MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: November 1, 2012
EFFECTIVE DATE: November 14, 2012

PMS2 Gene, Full Gene Analysis
Test ID: PMS2S

USEFUL FOR:
- Establishing a diagnosis of Lynch syndrome/hereditary nonpolyposis colorectal cancer Determining whether absence of PMS2 protein in tumor tissue, as demonstrated by immunohistochemistry, is associated with a germline mutation in the affected individual
- Identification of familial *PMS2* mutation to allow for predictive testing in family members

METHODOLOGY: Polymerase Chain Reaction (PCR) followed by DNA Sequence Analysis and Gene Dosage Analysis by Multiplex Ligation-Dependent Probe Amplification (MLPA).

REFERENCE VALUES: An interpretive report will be provided.

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.

NOTE:
- Molecular Genetics-Inherited Cancer Syndromes Patient Information Sheet (Supply T519) in Special Instructions
- **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 STABILITY INFORMATION:

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

CAUTIONS:

- Some individuals who have a diagnosis of *PMS2*-related Lynch syndrome may have a mutation that is not identified by this method (eg, deep intronic mutations, promoter mutations). The absence of a mutation, therefore, does not eliminate the possibility of a diagnosis of Lynch syndrome. For predictive testing of asymptomatic individuals, it is important to first document the presence of a *PMS2* gene mutation in an affected family member.
- In some cases, DNA alterations of undetermined significance may be identified.
- We strongly recommend that asymptomatic patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.
- Predictive testing of an asymptomatic child is not recommended.
- Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.
- In addition to disease-related probes, the multiplex ligation-dependent probe amplification technique utilizes probes localized to other chromosomal regions as internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing.

FEE: Please contact your Regional Manager for your account's fee information.

CPT CODE:
81317 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; full sequence analysis
81319 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; duplication/deletion variants

For non-participating payers:

83891- Isolation or extraction of highly purified nucleic acid
83898 x 7- Amplification, target, each nucleic acid sequence
83909 x 33- Separation and identification by high-resolution technique
83894 x 3- Separation by gel electrophoresis
83900- Amplification, target, multiplex, first 2 nucleic acid sequences
83914 x34 Mutation identification by enzymatic ligation or primer extension, single segment, each segment
83912- Interpretation and report

DAY(S) SET UP: Monday, 2 pm **ANALYTIC TIME**: 14 days

NOTE: The following referral test code(s) will become obsolete.

Test Name	Test ID	Referral Lab Code	Referral Lab
HNPCC/Lynch Syndrome (PMS2) Sequencing and Deletion/Duplication	ZW252	0051737	ARUP Laboratories
PMS2 Comprehensive - Sequence & Deletion/Duplication Analysis	ZW221	6890	Baylor Medical Genetics Laboratories

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710