ARUP Laboratories
500 Chipeta Way – Salt Lake City, UT 84108
(800)522-2787 - www.aruplab.com
Julio C. Delgado, M.D. M.S., Director of Laboratories

*****Example Report*****

Patient Age/Gender: Unknown Unknown
Printed: 22-Sep-17 07:38:21

Procedure	Result	Units	Ref Interval	Accession	Collected	Received	Reported/ Verified
NPM1 Quantitative, Source	Whole Blood			17-265-900010	22-Sep-17 06:47:00	22-Sep-17 06:47:00	22-Sep-17 07:25:13
NPM1 Quantitative, Result	**Detected *f**			17-265-900010	22-Sep-17 06:47:00	22-Sep-17 06:47:00	22-Sep-17 07:25:13
NPM1 Quantitative, Ratio	4.6500			17-265-900010	22-Sep-17 06:47:00	22-Sep-17 06:47:00	22-Sep-17 07:25:13

22-Sep-17 06:47:00 NPM1 Quantitative, Result:

A NPM1 mutation was detected.

This result has been reviewed and approved by Jay Patel, M.D.

22-Sep-17 06:47:00 NPM1 Quantitative, Result:
INTERPRETIVE INFORMATION: NPM1 Mutation Detection
by RT-PCR, Quantitative

This test is designed to detect and quantify NPM1 mutant transcripts. NPM1 mutations represent a common recurrent genetic abnormality found in a subset of patients with acute myeloid leukemia (AML). Approximately one-third of AML patients overall and one-half of cytogenetically normal AML patients harbor an NPM1 mutation, with the most common form being a TCTG insertion (type A) seen in approximately 80% of NPM1-mutated cases. Rarer forms, known as types B and D, compose around 10% of NPM1-mutated cases. This test is designed to detect and quantify NPM1-mutant transcripts of types A, B, and D only. Mutated NPM1 confers a favorable prognosis in cytogenetically normal AML patients who lack FLT3 internal tandem duplication mutations. Recent studies show that minimal residual disease (MRD) monitoring of AML patients after chemotherapy provides important prognostic information independent of other risk factors and may help to inform clinical decisions (see reference).
METHODOLOGY: Patient RNA is isolated, reverse transcribed into cDNA, and amplified using multiplex allele-specific primers targeting types A, B, and D NPM1 variants. A fragment of the ABL1 gene is co-amplified and quantification is performed using the delta-delta Ct method relative to a plasmid calibrator that harbors a 1:1 ratio of NPM1 type A and ABL1 cDNA fragments. Results are reported as a normalized ratio of NPM1 variant transcripts to ABL1 transcripts present in the sample.
LIMITATIONS: Rare NPM1 variants (non-type A, B, or D) may not be detected. The limit of detection for this assay is 1:100,000 cells (0.001%) for type A NPM1 mutants based on cell line dilution experiments. The sensitivity of this assay for type B and D mutants is expected to be similar but has not been demonstrated.
Results of this test must always be interpreted within the patient's clinical context and in conjunction with other relevant data, and should not be used alone for a diagnosis of malignancy. A negative result does not definitely exclude the possibility of an NPM1 mutation below the detection limit of this test and does not exclude the possibility of rare forms of NPM1 mutant transcripts (non-type A, B, or D) not detectable by this methodology.

For ongoing monitoring after initial diagnosis, this test should only be used in patients who are known to have NPM1-mutated AML.

Reference:

Ivey A et al. Assessment of Minimal Residual Disease in Standard-Risk AML.
N Engl J Med. 2016 Feb 4;374(5):422-33.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement B: aruplab.com/CS