

INFORMED CONSENT FOR FABRY DISEASE TESTING

PLEASE RETURN A COPY OF THIS SIGNED CONSENT FORM WITH YOUR BLOOD SAMPLE

I authorize Genetic Disease Diagnostic Laboratory of the Mount Sinai School of Medicine, New York to analyze a sample of my (my child's/my ward's) blood for the purpose of determining if I (my child/my ward) am (is) a male affected with Fabry disease or a female who is heterozygous for Fabry disease. The Genetic Disease Diagnostic Laboratory of the Mount Sinai School of Medicine is New York State/CLIA approved for these tests

I understand that:

- The purpose of this testing is to determine if I (my child/my ward) am (is) a male affected with Fabry disease or a female who is heterozygous for Fabry disease.
- Measurement of α -galactosidase A activity is accurate for diagnosis of males; however it is not reliable for diagnosis of females and mutation analysis may be required.
- In diagnostic testing, rare errors can occur, for example, due to sample mix-up, and/or laboratory errors. Genetic polymorphisms may also cause rare errors in mutation analysis.
- A written report of the results will be sent to me and/or my physician, as requested.
- Counseling will be available to me about the results of the tests and their effect on my family by a genetic counselor
- The results of this diagnostic testing and any medical information that I provide are confidential and will not be used for other purposes unless I give MY signed consent.
- I give the Mount Sinai Genetic Testing Laboratory permission to store my DNA labeled with my name indefinitely.

Please initial:

☐

Yes

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No

Signature: _____

Date: _____

Printed Name: _____

Witness: _____

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If you have any questions, please contact Dana O. Doheny, MS, CGC, Genetic Counselor, by telephone (212-659-6779, direct, or 866-322-7963, toll-free) or email (dana.doheny@mssm.edu) to discuss our Fabry disease testing and sample requirements. **THANK YOU.**