



EMORY
UNIVERSITY

Emory Genetics Laboratory

Dear Friend,

The American Association of Kidney Patients is the oldest and largest, fully independent kidney patient organization in the U.S. focusing on the various causes and complications of kidney disease. AAKP is dedicated to providing education and support to individuals with kidney disease and their care partners, helping them understand their condition, manage their care and achieve their best outcomes possible.

This is why AAKP, since 2007, has partnered with Emory University Medical Center to provide complimentary genetic testing for individuals at-risk and families affected by Fabry Disease. AAKP and Emory are able to provide this free service and Fabry disease education thanks to contributions from Sanofi Genzyme* and Amicus* – companies known for their expertise and commitment in the area of rare genetic diseases.

We want you to know that your genetic testing kit, postage and educational materials are being provided to you at no cost. AAKP and Emory understand the concerns that come with a genetic disease....*Do I have this disease?, Will I pass it along to my children?, How do I discuss this with my family?* AAKP and Emory are working hard together to answer these questions and help guide you and your family through this process.

Early diagnosis and treatment of Fabry Disease is important to help manage this condition which, left untreated, can damage the kidneys and other organs. Should your Fabry Disease begin to affect your kidneys, causing them to not to work properly, AAKP has a number of educational resources and support opportunities such as our website, brochures and webinars to answer your questions and help you manage your condition. We also offer FREE membership to patients and family members to keep them informed of the latest education, support and advancements in the kidney community. Join today at www.aakp.org/join or by calling us at (800) 749-2257.

AAKP and Emory are working together to better understand Fabry Disease so, as a community, we can provide as much information as possible regarding this genetic disease which will hopefully lead to advancements in the diagnosis, treatment and management of this condition.

Sincerely,

A handwritten signature in cursive script that reads "Diana Clynes".

Diana Clynes, Executive Director

**No personal information is ever shared with Sanofi Genzyme or Amicus.*

A: 14440 Bruce B. Downs Blvd.
Tampa, Florida 33613

T: 800-749-2257

F: 813-636-8122

W: www.aakp.org

The Independent Voice of Kidney Patients Since 1969™

EGL GENETICS TEST REQUISITION FORM - Page 1 of 1

2460 Mountain Industrial Boulevard, Tucker, GA. 30084 • (470) 378-2200 or 1-855-831-7447 • FAX (470) 378-2250 • eglgenetics.com

PATIENT INFORMATION:

Last Name _____ First _____ MI _____

Parents Names (if pt is a minor) _____

DOB _____ / _____ / _____ Gender: Male Female Unknown

Mailing Address _____

City _____ State _____ Zip _____

Preferred Phone# (_____) _____ - _____

Is this #: Home Work Cell Other: _____

Other # where pt. can be reached: (_____) _____ - _____

Ethnicity of Pt. (circle all that apply):
African-American Asian
Caucasian/NW European East Indian Hispanic
Jewish-Ashkenazi Jewish-Sephardic Mediterranean
Native American Native Hawaiian/Other Pacific Islander
Other

PHYSICIAN ORDERING TEST:

Name _____

Practice Name _____

Address _____

City _____ State _____ Zip _____

Preferred Phone# (_____) _____ - _____

FAX# (_____) _____

Genetic Counselor _____ Phone _____

DUPLICATE REPORTS TO:

Name: Dawn Laney, MS, CGC and Allison Foley, MS, CGC

Address 101 Woodruff Circle, 7th floor, Suite 7130

City Atlanta State GA Zip 30322

Preferred Phone#: (404) 778 - 8518

FAX#: (404) 778-8562

Email (preferred method): fabry.testing@emory.edu

SPECIMEN INFORMATION:

Date Collected _____ / _____ / _____ Time _____ : _____ AM/PM

SAMPLE TYPE:

_____ Blood(B)
_____ Saliva Sample
_____ Urine
_____ Other _____

REASONS FOR REFERRAL: (ICD-10; check all that apply)

Family history of Fabry Disease (272.7)

PREVIOUS LAB RESULTS:

See attached report with identified family mutation

SPECIAL INSTRUCTIONS:

Fabry Family Member Testing project (AAKP)

Paperwork to be reviewed by genetic counselor

Please mark this box if you do **NOT** want to be contacted about future Fabry related research studies.

BILLING INFORMATION:

Fabry Family Member Testing project, bill to
The Emory Clinic – Dawn Laney & Allison Foley
(Client number 39922)

Testing Options

- Fabry Family Member Testing (KM): Targeted mutation testing for previously identified family mutations. (A report of the family mutation **must** accompany submitted sample)
- Fabry Familial Mutation Testing (DKMDD): Targeted Deletions/Duplications. (A report of the family mutation **must** accompany submitted sample)
- Fabry Disease: *GLA* Gene Sequencing (DG)
- Fabry Disease: *GLA* Gene Deletion/Duplication (KX)
- Fabry Disease: Globotriaosylceramide (Gb3) Quantification, Urine (BGBCU)
- Fabry Disease: Alpha-Galactosidase Enzyme Activity, Leukocytes (LB)

NOTES:

Rec/d _____ / _____ /0__ Unboxed By _____ Sender: _____

TEMP	SPECIMEN	COLOR	#TUBES	VOL	COMPLETE	INC
R C F					PT. DATA	
R C F					TEST DATA	
R C F					LABELS	

THIS BOX FOR LAB USE ONLY

FABRY FAMILY MEMBER REQUISITION FORM

NAME/DOB OF PERSON BEING TESTED: _____

I. History of Individual Being Tested for Fabry Disease: (check all that apply)

- | | |
|--|---|
| <input type="checkbox"/> Pain in hands/feet | <input type="checkbox"/> Tinnitus or Hearing Loss |
| <input type="checkbox"/> Numbness/tingling | <input type="checkbox"/> Proteinuria |
| <input type="checkbox"/> Chronic fatigue | <input type="checkbox"/> Kidney Failure |
| <input type="checkbox"/> Purplish-red rash (Angiokeratoma) | <input type="checkbox"/> Cardiac Problems |
| <input type="checkbox"/> Decreased sweating | <input type="checkbox"/> TIAs/-strokes |
| <input type="checkbox"/> Heat and/or Cold Intolerance | <input type="checkbox"/> Depression/Anxiety |
| <input type="checkbox"/> Gastrointestinal Problems | <input type="checkbox"/> Corneal Changes (Whorls) |
| <input type="checkbox"/> Other _____ | |

II. Previous Testing:

Have YOU had any previous testing for Fabry disease?

- No prior testing/Unknown
- Enzyme analysis:
Date of testing: _____
Results: _____
Laboratory: _____
- Molecular/DNA analysis:
Date of testing: _____
Results: _____

III. Patient's Family History:

Which of your family members are affected by Fabry disease? (check all that apply)

- | | | |
|----------------------------------|---|--|
| <input type="checkbox"/> Mother | <input type="checkbox"/> Maternal Grandmother | <input type="checkbox"/> Paternal Aunt |
| <input type="checkbox"/> Father | <input type="checkbox"/> Maternal Grandfather | <input type="checkbox"/> Paternal Uncle |
| <input type="checkbox"/> Brother | <input type="checkbox"/> Maternal Aunt | <input type="checkbox"/> Maternal cousin |
| <input type="checkbox"/> Sister | <input type="checkbox"/> Maternal Uncle | <input type="checkbox"/> Paternal cousin |
| <input type="checkbox"/> Niece | <input type="checkbox"/> Paternal Grandmother | <input type="checkbox"/> Others _____ |
| <input type="checkbox"/> Nephew | <input type="checkbox"/> Paternal Grandfather | |

What is the name and relationship of the relative whose molecular test results including mutation are being sent with your sample?

Name: _____ Family Relationship: _____

Where and when was their molecular testing performed?

Date of testing: _____ Mutation: _____
Laboratory who performed the testing: _____

Informed Consent – MOLECULAR GENETIC TESTING

Instructions: Please obtain patient signature on consent form and provide a signed copy to EGL Genetics to permit testing and processing. If a signed consent is not submitted, EGL Genetics assumes that the ordering clinician has discussed testing with the patient and obtained the patient’s informed consent.

I, (name) _____ voluntarily request of EGL Genetics to perform DNA-based testing for (condition) _____ in myself/my child (child’s name _____),

in an attempt to determine whether I/my child (“patient”) am a carrier of a disease gene or are at increased risk to be affected by a genetic condition. The following points were explained and I understand that:

- DNA testing requires a blood sample, cheek or mouth swab, muscle or skin biopsy, all of which have risks associated with obtaining the sample. Additional samples may be needed if the sample is damaged in shipment or inaccurately submitted. In order to perform accurate prenatal testing, samples from the affected individual, parents, or additional family members may be required.
- DNA-based studies performed are specific to the condition indicated above. The accuracy of genetic testing is limited by the methods employed, the clinical diagnosis, and the nature of the specific condition for which testing is requested. In some cases, the test will detect an abnormality, called a pathogenic variant, in the gene. In other cases the test is unable to identify an abnormality although an abnormality may still exist. This event may be due to the current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of changes (pathogenic variant) in a gene. These tests are currently available for clinical laboratory testing; however, improvements will be made as scientific knowledge advances. As with any complex genetic test, there is always a small possibility of a failure or error in sample analysis. Extensive measures are taken to try to avoid these errors. The methods are not 100% accurate due to the possibility of rare genetic variations in the DNA of an individual or due to the complexity of the testing itself. A low error rate, approximately 1 in 1000 samples, is generally estimated to exist in a laboratory.
- It is the responsibility of the referring physician or health care provider to understand the specific use and limitations of the testing ordered, and to educate the patient regarding these limitations. Additional information describing indications, methodology and detection can be found on the EGL website at: <https://www.egl-eurofins.com/>
- Accurate interpretation of test results is dependent upon the patient’s clinical diagnosis or family medical history and upon reported family relationships being true biological relationships. An erroneous clinical diagnosis in the patient or family member can lead to an incorrect interpretation in the laboratory result. Genetic testing in family members can sometimes reveal that true biological relationships are not consistent with the reported biological relationships. For example, non-paternity may be detected, which means that the stated or assumed father of an individual is not the true biological father.
- Due to the complexity of DNA testing and potential implications of test results, results will be reported directly to the patient’s ordering provider, who will then review and discuss the test results with me. Patient-identifying results and information at EGL will remain confidential and may only be released to other parties with my expressed written consent or as permitted or required by applicable law.
- EGL Genetics is not a DNA banking facility and does not guarantee the future availability of isolated DNA. Any requests for additional studies must be ordered by the referring provider and charges will be incurred. Once the test is complete, identifying information may be removed and remaining DNA samples may be used for de-identified laboratory purposes. These samples will not be available for future clinical studies. Any results obtained cannot be related back to the original source, so no results can be reported.
- I can request that remaining DNA not be used for research purposes by initialing here: _____

The risks, benefits and limitations of DNA testing have been explained to me. I have read and will receive a copy of this consent form.

Patient Signature	Date	Parent / Guardian Signature	Date
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Physician/Counselor/Clinician Statement: I have explained DNA testing to the patient/parent/guardian. The consent form and limitations of genetic testing were reviewed with the patient/parent/guardian. I accept responsibility for pre- and post-test genetic counseling. I will use my independent professional judgment and the patient’s best interests in advising the patient/parent/guardian regarding DNA test results, the use and limitations of same, and any research study, clinical trial, drug, treatment or device brought to my attention by EGL or others.

Clinician Signature	Date
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Fabry Testing Checklist

The Fabry Family Member Testing project, sponsored by the American Association of Kidney Patients (AAKP), is a special program intended to test individuals with signs and symptoms of Fabry disease and/or with a known family history of Fabry Disease.

In order to be tested for Fabry Disease through this program, you must have some clinical signs or symptoms of Fabry disease OR you must be at risk to be affected by Fabry Disease based on the Fabry Disease pattern of inheritance in your family and know the genetic change or mutation causing Fabry Disease in your family. ***If a review of your symptoms does not meet criteria for testing, we will notify you that we cannot test your sample for Fabry disease, or if a review of your family history finds that you are not at an increased risk to be affected by Fabry Disease based on family history or you are unable to provide a diagnostic report detailing the mutation causing Fabry Disease in your family, we will notify you with the information that we cannot test your saliva sample for Fabry Disease.***

If you do not qualify for testing under the Fabry Family Member Testing project, clinical testing for a fee is also available at several diagnostic labs including EGL Genetics.

Please follow the following steps to make sure your test is not delayed:

____ Read the “**Things to consider BEFORE having genetic testing for Fabry Disease**” factsheet and complete consent form. Talk to your doctor, genetic counselor, or an Emory Fabry genetic counselor if you have questions about any information on the sheet or form. Emory Fabry genetic counselors can be emailing Allison Foley alfoley@emory.edu or Dawn Laney at dawn.laney@emory.edu.

____ If you are having testing because you have a family member with Fabry disease, send a copy of your affected family member’s molecular report that includes the family member’s mutation(s) with your saliva sample. If a family has already been tested at Emory, please write that information on the requisition form in the “notes” section and email Allison Foley alfoley@emory.edu or Dawn Laney at dawn.laney@emory.edu with that information. ****Please note, testing cannot be performed without this report or information****

____ Before sending in the sample to the lab, please make certain that it includes the saliva sample with tested person’s name on it, consent form, completed requisition forms including patient information, **physician information**, and specimen collection sections on the EGL test requisition form. ****please make sure that the tested person’s NAME is written on the saliva tube****

IMPORTANT INFORMATION:

Results will be faxed to the doctor and genetic counselor written on the requisition form when they are completed. Patients are able to call and receive results after they are faxed to their doctor from a clinical genetic counselor, but results will only be sent automatically to the doctor.

Questions?

Call the Emory Fabry Center at (404) 778-8518/(800) 200-1524 or email Allison Foley alfoley@emory.edu or Dawn Laney at dawn.laney@emory.edu.

Things to consider before having genetic testing for Fabry Disease

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 lacks α -galactosidase A, GL-3 builds up in these tissues causing the symptoms of Fabry disease. The early symptoms of Fabry disease, which usually 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.

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/lcdc>. 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.Fabrycommunity.com (Information on Fabry disease by Genzyme), and www.Rarediseases.org (National Organization of Rare Diseases or NORD).