Dear Friend,

The American Association of Kidney Patients is the oldest and the only patient-centered nonprofit 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 and EGL Genetics (formerly Emory Genetics Laboratory) 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 an educational research grant funded by Sanofi Genzyme* – a company known for its 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.

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,

Gary Green, Executive Director

*No personal information is ever shared with Sanofi Genzyme.
**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[ ] 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:**

This box for lab use only

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FABRY FAMILY MEMBER REQUISITION FORM

NAME/DOB OF PERSON BEING TESTED: __________________________

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

☐ Pain in hands/feet         ☐ Tinnitus or Hearing Loss
☐ Numbness/tingling          ☐ Proteinuria
☐ Chronic fatigue           ☐ Kidney Failure
☐ Purplish-red rash (Angiokeratoma) ☐ Cardiac Problems
☐ Decreased sweating        ☐ TIAs/Strokes
☐ Heat and/or Cold Intolerance ☐ Depression/Anxiety
☐ Gastrointestinal Problems ☐ Corneal Changes (Whorls)
☐ 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)

☐ Mother                  ☐ Maternal Grandmother ☐ Paternal Aunt
☐ Father                  ☐ Maternal Grandfather ☐ Paternal Uncle
☐ Brother                 ☐ Maternal Aunt       ☐ Maternal cousin
☐ Sister                  ☐ Maternal Uncle     ☐ Paternal cousin
☐ Niece                   ☐ Paternal Grandmother ☐ Others __________
☐ Nephew                  ☐ 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.

<table>
<thead>
<tr>
<th>Patient Signature</th>
<th>Date</th>
<th>Parent / Guardian Signature</th>
<th>Date</th>
</tr>
</thead>
</table>

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.

<table>
<thead>
<tr>
<th>Clinician Signature</th>
<th>Date</th>
</tr>
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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 emailed at alfoley@emory.edu or 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. *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.