

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

Ordering Guidance:

The initial screening test for congenital disorders of glycosylation is transferrin isoform analysis (CDG / Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum). The results of the transferrin isoform analysis should be correlated with the clinical presentation to determine the most appropriate testing strategy, which may include this test.

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.

Specimen Requirements:

Container/Tube:

Preferred: Yellow top (ACD solution B)

Acceptable: Yellow top (ACD solution A)

Specimen Volume: 6 mL

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

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 in Special Instructions:
- [Informed Consent for Genetic Testing](#) (T576)
 - [Informed Consent for Genetic Testing-Spanish](#) (T826)
2. [Biochemical Genetics Patient Information](#) (T602) in Special Instructions
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	YELLOW TOP/ACD
	Ambient	6 days	YELLOW TOP/ACD

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
50842	Phosphomannomutase, Leuko	Numeric	nmol/h/mg Prot	78970-1
50843	Phosphomannose Isomerase, Leuko	Numeric	nmol/h/mg Prot	78963-6
50840	Reason For Referral	Alphanumeric		42349-1
50836	Specimen	Alphanumeric		31208-2
50837	Specimen ID	Numeric		57723-9
50838	Source	Alphanumeric		31208-2

50839	Order Date	Alphanumeric		82785-7
50841	Method	Alphanumeric		85069-3
50845	Amendment	Alphanumeric		48767-8
50847	Release Date	Alphanumeric		82772-5
50844	Interpretation	Alphanumeric		59462-2
50846	Reviewed By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82657

Reference Values:

PHOSPHOMANNOMUTASE

Normal >350 nmol/h/mg protein

PHOSPHOMANNOSE ISOMERASE

Normal >1,300 nmol/h/mg protein