Test Definition: PMMIF

Phosphomannomutase (PMM) and Phosphomannose Isomerase (PMI), Fibroblasts

Reporting Title: PMM-PMI, Fibroblasts
Performing Location: Rochester

Specimen Requirements:

This test is not recommended for prenatal testing.

Forms:
1. 1. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.
2. 2. If not ordering electronically, submit a Biochemical Genetics Request Form (Supply T439) with the specimen.

Submit only 1 of the following specimens:

Specimen Type: Cultured fibroblasts
Container/Tube: T-75 or T-25 flask
Specimen Volume: 1 Full T-75 flask or 2 full T-25 flasks
Specimen Stability Information: Ambient (preferred)/Refrigerated 24 hours

Specimen Type: Skin biopsy
Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). 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 Volume: 4-mm punch
Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type	Temperature	Time
Tissue	Varies	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
50824	Specimen	Alphanumeric		31208-2
50825	Specimen ID	Numeric		N/A
50826	Source	Alphanumeric		N/A
50827	Order Date	Alphanumeric		N/A
50828	Reason For Referral	Alphanumeric		42349-1
50829	Method	Alphanumeric		In Process
50830	Phosphomannomutase, Fibro	Numeric	nmol/h/mg Prot	In Process

Result ID	Reporting Name	Type	Unit	LOINC®
50831	Phosphomannose Isomerase, Fibro	Numeric	nmol/h/mg Prot	In Process
50832	Interpretation	Alphanumeric		59462-2
50833	Amendment	Alphanumeric		In Process
50834	Reviewed By	Numeric		N/A
50835	Release Date	Alphanumeric		N/A

CPT Code: 1 × 82657

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FIBR	Fibroblast Culture	1	88233	Yes	Yes
CRYOB	Cryopreserve for Biochem Studies	1	88240	Yes	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FIBR	8482	Fibroblast Culture	Alphanumeric		In Process
CRYOB	88832	Cryopreserve for Biochem Studies	Alphanumeric		N/A

Reference Values:

PMM
Normal >700 nmol/h/mg Prot

PMI
Normal >1,500 nmol/h/mg Prot