BRAF somatic mutation testing – hematologic malignancies

The UNC Molecular Genetics Laboratory performs a targeted *BRAF* sequencing assay using next-generation sequencing to identify mutation that may aid in diagnosis and treatment of hematologic neoplasms

Rationale for testing:
The presence of somatic gene mutations in *BRAF*, specifically in codon 600, may assist in distinguishing hairy cell leukemia from other B-cell neoplasms.

Nearly 100% of cases of hairy cell leukemia have a *BRAF* codon 600 (V600E) mutation in exon 15, while other splenic B-cell neoplasms are usually negative. The presence of a *BRAF* V600E mutation can aid in the diagnosis of hairy cell leukemia.

Specimen Requirements for the Myeloid Mutation Panel:
Bone marrow aspirate (1 mL, EDTA), peripheral blood (3mL, EDTA), or formalin-fixed, paraffin-embedded bone marrow clot sections (10 unstained slides, minimum area of sampled marrow = $4mm^2$) having at least 20% neoplastic cells. The assay is sensitive to variants above 5% allele frequency (10% clonal cells). This test is NOT appropriate for MRD monitoring. For patients undergoing repeat testing, previously detected variants will be reported to 3% VAF in fresh samples (5% in FFPE samples).

Gene Regions Tested – *BRAF* exon 15

Limitations:
Gene amplifications, translocations, and insertions or deletions over 90 bases in length are not reliably detected by this assay. Normal tissue is not tested to determine whether a gene variant is somatic (acquired) or germline (heritable). If the patient has evidence of a heritable cancer syndrome (e.g. different tumor types, early age of onset, family history), genetic counseling is recommended. To make a patient appointment, call the Cancer Genetics Clinic at (919) 843-8724.

References:

1. National Comprehensive Cancer Network (NCCN) Clinical Practice Guidelines in Oncology: Hairy cell leukemia, www.nccn.org
2. Blombery PA, Wong SQ, Hewitt CA, et al. Detection of BRAF mutations in patients with hairy cell leukemia and related lymphoproliferative disorders. *Haematologica*. 2012; 97(5):780-3. PMID: 22133769

Questions?

Call the Molecular Genetics Lab at (984) 974-1825 or Dr. Nathan Montgomery at 919-445-6414, E-mail Nathan.Montgomery@unchealth.unc.edu
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