

Reporting Title: MPS III (Three) Panel, BS **Performing Location:** Rochester

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 older than 1 year 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 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)

ABORATORIES

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 |
|---------|-------------|---|-------------|------------|
| MPS3B | BG751 | Reason for Referral: • Rule out MPS III • Follow up of known MPS IIIA • Follow up of known MPS IIIB • Follow up of known MPS IIIC • Not Provided | Answer List | Yes |

Result Codes:

| Result ID | Reporting Name | Туре | Unit | LOINC® |
|-----------|---|--------------|-----------|----------|
| BG751 | Reason for Referral | Alphanumeric | | 42349-1 |
| 618419 | Heparan-N-sulfatase | Numeric | nmol/mL/h | 104113-6 |
| 618420 | N-acetyl-alpha-D-glucosaminidase | Numeric | nmol/mL/h | 104114-4 |
| 618421 | Heparan-alpha-glucosaminide N-acetyltransferase | Numeric | nmol/mL/h | 104115-1 |
| 618422 | Interpretation | Alphanumeric | | 59462-2 |
| 618418 | 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 |

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.