MAYO
Mayo Medical Laboratories
1-800-533-1710

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

PMS2 Gene, Known Mutation
Test ID: PMS2K

USEFUL FOR:
- Diagnostic testing of individuals with suspected diagnosis of Lynch syndrome/ hereditary nonpolyposis colorectal cancer when a mutation in the *PMS2* gene has been identified in an affected family member
- Predictive testing of at-risk individuals when a mutation in the *PMS2* gene has been identified in an affected family member

METHODOLOGY: Polymerase chain reaction (PCR) followed by DNA sequencing or 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:

- The identification of a disease-causing mutation in an affected family member is necessary before predictive testing for other family members can be performed. If a familial mutation has not been previously identified, order PMS2S/61173 *PMS2* Gene, Full Gene Analysis.
- Analysis is performed for the familial mutations provided only. This assay does not rule out the presence of other mutations within this gene or within other genes that may be associated with hereditary cancer syndromes.
- We strongly recommend that 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.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Any error in the diagnosis or in the pedigree provided to us, including false-paternity, could lead to erroneous interpretation of results.
- 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.

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

CPT CODE:
81318 PMS2 (postmeiotic segregation increased 2 [S. cerevisiae]) (eg, hereditary non-polyposis colorectal cancer, Lynch syndrome) gene analysis; known familial variants
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:

"PMS2 Gene, Known Mutation"
83891- Isolation or extraction of highly purified nucleic acid
83912- Interpretation and report

"PMS2 Gene, Large Deletion and Duplication Analysis"
83900- Amplification, target, multiplex, first 2 nucleic acid sequences (if appropriate)
83909- Separation and identification by high-resolution technique (if appropriate)
83914 x34- Mutation identification by enzymatic ligation or primer extension, single segment, each segment (if appropriate)

"DNA Sequence, Follow-up Analysis"
83898-Amplification, target, each nucleic acid sequence (if appropriate)
83909 x2- Separation and identification by high-resolution technique (if appropriate.

DAY(S) SET UP: Monday, 2 pm **ANALYTIC TIME**: 10 days

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