Cincinnati Children's

Division of Human Genetics

Fabry Disease

Gene tested: *GLA*

Molecular Genetics Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
Fax: (513) 636-4373
Email: moleculargenetics@cchmc.org

Additional information and test requisitions are available at:
www.cincinnatichildrens.org/molecular-genetics

Helping you fit the pieces together

Shipping Instructions

Please enclose test requisition with sample. All information must be completed before sample can be processed.

- All samples must be clearly labeled with the patient's name and birth date and initialed by the phlebotomist.
- All samples should be wrapped, cushioned against breakage and sealed in a plastic bag before placement in the shipping container.
- All samples should be sent at room temperature. Do not allow to freeze.
- Samples should be sent by overnight carrier to arrive Monday through Friday. Do not ship samples on Friday.

Ship to:
Molecular Genetics Laboratory
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229

Also known as:

- Alpha-Galactosidase A Deficiency (GLA Deficiency)
- Anderson-Fabry Disease

Fabry disease is a lysosomal storage disease caused by decreased activity of alpha-galactosidase A. Fabry disease is inherited as an X-linked dominant disorder and affects about 1 in 40,000 males in all ethnic groups. Heterozygous (carrier) females may be symptomatic although the incidents of Fabry disease in females is not well ascertained.

Males typically present in childhood with hand pain, temperature intolerance, and unexplained high fevers. Around puberty, males may develop flat or slightly raised vascular lesions, abdominal pain, nausea, diarrhea, and absence of sweating or diminished sweating. Life threatening complications of kidney failure, heart disease, and strokes may present as early as the third to fourth decade. Without any medical intervention, males with Fabry disease typically die in their mid forties. Modern medical interventions, such as kidney transplant and enzyme replacement therapy, may extend the lives of affected males. Heterozygous females may be completely asymptomatic or as severely affected as males. The majority of heterozygous women have a combination of signs and symptoms that present later than those in affected males. However, women remain at increased risk for kidney failure, heart disease, and early strokes.

In males, the diagnosis of Fabry disease is confirmed by measuring decreased GLA enzyme activity in plasma, leukocytes or cultured skin fibroblasts. Enzyme testing is unreliable in females. Molecular determination of the specific *GLA* mutation is recommended in all patients and is essential for accurate carrier testing in at-risk females.

INDICATIONS:

- Confirmation of diagnosis in a symptomatic individual
- Presymptomatic testing of at-risk relatives
- Carrier identification in individuals with a family history of Fabry disease
- Prenatal diagnosis of an at-risk fetus, after confirmation of mutation in the mother or sibling (by prior arrangement only)

SPECIMEN:

Blood samples should be collected in EDTA (lavender topped) tubes. A minimum of 5 mLs on adult or 3 mLs on child is required for analysis. Blood collected on Friday may be stored in refrigerator until Monday for overnight shipment.

For other tissue requirements please call 1-800-344-2462, extension 4474.

METHODOLOGY:

PCR-based sequencing of all seven exons and exon/intron boundaries of the *GLA* gene.

SENSITIVITY:

PCR-based sequencing of the *GLA* gene identifies a mutation in >99% of male patients with Fabry disease. The sensitivity of DNA sequencing is over 99% for the detection of nucleotide base changes, as well as small deletions and insertions in the regions analyzed. Multiple exon deletions and insertions may not be identified by this methodology but are rare in Fabry disease. *GLA* is the only gene associated with Fabry disease.

TURN-AROUND TIME:

Reports are routinely available within three weeks of sample receipt. Abnormal results will be called to the referring physician. All reports will be faxed to the referring physician.

COST:

Please call 1-866-450-4198 for prices or with any billing questions.

CPT CODES:

GLA Full Gene Sequencing	81405
Family Mutation Studies	81403

Updated 12/2011