Alpha-Galactosidase A Enzyme Analysis

Test Code	B0007
Test Summary	This test detects alpha-galactosidase A activity.
Turn-Around-Time (TAT)*	3 days
Acceptable Sample Types	Dried Blood Spots Whole Blood (EDTA)
Acceptable Billing Types	Self (patient) Payment Institutional Billing

Indications for Testing

This test may be appropriate for individuals with a clinical suspicion of Fabry disease and/or individuals with a family history of Fabry disease.

Test Description

This test evaluates the activity of alpha-galactosidase A enzyme that has been associated with Fabry disease.

Condition Description

Fabry disease is a lysosomal storage disorder with a wide spectrum of symptoms ranging from mild cases in females, to severe cases in classically affected hemizygous males. Symptoms may include abdominal and joint pain, vascular lesions and corneal abnormalities. Neurological symptoms include autonomic dysfunction and burning sensation or numbness in the extremities. Fabry disease affects an estimated 1 in 40,000 to 1 in 60,000 males. The prevalence is unknown in females.

Test Methods and Limitations

Flow injection analysis tandem mass spectrometry (FIA/MS/MS)

Detailed Sample Requirements

Dried Blood Spots

Whole Blood (EDTA)

Collection Container(s):

EDTA (purple top)

Collection:

Infants (< 2-years): 2 to 3 mL; Children (>2-years): 3 to 5 mL; Older children and adults: Minimum 5mL. The blood tube should be inverted several times immediately after blood collection to prevent coagulation.

Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.
Shipping: Ship overnight at ambient temperature ensuring receipt within 5-days of collection.

SPECIAL INSTRUCTIONS: Clotted or hemolyzed samples are not accepted.