**Condition Description**

Lysosomal storage diseases are genetic conditions most often caused by a lack of an enzyme that eliminates unwanted substances in cells. Lysosomes are small sac-like structures that act as the “recycling center” of each cell, breaking down unwanted material into simple products for the cell to use to build new material. The lack of certain enzymes causes a buildup of the substance that the enzyme would normally eliminate, and deposits accumulate in many cells of the body. Abnormal storage causes inefficient functioning of and damage to cells, which can lead to serious health problems. Currently, there are more than 40 known lysosomal storage diseases.

There is great variability in the clinical features of these diseases. Depending on the condition, symptoms can begin anytime from before birth until late in adulthood. Some patients with lysosomal storage diseases present in childhood with developmental delay or regression of learned skills, while others present with evidence of liver and/or spleen enlargement, bone or eye abnormalities, skin lesions, or facial coarsening, with/without a neurological component. In other diseases, adolescent and adult patients will present with weakness, psychosis, and mental deterioration. Most lysosomal storage disorders are autosomal recessively inherited; however, a few are X-linked, such as Fabry Disease and Hunter Syndrome (MPS II).

For further information about lysosomal storage diseases, please call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524.

**Indications**

This test is indicated for:

- Patients whose clinical evaluations are suggestive of a lysosomal disease.

The screening panel does not test for all known lysosomal storage diseases. It does include the following diseases:

- Alpha-Mannosidosis (alpha-mannosidase)
- B-Mannosidosis (beta-mannosidase)
- Fabry Disease (alpha-galactosidase)
- Fucosidosis (alpha-fucosidase)
- Gaucher Disease (beta-glucosidase)
- GM-1 Gangliosidosis (b-galactosidase)
- Mucopolysaccharidosis Type I, Hunter (alpha-L-iduronidase)
- Mucopolysaccharidosis Type VI, Maroteaux Lamy (arylsulfatase B)
- Mucopolysaccharidosis Type VII, Sly(beta-glucuronidase)
- Pompe (acid-alpha glucosidase)
- Tay-Sachs Disease (hexosaminidase A)

**Note:** This test will not detect carriers of these conditions.

**Methodology**

Enzyme Assay by fluorometric or spectrometric methods.

**Detection**

Dependent on level of clinical suspicion.

**Specimen Requirements**

**Type:** Whole Blood

Specimen Requirements:

In sodium heparin (green top) tube: 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.

**Special Instructions**

Customized panels with fewer enzymes available upon request.
Related Tests

- Mucopolysaccharide Screening (GA)
- Oligosaccharide Screening (OS)
- Lysosomal Storage Disease Screen, Urine (BLSDS) - includes both the GA and the OS
- All enzymes offered individually