LabPMM™

NPM1

NPM1 Mutation Assay

Indications for Testing

- Stratifying high and low risk AML
- At initial diagnosis of AML
- Reoccurrence of leukemia after induction therapy on patients not initially screened for NPM1mutations.

Test Code

NPM1

Specimen Requirements

5 ml of Peripheral Blood in Heparin, EDTA or ACD
3 ml of bone marrow in Heparin, EDTA or ACD
1 µg of previously isolated DNA

Specimen Transport

Ambient or cool
Do no Freeze

CPT Codes

81310

Turn-Around-Time

3-7 Days

Description of Testing

Primers targeting the area surrounding exon 12 of the NPM1 gene are used to amplify the patient's DNA. The size of the NPM1 PCR product is determined by capillary electrophoresis.

Clinical Utility of *NPM1* Mutation Assay

The Nucleophosmin (NPM1) gene is one of the most commonly mutated genes in acute myeloid leukemia (AML), occurring in about 35% of AML patients at diagnosis and in approximately 60% of adult cytogenetically normal AML (CN-AML). The CN-AML group is collectively the most common AML group and assigned an intermediate prognostic category. However studies have shown that the CN-AML group is not homogenous and significant diversity has been shown to exist within this population of AML patients. The vast majority of NPM1 mutations are insertions in exon 12 occurring near the C-terminus of the protein resulting in cytoplasmic localization. Currently there are over 40 known NPM1 mutations, most of which will be detected with our assay.

Clinical studies have found that NPM1 mutations are associated with increased blast counts, higher extramedullary involvement and increased platelet counts in AML. NPM1 mutations, in the absence of the FLT3 mutation, are associated with a good response to induction therapy and have a 60% improved 5 year survival. It has been suggested that the identification of mutations in both NPM1 and FLT3 genes allows for the stratification of the CN-AML patients into three different prognostic groups. A favorable prognosis is associated with NPM1+ and FLT3-, an intermediate prognosis with NPM1+ and FLT3+ or NPM1- and FLT3-, and a poor prognosis with NPM1- and FLT3+.

It is recommended that patients who are CN-AML should be screened for NPM1 mutations in efforts to assess prognosis and aid in treatment decisions. Utilizing both NPM1 and FLT3 mutations status is the most common method in stratification of the CN-AML population.

References

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* *NPM*1 testing is covered by a United States patent licensed from TrovaGene, Inc.