

Mucopolysaccharides, Screen and Quantitation

FOR SCREENING PATIENTS WITH SUSPECTED MUCOPOLYSACCHARIDOSES

Disease Overview

- Mucopolysaccharidoses (MPS) are a group of lysosomal storage disorders caused by deficiency of enzymes required for the degradation of glycosaminoglycans (GAGs).
- GAGs accumulate in the lysosomes of connective tissue, leading to tissue and organ damage.
- Clinical characteristics of MPS types can overlap, and include coarse facial features, organomegaly, progressive skeletal dysplasia, cardiomyopathy, and corneal clouding. CNS involvement and severity of symptoms can vary widely.
- The particular MPS type is defined by the deficient enzyme, which causes the accumulation of one or more specific GAGs.
- The GAGs identified with the MPS screen are dermatan, heparan, keratan, and chondroitin sulfate.
- Enzyme-replacement therapy is available for certain types of MPS if diagnosed early in life with no CNS involvement.

Epidemiology

Overall combined incidence of MPS is approximately one in 25,000.

Genetics

- Most MPS are autosomal recessive, except MPS II (Hunter), which is X-linked recessive.
- Each MPS gene is associated with many disease-causing mutations, several of which are family-specific.

Indications for Ordering

- For patients with clinical suspicion of MPS, order Mucopolysaccharides Electrophoresis & Quantitation, Urine (0081352).
- For monitoring GAG levels in patients with an established MPS diagnosis, order Mucopolysaccharides, Quantitative Urine (0081357).

Contraindications

This test will not detect carrier status.

Interpretation

- The presence of elevated GAG levels is suggestive of MPS.
- Abnormal results should be followed up with enzyme testing according to the GAGs identified.

Limitations

- The absence of elevated GAG levels does not exclude a diagnosis of MPS. Some patients with Sanfilippo syndrome (MPS III) can have normal urine mucopolysaccharides.
- This test cannot predict disease severity.
- This test is sometimes difficult to interpret in newborns or infants.

Methodology

- For Mucopolysaccharides Screen: Spectrophotometry and electrophoresis.
- For Mucopolysaccharides, Quantitative: Spectrophotometry.

References

1. Introduction to Mucopolysaccharidoses. In Atlas of Metabolic Diseases. 2nd ed. WN Nyhan, BA Barshop, PT Ozand, eds. 2005; London: Hodder Education.
2. Clarke LA. The mucopolysaccharidoses: a success of molecular medicine. *Expert Rev Mol Med* 2008;10(1):e1.
3. Muenzer J. The mucopolysaccharidoses: a heterogeneous group of disorders with variable pediatric presentations. *J Pediatr* 2004;144(5 Suppl):S27-34.

Test Information

0081352 **Mucopolysaccharides Electrophoresis & Quantitation, Urine**
0081357 **Mucopolysaccharides, Quantitative, Urine**

For specific collection, transport, and testing information, refer to the ARUP Web site at www.aruplab.com.

For information on test selection, ordering, and interpretation, refer to ARUP Consult® at www.arupconsult.com.