**TP53 somatic mutation testing – hematologic malignancies**

The UNC Molecular Genetics Laboratory performs a targeted TP53 sequencing assay using next-generation sequencing to provide prognostic information in chronic lymphocytic leukemia (CLL) and other hematologic neoplasms, such as mantle cell lymphoma (MCL).

**Rationale for testing:**
Testing for the presence of somatic gene mutations may assist in refining prognosis and therapy selection for hematologic malignancies.

1) **CLL:** Mutations in TP53 are correlated with decreased survival and impact therapy selection.
2) **MCL:** Mutations in TP53 are correlated with outcomes and impact therapy selection.

**Specimen Requirements for the Myeloid Mutation Panel:**
Bone marrow aspirate (1 mL, EDTA) or peripheral blood (3mL, EDTA) having at least 20% neoplastic cells, and refrigerated for up to 72 hours. Unacceptable sample types include: fresh, frozen, or paraffin embedded tissue. 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.

**Gene Regions Tested** – All exons of TP53 are sequenced and analyzed

**Limitations:**
Gene amplifications, copy number changes, loss of heterozygosity, 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:**

**Questions?**
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