MAYO CLINIC LABORATORIES

1-800-533-1710

JAKXB

JAK2 Exon 12 and Other Non-V617F Mutation Detection, Blood

Patient ID	Patient Name		Birth Date	Gender	Age
SA00100559	TESTINGRNV, REPORT		1999-11-11	M	17
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00100559	SA00100559	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		08 Nov 2017 06:00			

JAK2 Exon 12 Mutation Detection, B

JAK2 Sequencing Result

MCR

see interpretation

Final Diagnosis:

[1] MCR

Peripheral blood, JAK2 exons 12–15 genetic alteration analysis:

Positive. The following genetic alteration was detected in JAK2 : g.5072770G>T; c.1849G>T

This translates into the following predicted protein change: p.Val617Phe (JAK2 V617F)

JAK2 V617F alteration is found with high frequency in Philadelphia chromosome-negative myeloproliferative neoplasms (>95% in polycythemia vera and approximately 50–60% in primary myelofibrosis and essential thrombocythemia). It can also occur in other myeloid neoplasms, such as refractory anemia with ring sideroblasts associated with marked thrombocytosis (RARS-T) and rarely in myelodysplastic syndrome, chronic myelomonocytic leukemia, atypical chronic myeloid leukemia, and acute myeloid leukemia (Baxter et al, 2005, 15781101; Kralovics et al, 2005, 15858187; James et al, 2005, 15793561; Malcovati et al, 2009; Steensma et al, 2005, 15860661; Wang et al, 2014, 24627528). Clinicopathologic correlation is recommended for a definitive diagnosis.

Signing Pathologist: Melissa Tricker-Klar

ADDITIONAL INFORMATION

Method summary - JAK2 exons 12–15 mutation analysis: Total RNA was extracted and converted to cDNA, followed by Sanger sequencing of JAK2, exons 12–15 (see Mayo Clinic Laboratories Interpretive Handbook for method details). The sensitivity of this assay is approximately 20%, such that samples containing lower percentages of mutated nucleic acid will appear negative. The reference gene transcript used for variant annotation is: GRCh37(hg19)NM_004972.3.

Received: 08 Nov 2017 16:14 **Reported:** 08 Nov 2017 17:17

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