

Patient ID SA00100565	Patient Name TESTINGRNV, REPORT	Birth Date 1999-11-11	Gender M	Age 17
Order Number SA00100565	Client Order Number SA00100565	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 08 Nov 2017 06:00		

MPN (CALR, MPL) Reflex

MPNCM Reflex Result

see interpretation

Specimen Type

Peripheral blood

Final Diagnosis

Peripheral blood, MPL exon 10 mutation analysis:

Positive. The following mutation was detected in MPL:
c.1544G>T

This translates into the following predicted protein
change: p.Trp515Leu

Comment:

MPL exon 10 somatic mutations are seen in a subset of myeloproliferative neoplasms (MPN), with a frequency of approximately 5–10% in primary myelofibrosis (PMF) and 3–5% in essential thrombocythemia (ET). This mutation is considered pathogenic. The most common MPL mutations involve W515 or S505 at the protein level (Pikman et al, 2006, 16834459; Beer et al, 2008, 18451306; Onishi et al, 1996, 8695859; Staerk et al, 2006, 16249382). Rare cases of familial ET with S505N mutation

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have been reported (Ding et al, 2004, 14764528). Clinicopathologic correlation is recommended for a definitive diagnosis.

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Method summary - MPL exon 10 mutation analysis: Genomic DNA was extracted and Sanger sequencing used to evaluate for mutations in MPL, exon 10 (see Mayo Medical Laboratories Interpretive Handbook for method details). The sensitivity of this assay is approximately 20%, such that samples containing lower percentages of mutated DNA will appear negative.

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Peripheral blood, CALR mutation analysis, exon 9:

Negative. No deletion or insertion was detected in CALR, exon 9.

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).

Signing Pathologist: Melissa Tricker-Klar

Received: 08 Nov 2017 16:15

Reported: 08 Nov 2017 17:22

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