

Test Definition: MPS4B

Mucopolysaccharidosis IV Enzyme Panel, Blood Spot

Reporting Title: MPS IV 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 <u>Blood Spot Collection Instructions</u>
- 2. For collection instructions in Spanish, see <u>Blood Spot Collection Card-Spanish Instructions</u> (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

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)



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- -<u>Informed Consent for Genetic Testing-Spanish</u> (T826)
- 2. Biochemical Genetics Patient Information (T602)
- 3. If not ordering electronically, complete, print, and send a <u>Biochemical Genetics Test Request</u> (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
MPS4B	BG753	Reason for Referral:	Answer List	Yes
		Rule out MPS IV		
		Follow up of known MPS IVA		
		Follow up of known MPS IVB		
		• Follow up of known GM1		
		Not Provided		

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
BG753	Reason for Referral	Alphanumeric		42349-1
618424	N-acetylgalactosamine-6-sulfatase	Numeric	nmol/mL/h	88019-5
618425	Beta-galactosidase	Numeric	nmol/mL/h	55916-1
618426	Interpretation	Alphanumeric		59462-2
618423	Reviewed By	Alphanumeric		18771-6

 $\ensuremath{\mathsf{LOINC}}\xspace^{\ensuremath{\texttt{g}}}$ 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	Available Separately
MPSBS	Mucopolysaccharidosis, BS	1	83864	No	Yes



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Result Codes for Reflex Tests:

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

Reference Values:

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

Beta-galactosidase: >1.30 nmol/mL/hour

An interpretive report will be provided.