Beta-Mannosidosis: Beta-Mannosidase Enzyme Activity, Leukocytes

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

## 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.

## Related Tests

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