Things to consider BEFORE having genetic testing for Fabry Disease

What is Fabry disease?
Fabry disease is a medical condition that runs in families and affects children and adults. People with Fabry Disease have a deficiency of an enzyme in their body called α-galactosidase A. This enzyme usually helps break down a fatty substance called globotriacylceramide or GL-3, that is present in all tissues of the body, including the liver, kidney, skin, and blood vessels. When a person is missing α-galactosidase A, GL-3 builds up in these tissues causing the health problems of Fabry disease. There is a classic, more severe form of Fabry disease that usually begin in childhood and many “non-classic” forms of Fabry disease that may start during a person’s teen or adult years. The early symptoms of Fabry disease, which often begin in childhood, include decreased sweating, heat intolerance, protein in the urine, 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 stroke and vertigo. 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. Early treatment for Fabry disease decreases the risk for severe Fabry related health problems. Accordingly, knowing that you have Fabry disease is important. However, before you decide to have testing for Fabry disease it is a good idea to think through the implications of testing.

What if the test is positive?
Some people have genetic testing done expecting to not have Fabry disease; however, you must also be prepared to receive the news that you (and/or your family members) are affected by Fabry disease.

Before testing, think about how you would feel if the test tells you that you are affected by Fabry disease. Think about what these means for your other family members. Think about the process of getting evaluated or treated for Fabry disease. Feel free to talk to your physician or genetic counselor to discuss implication on yourself and your family before testing. Take your time when deciding whether or not to be tested for Fabry disease.

Information about your family
When going through the testing process, you may discover things you did not know about your family. For example, you may learn that your children are at risk to be affected by Fabry disease. This may bring on strong emotional times for you and your family. A parent may feel guilty for having “passed Fabry disease” on to their children, even if they know that they have no control over which gene is inherited. On the other hand, even though they do not want the health problems of Fabry disease, an individual found to be unaffected by Fabry disease may feel guilty that they aren’t affected or isolated on the outskirts of the family as the affected family members share treatments, support, and physicians. In addition to these considerations, in rare cases genetic testing for Fabry disease may reveal that there are individuals in your family who are not biologically related to one or more of their parents.
Insurance
Most health insurance companies will cover most costs related to genetic testing for Fabry disease. Your doctor or genetic counselor might need to write a letter explaining why testing is needed. If you are being testing for Fabry disease through the Fabry Family Testing Project, your testing will be free of charge and your insurance will not be billed for the test.

Many people are worried that the results of the genetic test will affect the chances of getting health insurance. The federal law known as Genetic Information Nondiscrimination Act (GINA) prohibits health insurers and employers from using your genetic information, which includes the following.

1. Employers cannot deny you a job because of the results of genetic testing
2. Health insurers cannot use genetic testing to deny your coverage or set your insurance rates
3. Employers and insurers cannot require you to have genetic testing.

In the United States, no federal legislation directly addresses the issue of genetic testing and life insurance. Although a few states have enacted statewide laws restricting the use of genetic information in life, disability and long-term care insurance, GINA does not cover these types of insurance and it is recommended that you explore life insurance options before genetic testing.

Many people are worried that the results of the genetic test will affect the chances of getting health insurance. The federal law known as Genetic Information Nondiscrimination Act (GINA) prohibits health insurers and employers from using your genetic information, which includes the following.

1. Employers cannot deny you a job because of the results of genetic testing
2. Health insurers cannot use genetic testing to deny your coverage or set your insurance rates
3. Employers and insurers cannot require you to have genetic testing.

In the United States, no federal legislation directly addresses the issue of genetic testing and life insurance. Although a few states have enacted statewide laws restricting the use of genetic information in life, disability and long-term care insurance, GINA does not cover these types of insurance and it is recommended that you explore life insurance options before genetic testing.

State by state information on genetic testing and life, disability, and long term care insurance can be found at the National Conference of State Legislatures website located at http://www.ncsl.org/default.aspx?tabid=14283

Confidentially
Some may feel concerned about maintaining the privacy of their genetic information. Your doctor isn’t allowed to tell anyone that you have had a gene test or the results without your consent. Consider discussing privacy concerns with a genetic counselor or doctor.

Deciding not to get tested
If your family history suggests that you are at risk to be affected by Fabry disease, but you decide not to have a genetic test, you can pursue all the services (kidney testing, heart evaluations, brain imaging, etc) available to people with Fabry disease. You don't need to have a genetic test in order to have screening. However, without a diagnosis, you will be unable to accurately determine the risk of passing on Fabry disease to your children, you will be unable to obtain treatment with the FDA approved medication, and it may be difficult to having monitoring tests covered by your insurance.

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 or at <http://genetics.emory.edu/lsdc>. Websites that may be of help are: www.nfdf.org (The National Fabry Disease Foundation), www.Fabry.org (Fabry Support and Information Group or FSIG), www.ThinkGenetic.com (interactive information on genetic conditions including Fabry disease), and www.Fabrycommunity.com (Information on Fabry disease by Genzyme).