

NPM1 Mutation Detection

Indication for Use: Nucleophosmin (NPM1) gene mutations, which cause aberrant cytoplasmic expression of nucleophosmin (NPMc+), are frequent genetic alterations in AML, being found in about 30% of cases. AML with mutated NPM1 usually carries normal karyotype. The NPM1 gene codes for a nucleoplasmic shuttling protein that is involved in many cellular functions. The clinical impact of NPM1 mutations is affected by the mutational status of the FLT3 gene. It has been proposed that combining the status of these two mutations allows for stratification into different prognostic groups, with NPM1 carrying mutants generally conferring a more favorable prognosis.

Testing Method: Amplicon based targeted next generation sequencing. NPM1 mutation detection can be ordered as an individual test or as part of De Novo AML panel.

Test Parameters:

Diagnostic sensitivity: This assay is designed to detect known single nucleotide variants only within defined regions. Nucleotide insertions and deletions more than 25bp or outside of the defined regions may not be detected. Gene rearrangements are not detected.

Technical sensitivity: This assay may not detect certain mutations if the proportion of tumor cells in the sample studied is less than 5%.

Turn Around Time: 5-7 business days

Sample Requirements:

- 3 ml peripheral blood in lavender top tube (EDTA)
- Bone marrow aspirate (anticoagulated with either heparin or EDTA and, if possible, placed into tissue culture medium)

CPT Codes: 81310