

Fabry Disease: Alpha-Galactosidase Enzyme Activity, Leukocytes

Test Code: LB

Turnaround time: 7 days - 10 days

CPT Codes: 82657 x1, 84155 x1

Condition Description

Fabry disease is an X-linked condition caused by a deficiency of alpha-galactosidase A enzyme activity. Affected individuals are unable to metabolize globotriaosylceramide (GL-3) in their lysosomes. The progressive accumulation of GL-3 results in symptoms that include: characteristic lysosomal disease skin rashes (angiokeratomas), decreased sweating (hypohidrosis), chronic fatigue, depression, neuropathic pain in the hands and feet (acroparesthesia), gastrointestinal issues, strokes, cardiac disease (including left ventricular hypertrophy), and renal disease (proteinuria to end stage renal disease). The diagnosis of Fabry disease has increased in importance since treatment with enzyme replacement therapy is now available.

Fabry disease affects both men (hemizygotes) and women (heterozygotes), but testing strategy varies based on gender. Alpha galactosidase A enzyme analysis alone should detect all affected males and may also detect ~60% of heterozygote females affected by Fabry disease. In order to increase the speed and detection of females with Fabry disease, we recommend combining an enzyme analysis with sequencing of the alpha galactosidase A gene. The enzyme analysis of alpha galactosidase A can be performed on samples at the same time as gene sequencing.

For further information about Fabry disease, please call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524. For general questions, call EGL Genetics at 470-378-2200.

Visit www.ThinkGenetic.com for patient-friendly information on [Fabry disease](#).

References: Click [here](#) for Gene Reviews Clinical Summary

Indications

This test is indicated for newborns, children, adolescents, and adults who are suspected to be affected by a lysosomal storage disease such as Fabry disease.

Methodology

Fluorometric Enzyme Assay using artificial 4-MU substrate. Alpha-galactosidase is evaluated to confirm a diagnosis of Fabry disease.

Detection

Males: In affected males, alpha-galactosidase A enzyme activity activity in peripheral blood leukocytes is a reliable test. An affected individual's enzyme activity will be found to be 0-20% of normal activity.

Females: Alpha-galactosidase A enzyme activity activity in females is varies from deficient activity to normal activity, and thus will not detect all affected females.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood (Sodium Heparin)

Specimen Requirements:

Sodium Heparin (Green Top)

3-5 ml

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. Not accepted on Saturday. (Late Friday collections may be stored at room temperature over the weekend for Monday receipt.)

Related Tests

Biochemical:

- Fabry Disease: Globotriaosylceramide, Gb3 (BGBCU)
- Lysosomal Enzyme Screening (LS)

Molecular:

- Fabry Disease: *GLA* Full Gene Sequencing (DG)
- Fabry Disease: *GLA* Gene Deletion/Duplication (KX)