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NPM1 Insertion Mutation DNA Screen and *FLT3 ITD*

Laboratory Medicine Home Page

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We are pleased to announce that starting November 1, 2007 the Molecular Hematopathology Laboratory will offer PCR assays for two mutations that are associated with prognosis in acute myeloid leukemia (AML). The *FLT3* internal tandem duplication (ITD) assay detects internal duplication mutations in the juxtamembrane region of the *FLT3* gene, resulting in activation of the FLT3 tyrosine kinase protein. The *NPM1* assay detects insertion mutations in the *NPM1* gene, resulting in altered cellular localization of the nucleophosmin 1 protein. *FLT3* ITD mutations have been associated with adverse prognosis in AML. In patients who have AML with a normal karyotype, the presence of a *NPM1* mutation without a *FLT3* ITD mutation has been associated with more favorable prognosis. These tests are intended to be used at the time of diagnosis when significant numbers of blasts are present; they are not designed to detect low-level involvement by AML following therapy. The tests can be performed on peripheral blood or bone marrow. Results for the *FLT3* and *NPM1* tests will be available within one week.

References:

Murphy, Kathleen M., et al (2003). Detection of FLT3 Internal Tandem Duplication and D835 Mutations by a Multiplex Polymerase Chain Reaction and Capillary Electrophoresis Assay, Journal of Molecular Diagnostics, 5(2): 96-102.

Falini B, Mecucci C, et al. (2005). Cytoplasmic Nucleophosmin in Acute Myelogenous Leukemia with a Normal Karyotype. N Engl J Med 352(3): 254-266.