NPM1 Gene Mutation Analysis

**Test Name:** NPM1  
**Test Code:** NPM1  
**CPT Code:** 81310  

**Clinical Use:**  
The nucleophosmin (NPM1) gene is located on chromosome 5q35 and encodes for a protein which shuttles between nucleus and cytoplasm. Approximately 50% of acute myeloid leukemia cases with normal cytogenetics, show positive NPM1 mutation\(^1\). The most commonly seen mutations in the gene encoding NPM1 are seen in exon 11 (>97%), although other rare mutations have been reported\(^2\). This frameshift mutation results in retention of the protein in the cytoplasm. In the absence of FLT3 mutation, the presence of NPM1 mutation is associated with a more favorable prognosis.

**Methodology:**  
This Sanger-based sequencing assay analyzes the exon 11 (formerly known as exon 12) region of NPM1 gene, where all common mutations are reported to occur.

**Reference Range:** NA  

**Specimen Requirements:**  
- 5-10 mL of blood or bone marrow-purple (EDTA) tube or yellow (ACD) tube within 24 hours of collection.  
- DNA 10ug at a minimum of 100ng/µL.

Note: If sample cannot arrive at laboratory within 24 hours of draw, refrigerate until sample can be transported and then transfer it at room temperature.

**Special Handling:** NA  

**Test Performance Schedule:** Weekly  

**Turn Around Time:** 7-10 days  

**STAT Availability:** NA
REFERENCES


