

Reporting Title: Multiple sulfatase deficiency, WBC**Performing Location:** Rochester**Shipping Instructions:**

For optimal isolation of leukocytes, it is recommended the specimen arrive refrigerated within 6 days of collection to be stabilized. Collect specimen Monday through Thursday only and not the day before a holiday. Specimen should be collected and packaged as close to shipping time as possible.

Necessary Information:

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

Specimen Requirements:

Container/Tube:

Preferred: Yellow top (ACD solution B)

Acceptable: Yellow top (ACD solution A) or lavender top (EDTA)

Specimen Volume: 6 mL

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

Specimen Minimum Volume:

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 ACD	Refrigerated (preferred)	6 days	
	Ambient	6 days	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
MSDW	BG771	Reason for Referral: <ul style="list-style-type: none">• Rule out MSD• Follow up of known MSD• Not Provided	Answer List	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
BG771	Reason for Referral	Alphanumeric		42349-1
618466	Iduronate-2-sulfatase	Numeric	nmol/h/mg Prot	24089-5
618467	Heparan-N-sulfatase	Numeric	nmol/h/mg Prot	24086-1
620156	N-acetylglucosamine-6-sulfatase	Numeric	nmol/h/mg Prot	24098-6
618468	N-acetylgalactosamine-6-sulfatase	Numeric	nmol/h/mg Prot	24096-0
618469	Interpretation	Alphanumeric		59462-2
618465	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82657

Reference Values:

Iduronate-2-sulfatase: >2.20 nmol/hour/mg protein
Heparan-N-sulfatase: >0.13 nmol/hour/mg protein
N-acetylglucosamine-6-sulfatase: >0.03 nmol/hour/mg protein
N-acetylgalactosamine-6-sulfatase: >1.60 nmol/hour/mg protein

An interpretive report will be provided.