

Patient ID SA00123354	Patient Name VALIDATIONSOF, TESTINGMO	Birth Date 1993-10-18	Gender M	Age 25
Order Number SA00123354	Client Order Number SA00123354	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 07 Oct 2019 14:05		

MPL Exon 10 Mutation Detection, V

Specimen Type

Bone marrow

MCR

Interpretation

Bone marrow, MPL exon 10 mutation analysis:

1 MCR

Positive. The following genetic alteration was detected in MPL :
Chr1(GRCh37): g.43815008T>A; NM_005373.2: c.1543T>A
This translates into the following predicted protein change:
pTrp515Arg (W515R)

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). The most common pathogenic MPL genetic alterations 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 have been reported (Ding et al, 2004, 14764528). Clinicopathologic correlation is recommended for a definitive diagnosis.

ADDITIONAL INFORMATION

Method Summary: Genomic DNA is extracted and PCR followed by Sanger sequencing is performed to evaluate for mutations in MPL, exon 10. The sensitivity of this assay is approximately 20%, such that samples containing lower percentages of mutated DNA may not be detected. The reference transcript used for variant annotation is: GRCh37(hg19)NM_005373.2.

Signing Pathologist

COLE ASPROS

MCR

Received: 08 Oct 2019 14:10

Reported: 08 Oct 2019 14:20

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