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)