### Condition Description

Mucopolysaccharidosis, Type I (MPS I) is caused by a deficiency of the lysosomal enzyme alpha-L-iduronidase. It should be considered in patients with corneal clouding, dysostosis multiplex, organomegaly as well as other symptoms of a storage disorder. Clinical symptoms are caused by progressive accumulation of glycosaminoglycans. There is a range of severity that may be associated with this enzyme deficiency. Patients with earlier onset, more severe course and central nervous system involvement have traditionally been described as having Hurler syndrome. Individuals with symptoms at the milder end of the spectrum have been termed Scheie syndrome and patients with intermediate symptoms are termed Hurler/Scheie.

Clinical features of MPS I include valvular heart disease, cardiomyopathy, obstructive sleep apnea, restrictive lung disease, reactive airway disease, joint stiffness, joint contractures, joint pain, spinal deformities, corneal clouding, glaucoma, developmental delay, mental retardation, communicating hydrocephalus, hearing loss, hepatomegaly, inguinal/umbilical hernia and chronic infections.

Most affected individuals have less than 1% enzyme levels regardless of disease severity. It is not clear why phenotypes vary, however, in a single family, siblings are usually similarly affected.

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, call the Emory Genetics Laboratory at (404) 778-8500.

[Click here](#) for the GeneReviews summary on this condition.

### Indications

This test is indicated for children or adults with symptoms of MPS I (alpha-iduronidase deficiency).

### Methodology

Flurometric Enzyme Assay using artificial 4-MU substrate. Alpha-iduronidase activity is evaluated to confirm a diagnosis of MPS I.

### 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.

### Related Tests

- Mucopolysaccharide screen, GAG's (GA)
- Lysosomal enzyme screening panel (LS)