

Mucopolysaccharidoses, Eight-Enzyme Panel, Blood Spot

Reporting Title: MPS (Eight) Panel, BS

Performing Location: Rochester

Ordering Guidance:

To evaluate newborn patients in follow-up to an abnormal newborn screen for MPSI, the recommended tests are IDUAW / Alpha-L-Iduronidase, Leukocytes and MPSBS / Mucopolysaccharidosis, Blood Spot, MPSWB / Mucopolysaccharidosis, Blood, MPSER / Mucopolysaccharides Quantitative, Serum or MPSQU / Mucopolysaccharides Quantitative, Random, Urine.

To evaluate newborn patients in follow-up to an abnormal newborn screen for MPSII, the recommended tests are I2SB / Iduronate-2-Sulfatase, Blood Spot or I2SWB / Iduronate-2-Sulfatase, Leukocytes and MPSBS / Mucopolysaccharidosis, Blood Spot, MPSWB / Mucopolysaccharidosis, Blood, MPSER / Mucopolysaccharides Quantitative, Serum or MPSQU / Mucopolysaccharides Quantitative, Random, Urine.

Necessary Information:

- 1. Patient's age is required.
- 2. Reason for testing is required

Specimen Requirements:

Submit only 1 of the following specimen types:

Preferred:

Specimen Type: Blood spot

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

Container/Tube:

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. For detailed instructions, see How to Collect Dried Blood Spot Samples.
- 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)

Acceptable:

Specimen Type: Whole Blood



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Container/Tube:

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

Specimen Minimum Volume:

Blood Spots: 1

Whole Blood: 0.5 mL

Forms:

- 1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:
- -Informed Consent for Genetic Testing (T576)
- -Informed Consent for Genetic Testing-Spanish (T826)
- 2. Biochemical Genetics Patient Information (T602)
- 3. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole blood	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Туре	Reportable
MP8BS	BG743	Reason for Referral: • Rule out Mucopolysaccharidoses • Not Provided	Answer List	Yes

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®	
BG743	Reason for Referral	Alphanumeric		42349-1	
618405	Iduronate-2-sulfatase	Numeric	nmol/mL/h	79462-8	
618406	Heparan-N-sulfatase	Numeric	nmol/mL/h	104113-6	

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Result ID	Reporting Name	Туре	Unit	LOINC®
618407	N-acetyl-alpha-D-glucosaminidase	Numeric	nmol/mL/h	104114-4
618408	Heparan-alpha-glucosaminide N-acetyltransferase	Numeric	nmol/mL/h	104115-1
618409	N-acetylgalactosamine-6-sulfatase		nmol/mL/h	88019-5
618410	Beta-galactosidase	Numeric	nmol/mL/h	55916-1
618411	Arylsulfatase B	Numeric	nmol/mL/h	55912-0
618412	Beta-glucuronidase	Numeric	nmol/mL/h	79457-8
618413	Interpretation	Alphanumeric		59462-2
618404	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82657 83864 (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
MPSBS	Mucopolysaccharidosis, BS			No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Туре	Unit	LOINC®
MPSBS	43695	Interpretation (MPSBS)	Alphanumeric		59462-2
MPSBS	43693	Dermatan Sulfate	Numeric	nmol/L	90233-8
MPSBS	43694	Heparan Sulfate	Numeric	nmol/L	90235-3
MPSBS	BA2869	Total Keratan Sulfate	Alphanumeric	nmol/L	90236-1
MPSBS	43696	Reviewed By	Alphanumeric		18771-6



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Reference Values:

Iduronate-2-sulfatase: >4.30 nmol/mL/hour 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

N-acetylgalactosamine-6-sulfatase: >0.70 nmol/mL/hour

Beta-galactosidase: >1.30 nmol/mL/hour Arylsulfatase B: >0.90 nmol/mL/hour Beta-glucuronidase: >2.60 nmol/mL/hour

An interpretive report will be provided.