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

1. Mrozek K, Marcucci G, Paschka P, et al. "Clinical relevance of mutations and gene-expression changes in adult acute myeloid leukemia with normal cytogenetics: are we ready for a prognostically prioritized molecular classification?" *Blood* 2007, 109(2): 431-448.
2. Schnittger S, Schoch C, Kern W, et al. "Nucleophosmin gene mutations are predictors of favorable prognosis in acute myelogenous leukemia with a normal karyotype." *Blood* 2005, 106(12): 3733-3739.
3. Thiede C, Koch S, Creutzig E, et al. "Prevalence and prognostic impact of NPM1 mutations in 1485 adult patients with acute myeloid leukemia." *Blood* 2006, 107(10): 4011-4020.
4. Falini B, Mecucci C, Tiacci E, et al. Cytoplasmic nucleophosmin in acute myelogenous leukemia with a normal karyotype. N Engl J Med. 2005; 352: 254-266.