

**PHOSPHOMANNOMUTASE AND PHOSPHOMANNOSE ISOMERASE,
FIBROBLASTS
#89657**

USEFUL FOR: The detection of, and distinguishing between, deficiencies of phosphomannomutase (PMM, CDG-1a) and phosphomannose isomerase (PMI, CDG-1b) as associated with congenital disorders of glycosylation (CDG).

As a follow-up test in patients of all ages with an abnormal transferrin isoform profile as determined by isoelectric focusing or liquid chromatography-mass spectrometry.

METHODOLOGY: Colorimetric

REFERENCE VALUES: PMM Fibroblasts: Normal > 700 nmol/hr/mg protein

PMI Fibroblasts: Normal > 1,500 nmol/hr/mg protein

SPECIMEN REQUIREMENTS: Submit only 1 of the following specimens.

Cultured Fibroblasts

1 T-75 or 2 T-25 flask(s) filled to neck with culture media. (**Specimen received in formalin or fixative preservative is not acceptable.**) Maintain sterility and forward promptly at ambient temperature. Specimen cannot be frozen.

Skin Biopsy

Collect sterile skin biopsy (4-mm punch) in a screw-capped, sterile container filled with any standard cell culture media (eg, minimal essential media, RPMI 1640, etc.). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]). (**Specimen received in formalin or fixative preservative is not acceptable.**) Send specimen refrigerated. Specimen cannot be frozen. Maintain sterility and forward promptly.

NOTE: When an order is filed for this test, #8482 Fibroblast Culture and #88832 Cryopreservation for Biochemical Studies are automatically added by the system.

LIST FEE: \$ 271.30

CPT CODE: 82657

ANALYTIC TIME: 30-45 days

DAY(S) SET-UP: Varies

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

TEST DEFINITION

8/27/2009

CODE NAME

89657 PMM-PMI, FIBROBLASTS

ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PRINT NORMALS (# CODED)	PERFORM SITE *
89657	7/10/2009		PMM-PMI, FIBROBLASTS			MCR
			TRANSPORT TEMP : VARIES			
			AMBIENT\REFRIG <24 HOURS OK\FROZEN NO-CULTURED FIBROBLASTS			
			REFRIG\AMBIENT OK\FROZEN NO-SKIN BIOPSY			
50824			SPECIMEN			
50825			SPECIMEN ID			
50826			SOURCE			
50827			ORDER DATE			
50828			REASON FOR REFERRAL			
50829			METHOD			
50830			PHOSPHOMANNOMUTASE, FIBRO			
			UNITS: NMOL/HR/MG PROT			
			NO SEX			
			ALL AGES : 701-		>700	
			MALE			
			ALL AGES : 701-		>700	
			FEMALE			
			ALL AGES : 701-		>700	
50831			PHOSPHOMANNOSE ISOMERASE, FIBRO			
			UNITS: NMOL/HR/MG PROT			

NO SEX
ALL AGES : 1501- ; >1500
MALE
ALL AGES : 1501- ; >1500
FEMALE
ALL AGES : 1501- ; >1500

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50832 INTERPRETATION
- - - - -
50833 AMENDMENT
- - - - -
50834 REVIEWED BY
- - - - -
50835 RELEASE DATE
- - - - -

*PERFORMING SITE LEGEND

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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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1-800-533-1710

PATIENT NAME TESTING, PMMIF		PATIENT NUMBER		AGE 33	SEX F	ACCESSION # G9133046
ORDERING PHYSICIAN		CLIENT ORDER #				ACCOUNT # LIAISONS
COLLECTION 08/04/09 12:26 P DATE TIME	RECEIVED 08/04/09 12:26 P DATE TIME	REPORT PRINTED 08/27/09 02:38 P DATE TIME		SPECIMEN INFORMATION DATE OF BIRTH:		
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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Fibroblast Culture

Fibroblast Culture

Not applicable MCR

Fibroblasts successfully cultured.

Cryopreserve for Biochem Studies

 Cryopreserve for
 Biochem Studies

MCR

Fibroblasts successfully cryopreserved.

PMM-PMI, Fibroblasts

Specimen	Fibroblasts	MCR
Specimen ID	1038485	MCR
Order Date	14 Aug 2009 11:46	MCR
Reason For Referral		MCR

Reason for referral not provided.

Method	Colorimetric	MCR
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Phosphomannomutase, Fibro	701	nmol/hr/mg prot	>700	MCR
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Phosphomannose Isomerase, Fibro	1501	nmol/hr/mg prot	>1500	MCR
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Interpretation		MCR
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In this sample, the activities of both phosphomannomutase II (PMM) and phosphomannose isomerase (PMI) were normal. These results indicate this individual is NOT affected with either congenital disorder of glycosylation (CDG) type Ia (OMIM 212065) or CDG type Ib (OMIM 602579). However, please note that this test does not rule out other types of CDG. If clinically indicated and not done already, consider repeating the Carbohydrate Deficient Transferrin, Serum (MML 82414), assay. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) to

* Perform Site Legend on last page of report

PATIENT NAME TESTING, PMMIF	ORDER STATUS Final	COLLECTION DATE AND TIME 08/04/09 12:26 P
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1-800-533-1710

PATIENT NAME TESTING, PMMIF		PATIENT NUMBER		AGE 33	SEX F	ACCESSION # G9133046
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DATE TIME	DATE TIME	DATE TIME				
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TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
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determine the best testing strategy or if you have any questions.

Reviewed By	Kimiyo M Raymond MD	MCR
Release Date	18 Aug 2009 09:07	MCR

* 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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