University of Nebraska Medical Center
REGIONAL PATHOLOGY SERVICES

LAB ALERT

Date: October 16, 2018

New Tests Update: CEBPA, KIT for AML Patients and TP53 Mutation Analysis by Next Generation Sequencing (NGS)

Dear Regional Pathology Services Clients,
We are very excited to announce that we are expanding our Next Generation Sequencing (NGS) testing menu to include CEBPA, KIT mutation for AML patients, and TP53 mutation analysis.

I. CEBPA Mutation Analysis

CEBPA mutations are observed in 10-20% of patients diagnosed with AML. CEBPA biallelic mutations (as opposed to monoallelic) are now associated with a favorable prognosis in AML (in the absence of FLT3-ITD) and biallelic mutation is required for recognition as the separate entity under the 2017 World Health Organization (WHO) classification "AML with Biallelic Mutation of CEBPA" (Swerdlow, S. et al 2017, Revised 4th Edition). The limit of detection for this assay is approximately 5% mutant allele, but may vary depending on the specific mutation.

Test Name: CEBPA MUTATION DETECTION/ BONE MARROW or CEBPA MUTATION DETECTION/ BLOOD
Test Code: CEBPA
CPT Code: 81218

II. KIT Mutation Analysis for Acute Myelogenous Leukemia (AML)

KIT is mutated in 8.0% of AML (COSMIC). Oncogenic KIT mutations occur primarily in core binding factor (CBF) AML, most often in exon 17 (50-60% in codon 816). KIT mutations in CBF-AML may have prognostic significance (Chen, W. et al. PLoS One. 2016; 11(1): e0146614; Ayatollahi, H. et al Hematol Oncol Stem Cell Ther. 2017 Mar;10(1):1-7). The limit of detection for this assay is approximately 5% mutant allele, but may vary depending on the specific mutation. We offer analysis for somatic mutations in the KIT gene for AML (covers hotspots in exons 1, 2, 8-11, 13, 17).
Test Name: KIT MUTATION PANEL
Test Code: KITAML
CPT Code: 81272

III. TP53 Mutation Analysis by Next Generation Sequencing

Aberrant p53 function, due to TP53 mutation, is associated with poor prognosis in chronic lymphocytic leukemia (CLL). Mutations in TP53 occur in about 5-10 % of untreated CLL patients. The presence of a TP53 mutation in CLL can indicate resistance to chemotherapy and may aid in determining the need for alternative targeted agents (Malcikova, J. et al. Leukemia 2018 32:1070-1080).
The limit of detection for this assay is approximately 5% mutant allele, but may vary depending on the specific mutation.

Test Name: TP53 MUTATION DETECTION / BONE MARROW or TP53 MUTATION DETECTION/ BLOOD
Test Code: TP53
CPT Code: 81405

Please specify on the requisition the surgical pathology case number and/or date of collection of the specimen for identification of the specimen to be tested, especially if previously extracted by our laboratory.

Acceptable Specimens:
Bone Marrow Aspirate, EDTA anticoagulated, minimum 1.0 mL (including Ficoll Gradient Flow Cells or unstained smears)

Peripheral Blood, EDTA anticoagulated, minimum 2.0 mL (including Ficoll Gradient Flow Cells)

Unacceptable specimens: Blood/Bone Marrow collected in heparin (green-top) tubes and Formalin-Fixed, Paraffin Embedded Tissue

Testing will be performed M-F; turnaround time is 7-10 business days. Inquires may be called to the laboratory at 559-7745, Jill Branson, Mgr, 559-7611, or Allison Cushman-Vokoun, Medical Director 559-3512.

The Molecular Diagnostics lab in addition performs: Melanoma Mutation Panel, GIST Panel, IDH1/IDH2 Mutation testing, KITD816V Mutation Testing, a 50 Gene Cancer Panel, Myeloid Panel, MPN Panel for Myeloproliferative Disorders, JAK2 extended testing for Polycthemia Vera and a 5 Gene Colorectal Panel. More information can be found on our test directory at www.reglab.org.

If you have any questions about this lab alert please contact client services at 402-559-6420 and ask to speak with one of the client coordinators.
