



Division of Human Genetics

NPM1 Mutation Assay (NPM1)

Molecular Genetics Laboratory

CLIA#: 36D0965791

Phone: (513) 636-4474

Fax: (513) 636-4373

Email: moleculargenetics@cchmc.org

Additional information and test requisitions are available at:

www.cincinnatichildrens.org/molecular-genetics



Shipping Instructions

Please enclose an oncology test requisition with sample. All information must be completed before sample can be processed.

Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday

Ship to:
Cytogenetics and Molecular Genetics
Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229

The NPM1 gene is a nucleocytoplasmic shuttling protein that is involved in many cellular functions. If there is a mutation in the NPM1 gene, this can disrupt those functions. A common mutation is known to be present in exon 12 of the NPM1 gene, and involves 4bp insertions into this region of the gene. However, the NPM1 mutation leads to a more FAVORABLE prognosis in patients with Acute Myeloid Leukemia (AML) when their cancer is also found to also have a normal karyotype.

INDICATION

The main indication for NPM1 testing at this time is at the time of diagnosis in a patient with Acute Myeloid Leukemia. If the patient has AML and testing has revealed a normal karyotype (about 50% of AML cases) then the prognosis is often considered intermediate or mixed. NPM1 testing is indicated and can give more information about the patient's prognosis. In those patients with an NPM1 mutation and a normal karyotype, a more FAVORABLE prognosis is suspected. Other genetic markers can also affect prognosis. Mutations in the FLT3 gene have also been shown to affect prognosis in patients with AML and normal karyotype. Consideration of testing for mutations in this gene may provide a more complete prognostic picture.

METHODOLOGY

A single polymerase chain reaction (PCR) amplifies the region of the gene that contains exon 12. The PCR reaction is followed by capillary gel electrophoresis to identify the presence of 4 bp NPM1 insertion mutation based on fragment size analysis. This test utilizes the Applied Biosystems 3130xl system. It has not been cleared or approved by the United States Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical laboratory testing.

SPECIMEN: 5-10mls peripheral blood OR 3-5mls bone marrow in EDTA tube

TURN-AROUND TIME: 7-10 days

COST: Please contact laboratory for pricing

CPT CODES: 83890, 83900 x8, 83909 x8, 83912

REFERENCES:

Falini, B., C. Mecucci, et al. (2005). "Cytoplasmic nucleophosmin in acute myelogenous leukemia with a normal karyotype." *N Engl J Med* 352(3): 254-66.

Schneider F., et al. (2009), "NPM1, but not FLT3-ITD mutations predict early blast cell clearance and CR rate in patients with normal karyotype AML (NK-AML) or high risk myelodysplastic syndrome (MDS)." *Blood*, online March 11, 2009.

Thiede, C., S. Koch, et al. (2006). "Prevalence and prognostic impact of NPM1 mutations in 1485 adult patients with acute myeloid leukemia (AML)." *Blood* 107(10): 4011-20.