Mucopolysaccharidosis: Glycosaminoglycans (GAGs), Quantitative and Qualitative, Urine

**Condition Description**

Mucopolysaccharidosis (MPS) diseases are genetic lysosomal storage diseases (LSDs) caused by the body’s inability to produce specific enzymes. Normally, the body uses enzymes to break down and recycle materials in cells. In individuals with MPS and related diseases, the missing or insufficient enzyme prevents the proper recycling process, resulting in the storage of materials, called glycosaminoglycans or GAGS, in virtually every cell of the body. As a result, cells do not perform properly and may cause progressive damage throughout the body, including the heart, bones, joints, respiratory system and central nervous system. While the disease may not be apparent at birth, signs and symptoms develop with age as more cells become damaged by the accumulation of cell materials.

While the symptoms of the diseases may vary from one syndrome to another, there are many similarities. Affected individuals often have mental retardation, cloudy corneas, short stature, stiff joints, speech and hearing impairment, large liver and spleen, hernias, heart disease, hyperactivity, pain, and a dramatically shortened life span. Currently, there are six identified MPS conditions. Most MPS diseases are autosomal recessively inherited, however one is X-linked recessively inherited, Hunter syndrome or MPS II. MPS diseases include: MPS type I (Hurler, Scheie, and Hurler-Scheie syndrome), MPS Type II (Hunter syndrome), MPS Type III (Sanfilippo Types A, B, C, and D), MPS Type IV (Morquio Types A and B), MPS Type VI (Maroteaux-Lamy syndrome), and MPS Type VII (Sly syndrome).

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

Click here for the GeneReviews summary on this condition.

**Indications**

This test is a quantitative and qualitative mucopolysaccharide screen indicated:

- for patients whose clinical evaluations are suggestive of a MPS disease.

Abnormal results should be confirmed by lysosomal enzyme analysis.

**Methodology**

Dimethylene Blue binding Quantitation and Thin Layer Chromatography.

**Detection**

Dependent on level of clinical suspicion. Abnormal results should be confirmed by lysosomal enzyme analysis.

**Reference Range**

Click here for reference range.

**Specimen Requirements**

**Type:** Urine

Specimen Requirements:

In a clean container without preservatives: 15-30 ml. Freeze.

Fasting or first void sample is preferable.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

**Related Tests**

- Lysosomal Enzyme Screen
- Oligosaccharide Screen