

ALPHA-GALACTOSIDASE, BLOOD SPOTS
#89407

USEFUL FOR: Evaluation of patients with a clinical presentation suggestive of Fabry Disease.

METHODOLOGY: Fluorometric Enzyme Assay

REFERENCE VALUES: Males > or = 1.2 nmol/mL blood/hr
Females > or = 2.8 nmol/mL blood/hr

SPECIMEN REQUIREMENTS: Two blood spots from Whatman 903 filter paper are preferred (Mayo Newborn Screening Card [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 finger stick.

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.

LIST FEE: \$271.30

CPT CODE: 82657

ANALYTIC TIME: 2 days

DAY(S) SET-UP: Thursday; morning.

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



TEST DEFINITION

9/25/2009

ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PRINT NORMALS (# CODED)	PERFORM SITE *
89407	9/10/2009		ALPHA-GALACTOSIDASE, BS			MCR
		50883	TRANSPORT TEMP : AMBIENT\REFRIG OK\FROZEN OK SPECIMEN			
		50884	SPECIMEN ID			
		50885	SOURCE			
		50886	ORDER DATE			
		50887	REASON FOR REFERRAL			
		50888	METHOD			
		50889	ALPHA-GALACTOSIDASE, BS			
			UNITS: NMOL/ML/HR			
			MALE		; >=1.2	
			FEMALE			
			ALL AGES : 2.8-		; >=2.8	
		50890	INTERPRETATION			
		50891	AMENDMENT			
		50892	REVIEWED BY			

ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PRINT NORMALS (# CODED)	PERFORM SITE *
89407		50893	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.

*** END OF REPORT ***

MML
LAB

MESSAGES USED AS NORMALS

CODE	TEXT
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TOTAL OF 0 NORMALS CODES

*** END OF REPORT ***

1-800-533-1710

PATIENT NAME TESTING, AGABSQA		PATIENT NUMBER		AGE 33	SEX F	ACCESSION # G9134592
ORDERING PHYSICIAN		CLIENT ORDER #				ACCOUNT # LIAISONS
COLLECTION 09/11/09 10:24 A	RECEIVED 09/11/09 10:24 A	REPORT PRINTED 09/25/09 10:44 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 *
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Alpha-Galactosidase, BS

Specimen	Blood Spot	MCR
Specimen ID	1038550	MCR
Order Date	14 Sep 2009 11:26	MCR
Reason For Referral		MCR

Reason for referral not provided.

Method		MCR
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Fluorometric Enzyme Assay

Alpha-Galactosidase,	L	2.7	nmol/mL/hr	>=2.8	MCR
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BS

Interpretation		MCR
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In this sample, the activity of alpha-galactosidase enzyme is reduced indicating that this individual is a carrier for Fabry disease (OMIM #301500). Carriers may or may not be symptomatic. If clinically indicated, consider follow-up investigations to include molecular genetic analysis of the GLA gene (MML 88264). If not already ordered, this could be done on the existing sample by calling MML (1-800-533-1710) within one month. In addition, the Fabry Registry website (<http://www.Fabryregistry.com>) provides information and resources for health professionals and families. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

Reviewed By	Kimiyo M Raymond MD	MCR
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Release Date	15 Sep 2009 10:13	MCR
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* 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.
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PATIENT NAME TESTING, AGABSQA	ORDER STATUS Final	COLLECTION DATE AND TIME 09/11/09 10:24 A
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