

LYSOSOMAL DISORDERS SCREEN, BLOOD SPOT #89406

USEFUL FOR: Evaluation of patients with a clinical presentation suggestive of one of the following lysosomal storage disorders: Gaucher, Niemann-Pick A/B, Pompe, Krabbe, or Fabry disease.

METHODOLOGY: Tandem Mass Spectrometry (MS/MS)

REFERENCE VALUES:

DISEASE NAME	ENZYME DEFICIENT	NORMAL WHEN
Gaucher disease	Acid beta-glucosidase (GBA)	> or = 4.1 nmol/mL blood/hr
Niemann-Pick A/B disease	Acid sphingomyelinase (ASM)	> or = 1.0 nmol/mL blood/hr
Pompe disease	Acid alpha-glucosidase (GAA)	> or = 4.0 nmol/mL blood/hr
Krabbe disease	Galactocerebrosidase (GALC)	> or = 0.5 nmol/mL blood/hr
Fabry disease	Alpha-galactosidase A (GLA)	> or =2.0 nmol/mL blood/hr (males) > or =4.2 nmol/mL blood/hr (females)

SPECIMEN REQUIREMENTS: 2 blood spots on Whatman 903 filter paper is preferred (Mayo Newborn Screening Card [Supply T-493] is also acceptable.) Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours. An alternative blood collection option for a patient > 1 year of age is a fingerstick.

NOTE:

1. Do not expose specimen to heat or direct sunlight.
2. Do not stack wet specimens.
3. Do not use device or capillary tube containing EDTA to collect specimen.
4. Keep specimen dry.

CAUTIONS: Samples exposed to heat >25 degrees C for more than 48 hours will yield higher activity levels than properly submitted samples. This may cause false-normal (false-negative) results in affected patients.

LIST FEE: \$ 88.20

CPT CODE: 83789

ANALYTIC TIME: 3 days

DAY(S) SET-UP: Monday, 8:00 a.m.
Thursday, 8:00 a.m.

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Sara Siewert, Mayo Medical Laboratories' Technologist Support
Telephone: 800-533-1710



TEST DEFINITION

10/28/2009

ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PRINT NORMALS (# CODED)	PERFORM SITE *
89406	8/21/2009		LYSOSOMAL DISORDERS SCREEN, BS			MCR
		50859	TRANSPORT TEMP : AMBIENT\REFRIG OK\FROZEN OK SPECIMEN			
		50860	SPECIMEN ID			
		50861	SOURCE			
		50862	ORDER DATE			
		50863	REASON FOR REFERRAL			
		50864	METHOD			
		50865	ACID BETA-GLUCOSIDASE			
			UNITS: NMOL/ML/HR			
			NO SEX			
			ALL AGES : 4.1-		; >=4.1	
			MALE			
			ALL AGES : 4.1-		; >=4.1	
			FEMALE			
			ALL AGES : 4.1-		; >=4.1	
		50866	SPHINGOMYELINASE			
			UNITS: NMOL/ML/HR			
			NO SEX			
			ALL AGES : 1.0-		; >=1.0	

ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PRINT NORMALS (# CODED)	PERFORM SITE *
89406 (CONTINUED)		50866	SPHINGOMYELINASE (CONTINUED)			
			MALE			
			ALL AGES : 1.0-		; >=1.0	
			FEMALE			
			ALL AGES : 1.0-		; >=1.0	

```

- - - - -
50867  ACID ALPHA-GLUCOSIDASE
        UNITS: NMOL/ML/HR
        NO SEX
        ALL AGES : 4.0-           ; >=4.0
        MALE
        ALL AGES : 4.0-           ; >=4.0
        FEMALE
        ALL AGES : 4.0-           ; >=4.0
- - - - -
50868  GALACTOCEREBROSIDASE
        UNITS: NMOL/ML/HR
        NO SEX
        ALL AGES : 0.5-           ; >=0.5
        MALE
        ALL AGES : 0.5-           ; >=0.5
        FEMALE
        ALL AGES : 0.5-           ; >=0.5
- - - - -
50869  ALPHA-GALACTOSIDASE
        UNITS: NMOL/ML/HR
        MALE
        ALL AGES : 2.0-           ; >=2.0
        FEMALE
        ALL AGES : 4.2-           ; >=4.2
- - - - -
50870  INTERPRETATION
- - - - -
50871  AMENDMENT
- - - - -
50872  REVIEWED BY
- - - - -
50873  RELEASE DATE
- - - - -

```

*PERFORMING SITE LEGEND

MCR MAYO CLINIC DPT OF LAB MED & PATHOLOGY
 200 FIRST STREET SW
 ROCHESTER, MN 55905

LAB DIRECTOR: FRANKLIN R. COCKERILL, III, M.D.

MML MESSAGES USED AS NORMALS

CODE TEXT

TOTAL OF 0 NORMALS CODE
 *** END OF REPORT ***

1-800-533-1710

PATIENT NAME TESTING, LDSQAMALE		PATIENT NUMBER		AGE 33	SEX M	ACCESSION # G9134937
ORDERING PHYSICIAN		CLIENT ORDER #			ACCOUNT # LIAISONS	
COLLECTION 09/17/09 10:58 A	RECEIVED 09/17/09 10:58 A	REPORT PRINTED 10/28/09 11:19 A		SPECIMEN INFORMATION DATE OF BIRTH:		
DATE	TIME	DATE	TIME	DATE	TIME	
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI	LO	REF RANGE	PERFORM SITE *
----------------	----	----	-----------	----------------

Lysosomal Disorders Screen, BS

Specimen	Blood Spot			MCR
Specimen ID	1038571			MCR
Order Date	18 Sep 2009 10:10			MCR
Reason For Referral				MCR
Reason for referral not provided.				
Method				MCR
Tandem Mass Spectrometry (MS/MS)				
Acid Beta-Glucosidase	4.1	nmol/mL/hr	>=4.1	MCR
Sphingomyelinase	1.0	nmol/mL/hr	>=1.0	MCR
Acid Alpha-Glucosidase	4.0	nmol/mL/hr	>=4.0	MCR
Galactocerebrosidase	0.5	nmol/mL/hr	>=0.5	MCR
Alpha-Galactosidase	L 1.9	nmol/mL/hr	>=2.0	MCR
Interpretation				MCR

In this specimen, the activity of alpha-galactosidase is reduced indicating that this individual is affected with Fabry disease (OMIM #301500). The other enzymes tested by this method were normal (beta-glucosidase, acid alpha-glucosidase, sphingomyelinase, and galactosylceramide beta-galactosidase). For confirmation, consider ordering enzyme analysis in leukocytes (MML#8785) or molecular genetic analysis of the GLA gene (MML #88264). If not already ordered, molecular genetic analysis could be performed on the existing sample by calling MML (1-800-533-1710) within one month. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

Reviewed By	Dietrich Matern MD	MCR
Release Date	18 Sep 2009 11:02	MCR

* Perform Site Legend on last page of report

PATIENT NAME TESTING, LDSQAMALE	ORDER STATUS Final	COLLECTION DATE AND TIME 09/17/09 10:58 A
------------------------------------	-----------------------	--

1-800-533-1710

PATIENT NAME TESTING, LDSQAMALE		PATIENT NUMBER		AGE 33	SEX M	ACCESSION # G9134937
ORDERING PHYSICIAN		CLIENT ORDER #				ACCOUNT # LIAISONS
COLLECTION 09/17/09 10:58 A	RECEIVED 09/17/09 10:58 A	REPORT PRINTED 10/28/09 11:19 A		SPECIMEN INFORMATION DATE OF BIRTH:		
DATE TIME	DATE TIME	DATE TIME				
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
-----------------------	------------------------	------------------	-----------------------

* PERFORMING SITE

MCR Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
---	--

PATIENT NAME TESTING, LDSQAMALE	ORDER STATUS Final	COLLECTION DATE AND TIME 09/17/09 10:58 A
---	------------------------------	---