

Reporting Title: PMS2 Gene, Known Mutation

Performing Location: Rochester

Specimen Requirements:

Specimen must arrive within 96 hours of collection.

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.

Forms:

1. Molecular Genetics-Inherited Cancer Syndromes Patient Information Sheet (Supply T519) in Special Instructions
2. 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.

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
33625	Reason for Referral	Alphanumeric		In Process
33626	Result	Alphanumeric		In Process
33627	Interpretation	Alphanumeric		In Process
33628	Method	Alphanumeric		In Process
33629	Amendment	Alphanumeric		In Process
33630	Specimen	Alphanumeric		In Process
33631	Source	Alphanumeric		In Process
33632	Reviewed By	Alphanumeric		In Process
33633	Release Date	Alphanumeric		In Process

Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
Billing only	MOLECULE ISOLATE NUCLEIC	1	83891		
Billing only	GENETIC EXAMINATION	1	83912		

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FUSEQ	DNA Sequence, Follow-up Analysis		Profile	No	No
PMSLD	PMS2 Gene, Large Del/Dup Analysis		Profile	No	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FUSEQ	17231	Specimen	Alphanumeric		31208-2
FUSEQ	17232	Specimen ID	Alphanumeric		In Process
FUSEQ	17233	Source	Alphanumeric		N/A
FUSEQ	17234	Order Date	Alphanumeric		N/A
FUSEQ	17235	Result	Alphanumeric		In Process
FUSEQ	17236	Amendment	Alphanumeric		In Process
FUSEQ	17237	Reviewd By	Alphanumeric		In Process
FUSEQ	17238	Released Date	Alphanumeric		N/A
PMSLD	33636	Result	Alphanumeric		In Process
PMSLD	33637	Amendment	Alphanumeric		In Process
PMSLD	33638	Specimen	Alphanumeric		In Process
PMSLD	33639	Source	Alphanumeric		In Process
PMSLD	33640	Reviewed By	Alphanumeric		In Process
PMSLD	33641	Release Date	Alphanumeric		In Process

Reference Values:

An interpretive report will be provided.