Information Group or FSIG)
- [www.Fabrycommunity.com](http://www.Fabrycommunity.com) (Information on Fabry disease by Genzyme)
- [www.Rarediseases.org](http://www.Rarediseases.org) (National Organization of Rare Diseases or NORD)