MAYO CLINIC LABORATORIES

Test Definition: MSDBS

Multiple Sulfatase Deficiency, Blood Spot

Reporting Title: Multiple sulfatase deficiency, 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 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
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)
-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	Type	Reportable
MSDBS	BG755	Reason for Referral: • Rule out MSD • Follow up of known MSD • Multiple Sulfatase deficiency • Not Provided	Answer List	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
BG755	Reason for Referral	Alphanumeric		42349-1
618430	Iduronate-2-sulfatase	Numeric	nmol/mL/h	79462-8
618431	Heparan-N-sulfatase	Numeric	nmol/mL/h	104113-6
618432	N-acetylgalactosamine-6-sulfatase	Numeric	nmol/mL/h	88019-5
618433	Interpretation	Alphanumeric		59462-2
618429	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	Available Separately
MPSBS	Mucopolysaccharidosis, BS	1	83864	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	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:

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

Heparan-N-sulfatase: >0.06 nmol/mL/hour

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

An interpretive report will be provided.