

Test Definition: PMPDD

PMP22 Gene, Large Deletion/Duplication Analysis, Varies

Reporting Title: PMP22 Gene, Deletion/Duplication **Performing Location:** Rochester

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.

2. Send specimen in original tube.

Additional Information: To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

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. Molecular Genetics: Neurology Patient Information in Special Instructions

3. If not ordering electronically, complete, print, and send a <u>Neurology Specialty Testing Client Test Request</u> (T732) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
113371	Result Summary	Alphanumeric		50397-9
113374	Result	Alphanumeric		75384-8
113375	Interpretation	Alphanumeric		69047-9
113376	Additional Information	Alphanumeric		48767-8
113377	Specimen	Alphanumeric		31208-2
113378	Source	Alphanumeric		31208-2



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113379	Released By	Alphanumeric	18771-6

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

Supplemental Report: No

CPT Code Information: 81324

Reference Values:

An interpretive report will be provided.