MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: June 28, 2013
EFFECTIVE DATE: July 3, 2013

TP53 Gene, Full Gene Analysis
Test ID: P53MS

USEFUL FOR:
- Confirmation of suspected clinical diagnosis of Li-Fraumeni syndrome or Li-Fraumeni-like syndrome
- Identification of familial TP53 mutation to allow for predictive testing in family members

METHODOLOGY: Polymerase Chain Reaction (PCR) Amplification/DNA Sequencing and gene dosage analysis by array comparative genomic hybridization (aCGH)

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: **Specimen must arrive within 96 hours of draw.**

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- Colon Cancer Patient Information Sheet (Supply T521) 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 Li-Fraumeni syndrome or Li-Fraumeni-like 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 Li-Fraumeni syndrome or Li-Fraumeni-like syndrome. For predictive testing of asymptomatic individuals, it is important to first document the presence of a TP53 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.

CPT CODE:
81405-TP53 (tumor protein 53) (eg, Li-Fraumeni syndrome, tumor samples), full gene sequence or targeted sequence analysis of >5 exons

Hereditary Colon Cancer CGH Array
81228-Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number variants (eg, Bacterial Artificial Chromosome [BAC] or oligo-based comparative genomic hybridization [CGH] microarray analysis)

DAY(S) SET UP: Wednesday 10 a.m. **ANALYTIC TIME**: 14 days

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

Test Name	Test ID	Referral Lab Code	Referral Lab
Li Fraumeni Syndrome (p53 Tumor Supressor Gene)	ZW172	MML171	Center for Genetic Testing
Li-Fraumeni Syndrome (LFS) TP53 Amplified	ZW185	2866	Ambry
Li-Fraumeni Syndrome (LFS) TP53 Deletion/Duplication Analysis	ZW185	2864	Ambry
p53 Gene MLPA Analysis	ZW171	P53-DEL-CAS	City of Hope
p53 Gene MLPA Analysis	ZW171	P53-DEL	City of Hope
p53 Gene MLPA Analysis	ZW171	P53-CAS	City of Hope
p53 Gene MLPA Analysis	ZW171	P53-SEQ	City of Hope
TP53 (p53) Gene Analysis in Li-Fraumeni Syndrome (LFS)	ZW168	559	GeneDx
TP53 Comprehensive - Sequence & Deletion / Duplication Analysis	ZW221	6821	Baylor

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