Beta-Mannosidosis: Beta-Mannosidase Enzyme Activity, Leukocytes

Condition Description

Beta-mannosidosis is an autosomal recessive disorder caused by deficiency of the lysosomal enzyme beta-mannosidase. The most common features of beta-mannosidosis include:

- mental retardation
- speech impairment
- low muscle tone
- recurrent respiratory infections
- hearing loss

Some affected individuals may also have a purplish-red rash called angiokeratomas and tortuosity of conjunctival vessels.

Please click here for OMIM clinical summary.

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 EGL Genetics at 470-378-2200.

Genes

SMPD

Indications

This test is indicated for:

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

Methodology

Flurometric enzyme assay using artificial 4-MU substrate. Beta-mannosidase activity is evaluated to confirm a diagnosis of beta-mannosidosis.

Detection

In affected individuals, acid beta-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. Beta-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

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

Lysosomal Enzyme Screening (LS)
Lysosomal Storage Disease Screen, Urine (BLSDS)
Oligosaccharide and Glycan Screening (OS)