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


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.