UNCH Molecular Testing for T-cell Large Granular Lymphocytic Leukemia

The UNCH Molecular Genetics Laboratory performs a sequencing panel targeting selected genes using next-generation sequencing to aid in the diagnosis of T-cell large granular lymphocytic leukemia.

Rationale for testing:

Testing for the presence of somatic mutations may assist in the diagnosis of T-cell large granular lymphocytic leukemia (T-LGLL). Mutations of *STAT3* and *STAT5B* can aid in distinction of reactive and neoplastic proliferations of T-cell large granular lymphocytes. Activating mutations in *STAT3* and *STAT5* are identified in 30-40% and 5% of T-LGLL, respectively, and can aid in the diagnosis.

Clinical indications for testing:

Expansion of T-cell large granular lymphocytes by flow cytometry and/or clinical concern for T-LGL Leukemia.

Specimen requirements:

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% T-cell large granular lymphocytes. The assay is sensitive to 5% variant allele frequency (VAF; 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% VAF in FFPE).

Limitations:
Gene amplifications, translocations, and insertions or deletions over 90 bases in length are not reliably detected by this assay. Variants predicted to be non-deleterious (such as synonymous coding changes and population variants) are not reported. Lack of mutation does not exclude neoplasia. Presence of clonality does not establish a diagnosis of malignancy.

Gene Regions Tested: These regions are covered by the T-cell LGL panel

Genes	RNA	Exons
STAT3	NM_139276.2	All
STAT5B	NM_012448.3	2-5,10-19

Questions?
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