Beta-Mannosidosis: Beta-Mannosidase Enzyme Activity, Leukocytes

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

**Condition Description**

Notice: This test will soon be discontinued.  
As of 02/01/2020, EGL can no longer accept samples for this test. For questions, please call: 470-378-2200.

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**

Fluorometric 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)