Alpha-Mannosidosis: Alpha-Mannosidase Enzyme Activity, Leukocytes

Test Code: LM
Turnaround time: 7 days - 10 days
CPT Codes: 82657 x1

Condition Description

Alpha-mannosidosis is an autosomal recessive disorder caused by deficiency of lysosomal alpha-mannosidase. The age of onset and symptoms of alpha-mannosidosis range from a severe perinatal/infantile form to attenuated adult onset. The presenting symptoms are often very different depending on the severity of the disease in an individual. Individuals affected by a severe form of alpha-mannosidosis, called Type 3, often die before birth, in infancy, or early childhood as a result of primary nervous system involvement and/or infections. Symptoms in affected individuals include:

- enlarged liver
- hypotonia
- frequent infections

Individuals affected by a moderate form of alpha-mannosidosis, called Type 2, develop symptoms before age 10. Symptoms of the moderate form of alpha-mannosidosis include:

- coarse facial features
- hypotonia
- skeletal abnormalities
- mental retardation
- progressive muscle weakness
- hearing loss
- ataxia
- arthritis
- corneal clouding
- and slow disease progression

Individuals affected by the attenuated form of alpha-mannosidosis, often called Type 1, have symptoms including:

- mild to moderate mental retardation
- progressive muscle weakness
- hearing loss
- retinal abnormalities
- ataxia

Individuals affected by the milder form of alpha-mannosidosis tend to exhibit symptoms after age ten and can survive into the sixth decade of life.

Please [click here](#) for the GeneReviews summary of this condition.

For further information about lysosomal storage diseases, please call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524. For general questions, please call the Emory Genetics Laboratory at (404) 778-8500.

Genes

DM

Indications

This test is indicated for:

- Newborns, children, adolescents, and adults who are suspected to be affected by a lysosomal storage disease such as alpha-mannosidosis.

Methodology

Fluorometric Enzyme Assay using artificial 4-MU substrate. Alpha-mannosidase activity is evaluated to confirm a diagnosis of alpha-mannosidosis.

Detection

In affected individuals, acid alpha-mannosidase activity in peripheral blood leukocytes is a reliable test. An affected individual's enzyme activity will be found to be 5-10% of normal activity. Acid alpha-mannosidase activity in carriers is usually 40-60% of normal, and is therefore unreliable for carrier detection given the overlap in carriers and non-carriers. Molecular testing may be more informative for carrier testing.

Specimen Requirements

Type: Whole Blood

Specimen Requirements:
5-10ml of blood in sodium heparin (green top) tube.

Specimen Collection and Shipping: Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

Related Tests

Lysosomal Enzyme Screening (LS)