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Specimen Collected: 13-Aug-25 10:55

JAK2 Exon 12-Mutation Analysis by |Received: 13-Aug-25 10:55

Report/Verified: 13-Aug-25 13:02

PCR

Procedure	Result	Units	Reference Interval
JAK2EX12, Source	Bone Marrow		
JAK2EX12, Interpretation	Positive * f1 i1		

**Result Footnote**

f1: JAK2EX12, Interpretation

There is evidence of a JAK2 mutation in Exon 12.

This result has been reviewed and approved by [REDACTED]

**Test Information**

i1: JAK2EX12, Interpretation

INTERPRETIVE INFORMATION: JAK2 Exon 12-Mutation Analysis  
by PCR

DNA from whole blood or bone marrow is isolated and subjected to PCR amplification in the presence of a short blocking oligonucleotide homologous to codons 537-544 of exon 12 of the wild-type JAK2 gene. The oligonucleotide is designed to specifically suppress PCR amplification of wild-type JAK2 exon 12 sequence. In contrast, JAK2 exon 12 mutations located between codons 537-544 disrupt proper binding of the blocking oligonucleotide during PCR amplification resulting in a product of approximately 225 base-pairs. Each assay includes control DNA from mutation positive and wild-type negative samples; all samples are tested in paired reactions with and without blocking oligonucleotide. A PCR product formed in the presence of blocking oligonucleotide indicates the presence of a mutation.

Results of this test must always be interpreted in the context of clinical and other relevant laboratory data such as erythropoietin level, exclusion of other causes of elevated hemoglobin, and should not be used alone for a diagnosis of polycythemia vera which is a form of malignancy, i.e., myeloproliferative disorder. This test does not identify JAK2 mutation outside of codons 537-544, and duplications or missense variants that compromise oligonucleotide binding may not be detected.

Limit of Detection: 5 percent mutant alleles for length-altering mutations.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

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\*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

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**Unless otherwise indicated, testing performed at:****ARUP Laboratories**

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Jonathan R. Genzen, MD, PhD

**ARUP Accession:** 25-225-900152**Report Request ID:** 20842504**Printed:** 13-Aug-25 15:55

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