

Mucopolysaccharidosis III, Three-Enzyme Panel, Blood Spot

Test ID: MPS3B

Useful for:

Supporting the biochemical diagnosis of mucopolysaccharidoses types IIIA, IIIB, IIIC

This test is **not useful for** carrier detection.

Genetics Information:

This test provides diagnostic testing for individuals with clinical signs and symptoms suspicious for mucopolysaccharidosis type IIIA, IIIB or IIIC. Enzymatic analysis for mucopolysaccharidosis (MPS) IIID is not included in this assay, however it is included in test MPS3W. If an enzyme deficiency is detected by this screening test, additional biochemical or molecular testing is required to confirm a diagnosis.

Methods:

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

Reference Values:

HEPARAN-N-SULFATASE:

>0.06 nmol/mL/hour

N-ACETYL-ALPHA-D-GLUCOSAMINIDASE:

>0.70 nmol/mL/hour

HEPARAN-ALPHA-GLUCOSAMINIDE N-ACETYLTRANSFERASE:

>0.50 nmol/mL/hour

An interpretive report will be provided.

Specimen Requirements:

Submit only 1 of the following specimen types:

Preferred:

Specimen Type: Blood spot

Supplies: Card-Blood Spot Card Collection (Filter Paper) (T493)

Preferred: Blood Spot collection Card

Acceptable: Whatman Protein Saver 903 Paper, PerkinElmer 226 filter paper, Munktell filter paper, or blood collected in tubes containing ACD or EDTA and dried on filter paper.

Specimen Volume: 2 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient 1 year of age or older is a fingerstick. See How to Collect Dried Blood Spot Samples via fingerstick.
2. At least 2 spots should be complete (ie, unpunched).
3. Let blood dry on filter paper at room temperature in a horizontal position for a minimum of 3 hours.
4. Do not expose specimen to heat or direct sunlight.
5. Do not stack wet specimens.
6. Keep specimen dry.

Specimen Stability Information: Refrigerated (preferred) 60 days/Ambient 7 days/Frozen 60 days

Additional Information:

1. For collection instructions, see [Blood Spot Collection Instructions](#)
2. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)
3. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800)

Minimum Volume: 1 Blood Spot

Acceptable:

Specimen Type: Whole blood

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD)

Specimen Volume: 2 mL

Collection Instructions: Send whole blood specimen in original tube. **Do not aliquot.**

Specimen Stability Information: Refrigerate (preferred) 7 days/Ambient: 48 hours

Minimum Volume: 0.5 mL

Specimen Stability Information:

Specimen Type	Temperature
Whole blood	Varies

Cautions:

The enzyme for mucopolysaccharidosis (MPS) IIID (N-acetylglucosamine 6-sulfatase) is not evaluated on this panel.

Individuals with pseudodeficiency alleles can show reduced enzyme activity.

Carrier status (heterozygosity) for these conditions cannot be reliably detected.

Enzyme levels may be normal in individuals receiving enzyme replacement therapy or who have undergone hematopoietic stem cell transplant.

CPT Code:

82657

83864 (if appropriate)

Day(s) Performed: Thursday

Report Available: 8 to 15 days

Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.