Fabry Family Member 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 a known family history of Fabry Disease. In order to be tested for Fabry Disease through this program, 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 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 Emory Genetics Laboratory.

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 by emailing Allison Foley alfoley@emory.edu or Dawn Laney at dawn.laney@emory.edu.

_____ 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, two 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.