



NPM1 Mutation Analysis, V

PATIENT NAME TESTINGRNA, VALIDATION			ORDER NUMBER J413000253
PATIENT ID SA00129082	DATE OF BIRTH 09/09/1999	SEX Male	REQUESTED BY CLIENT CLIENT
COLLECTED 5/12/2020, 12:00 AM	RECEIVED 5/13/2020, 11:34 AM	REPORTED 5/13/2020, 12:30 PM	
7028846 DLMP Rochester Rochester MN 55901			CLIENT ORDER NUMBER SA00129082
			CLIENT MRN SA00129082
Specimen Type	Peripheral blood		
Signing Pathologist	BENJAMIN BAILEY		

INTERPRETATION

Peripheral blood, NPM1 gene Mutation analysis:

Positive for NPM1 mutation. The quantitative level of mutated NPM1 transcript is 2735/10,000 ABL1 copies (27.35%). See comments.

Comments: NPM1 mutation is seen in approximately 30% of acute myeloid leukemia (AML) patients. It is prevalent in normal karyotype AML and frequently associated with FLT3 gene mutations. AML patients carrying the NPM1 mutation in the absence of FLT3-ITD have been shown to have better overall and disease free survival, and lower cumulative incidence of relapse (Thiede et al, 2006, 16455956; Suzuki et al, 2005, 15994285; Falini et al, 2005; 15659725).

In NPM1-mutated AML patients, persistence of NPM1-mutated transcript, particularly at higher levels (>100-200/10,000 ABL1 copies) after at least two cycles of cytotoxic chemotherapy is associated with increased risk of relapse and lower rate of survival (Kronke et al, 2011, 21555683; Shayegi et al, 2013, 23656730; Hills et al, 2016, 26789727). Trends in the level of NPM1-mutated transcript should be followed carefully. An increase of NPM1-mutated transcript level greater than or equal to 1 log10 between two positive samples is consistent with molecular relapse/molecular progression (Schuurhuis et al, 2018, 29330221). The reproducibility of this assay is such that results within 0.5 log10 should be considered equivalent. Suggest correlation with historical NPM1 test results, as well as clinical and other laboratory findings.

METHOD

The assay starts with a sensitive quantitative reverse transcription real-time polymerase chain reaction (RT-PCR) which detects and quantifies the most common mutant NPM1 transcripts (A, B, D forms, account for approximately 90% of NPM1 mutations in acute myeloid leukemia (AML)). If a mutant NPM1 transcript is not detected, the test reflexes to a qualitative NPM1 exon 12 mutation screen that detects essentially all mutant forms reported in AML, including the rare non-A, B, D forms (with lower sensitivity). For the quantitative test, extracted RNA is reverse transcribed into cDNA and quantitative PCR is then performed. The quantitative value of NPM1 transcript copy number is determined relative to ABL1 as the reference transcript. The analytic sensitivity of this quantitative assay is approximately 1/10,000 (mutated NPM1/ABL1 transcript copies, 0.01%). The quantitative test will not detect other rarer NPM1 mutation types. The qualitative PCR assay is performed on extracted DNA using primers that amplify a fragment of NPM1 DNA containing the region susceptible to insertion mutation and detects essentially all types of NPM1 mutations (insertion/deletion) described in acute myeloid leukemia. The amplified fragments are size separated by capillary electrophoresis. The analytical sensitivity of the qualitative test is approximately 5% (mutated/wild-type NPM1 DNA).

DISCLAIMER

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.



1-800-533-1710

NPM1Q

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Code : MCR Laboratory : Mayo Clinic Laboratories - Rochester Main Campus

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The collected, received, and reported dates and times on the report are in the time zone of the performing location.