

Myeloproliferative Neoplasm, JAK2 V617F with
Reflex to CALR and MPL, Varies

Patient ID SA00100563	Patient Name TESTINGRNV, REPORT	Birth Date 1999-11-11	Gender M	Age 17
Order Number SA00100563	Client Order Number SA00100563	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 08 Nov 2017 06:00		

MPN (JAK2 V617F, CALR, MPL) Reflex

MPNR Result

see interpretation

MCR

Final Diagnosis

1 MCR

Peripheral blood, CALR mutation analysis, exon 9:

Positive. A 5bp insertion-type mutation was detected in CALR, exon 9.

Comment: CALR mutation is identified in approximately 49–88% of JAK2 and MPL-wild type essential thrombocythemia (ET) and primary myelofibrosis (PMF) patients, and at significantly lower frequencies in other myeloid neoplasms such as refractory anemia with ringed sideroblasts associated with marked thrombocytosis (13%), myelodysplastic syndrome (8%), chronic myelomonocytic leukemia (3%) or atypical chronic myeloid leukemia (3%). Correlation with clinical and pathologic findings is required for a definitive diagnosis.

No further testing for MPL mutation was performed as it is not indicated per MPNR test algorithm. See test catalogue for additional information: <http://www.mayocliniclabs.com/test-catalog/Overview/63031>.

Method Summary - CALR: Exon 9 of CALR was amplified from genomic DNA by polymerase chain reaction (PCR). The size of the PCR product was analyzed by capillary electrophoresis on the ABI 3130xl Genetic Analyzer. The analytic sensitivity of this assay is approximately 6% for the majority of CALR mutations and is approximately 20% for the rare type 1-bp deletion (See Mayo Medical Laboratories Interpretive Handbook for details).

Peripheral blood, JAK2 V617F mutation analysis:

Negative for JAK2 V617F.

Method summary - JAK2 V617F analysis: Quantitative, allele-specific polymerase chain reaction (PCR) assay was performed using extracted genomic DNA to evaluate for the point mutation causing JAK2 V617F. The analytic sensitivity of this assay has been determined at 0.06% (see Mayo Medical Laboratories Interpretive Handbook for method details).

Signing Pathologist: Melissa Tricker-Klar

Received: 08 Nov 2017 16:15

Reported: 08 Nov 2017 17:19

Laboratory Notes

- 1 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.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292