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**

**Type:** Whole Blood

Specimen Requirements:

5-10ml 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)
- Lysosomal Storage Disease Screen, Urine (BLSDS)
- Oligosaccharide and Glycan Screening (OS)