MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: January 28, 2014
EFFECTIVE DATE: January 30, 2014

Hematologic neoplasms, *TP53* Somatic Mutation, DNA Sequencing Exons 4-9

Test ID: P53CA

USEFUL FOR:

- Evaluating chronic lymphocytic leukemia patients at diagnosis or during disease course for the presence of TP53 gene mutations indicating high risk of disease progression and adverse outcome.

- This test is complementary to FISH analysis for the 17p- abnormality, but more appropriately identifies the presence of mutational alteration and gene inactivation in tumor cells. For hereditary (germ line) TP53 mutation syndrome testing, see P53MS / TP53 Gene, Full Gene Analysis.

REFLEX TESTS:

Test ID	Reporting Name	Available Separately	Always Performed
CKP53	CKP53 Protocol, B	No	No

TESTING ALGORITHM: Flow cytometry CKP53 / CKP53 Protocol, Blood may be performed on peripheral blood samples to verify diagnosis of chronic lymphocytic leukemia (CLL) and determine the % B-cells in the sample prior to TP53 testing. See TP53 Sequencing Testing Algorithm in Special Instructions.

METHODOLOGY: Polymerase Chain Reaction (PCR) and Sanger Sequencing
(PCR is utilized pursuant to a license agreement with Roche Molecular Systems, Inc.)

REFERENCE VALUES: Mutation(s) present or absent as compared to a reference sequence of the normal TP53 gene

SPECIMEN REQUIREMENTS:
The following information is required:

1. Pertinent clinical history
2. Clinical or morphologic suspicion
3. Date of collection
4. Specimen source

Forms: Molecular Hematopathology Patient Information: B-Cell Chronic Lymphocytic Leukemia (CLL) for IGVH and/or TP53 Somatic Mutation Testing.

Blood and bone marrow specimens must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Specimen Type: Blood (preferred)
Container/Tube: Lavender top (EDTA), yellow top (ACD solution B), or green top (heparin)
Specimen Volume: 10 mL
Minimum Volume: 5 mL
Collection Instructions:

1. Invert several times to mix blood.
2. Send specimen in original tube.
3. Label specimen as blood.

Specimen Stability Information: Ambient <96 hours

Specimen Type: Bone marrow
Container/Tube: Lavender top (EDTA), yellow top (ACD solution B), or green top (heparin)
Specimen Volume: 3 mL
Minimum Volume: 2 mL
Collection Instructions:

1. Invert several times to mix bone marrow.
2. Send specimen in original tube.
3. Label specimen as bone marrow.

Specimen Stability Information: Ambient <96 hours

Specimen Type: Tissue
Container/Tube: Plastic container
Specimen Volume: 100 mg
Collection Instructions: Freeze immediately after collection.
Specimen Stability Information: Frozen

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	4 days
	Refrigerated	4 days

CAUTIONS:

- This test will not detect all possible acquired mutations in the TP53 gene, because it is restricted to analyzing exons 4-9. However, this region encompasses >90% of described pathologic mutations and covers the coding exons of the critical DNA binding regions.

- The analytical sensitivity of the assay can be affected by the absolute B-cell number in the peripheral blood or tissue sample, as well as the often subclonal nature of this tumor genetic abnormality. The assay attempts to compensate in part for this by performing an initial screening flow cytometry to assess B-cell quantity and by performing the cell enrichment step (for the peripheral blood specimens only) to isolate relatively pure CD19+ B-cells for analysis. Nevertheless, the nature of the Sanger sequencing method is such that typical reproducible analytic sensitivity will be in the order of 25% mutant allele burden.

- Because optimal cell enrichment is dependent on the absolute B-cell quantity, samples with very low WBC and/or initial %B cells (determined from flow cytometry or WBC automated cell count) will likely result in poor assay performance and inability to detect possible TP53 gene mutations in the tumor population.

CPT CODE: 81405

DAY(S) SET UP: Monday, Wednesday **ANALYTIC TIME**: 7 days