Fucosidosis: Alpha-Fucosidase Enzyme Activity, Leukocytes

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

Condition Description

Alpha-fucosidosis is an autosomal recessive disorder caused by deficiency of the lysosomal enzyme alpha-fucosidase. The disease is found more often in individuals from Italy and the Southwestern United States. There are two main types of fucosidosis characterized by the age of onset and type of physical and mental manifestations.

Babies affected by Type I alpha-fucosidosis begin exhibiting features of the disease in the first 3-18 months of life. Symptoms of Type I alpha-fucosidosis often include:

- mental retardation
- muscle weakness
- seizures
- coarsening of facial features
- organomegaly
- cherry red spots
- purplish-red angiokeratoma
- dysostosis multiplex

Children affected by Type II alpha-fucosidosis usually begin exhibiting symptoms of the disease in the first 12-24 months of life. Symptoms of Type II alpha-fucosidosis often include:

- mental retardation
- seizures
- coarsening of facial features
- organomegaly
- tortuosity of conjunctival vessels
- purplish-red angiokeratoma
- dysostosis multiplex resulting in growth retardation

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.

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-fucosidosis.

Methodology

Fluorometric Enzyme Assay using artificial 4-MU substrate. Alpha-fucosidase is evaluated to confirm a diagnosis of alpha fucosidosis.

Detection

In affected individuals, alpha-fucosidase 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. Alpha-fucosidase 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)