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NPM1 Nucleophosmin Gene Mutation

Mutation Detection in Acute Myeloid Leukemia (AML)

Indications for testing

- Suspected diagnosis Acute Myeloid Leukemia (AML) Nucleophosmin-1 gene mutations within exon 12 (*NPM1* mutations) are among the most frequent molecular aberration detected in patients with acute myeloid leukemia (AML). The overall frequency of *NPM1* mutations is 20-30% of all AML subtypes and in ~50% of AML with normal cytogenetic findings. In the absence of other, deleterious mutations, *NPM1* mutations are associated with a better prognosis

Testing Methodology

NPM1 insertion mutation detection involves polymerase chain reaction (PCR) amplification of exon 12 followed by capillary electrophoresis separation to detect the DNA fragments of increased size.

Interpretation of DNA analysis

The presence of a somatic 4-11 base pair insertion in the exon 12 of the *NPM1* gene is consistent with a better prognosis (increase in disease-free and overall survival) in patients with AML, in the absence of additional deleterious somatic mutations or cytogenetic abnormalities. . For example, patients with cytogenetically normal AML tumors that lack an *NPM1* mutation but possess a *FLT3* internal tandem duplication mutation (ITD) have a worse prognosis, as gauged by both disease-free and overall survival, than do patients whose tumors possess an *NPM1* mutation without a concomitant *FLT3*-ITD.

Specimen Requirements

Peripheral blood--1 lavender-top (EDTA) tube. Invert several times to mix blood. Forward promptly at ambient temperature.
Bone Marrow--Place 1-2 mL of anticoagulated bone marrow in a lavender-top (EDTA) tube. Invert several times to mix bone marrow. Forward promptly at ambient temperature.
Frozen Tissue--10 mm^3 of fresh frozen tissue in sterile, plastic container. Forward frozen tissue on dry ice.

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425 South Euclid Avenue, Room 5970
St. Louis, MO 63110

Clinical information must be provided with specimen referral in order to correctly interpret test results.

Current Pricing

Contact Lab Customer Service for current pricing 314 362-1470.
CPT code: 81310

1. Falini B, Mecucci C, Tiacci E, et al. “Cytoplasmic nucleophosmin in acute myelogenous leukemia with a normal karyotype”. New Engl J Med 2005; 352:254-266.
2. Thiede C, Koch S, Creutzig E, et al. “Prevalence and prognostic impact of NPM1 mutations in 1485 adult patients with acute myeloid leukemia (AML)” Blood 2006; 107:4011-4020.
3. Falini B, Nicoletti I, Martelli MF, et al. “Acute myeloid leukemia carrying cytoplasmic/mutated nucleophosmin: biological and clinical features”. Blood 2007; 109:874-885.
4. Dohner K, Schlenk RF, Habdank M, et al. “Mutant nucleophosmin (NPM1) predicts favorable prognosis in younger adults with acute myeloid leukemia and normal cytogenetics: interaction with other gene mutations” Blood 2005; 106:3740-3746.