


Dr. Vinay Chopra
 MD (Pathology & Microbiology)
 Chairman & Consultant Pathologist

Dr. Yugam Chopra
 MD (Pathology)
 CEO & Consultant Pathologist

 220054000256658	Mr. GURMINDERJIT SINGH AMBALA, CHEMBUR Tel No : 9509477777 PIN No: 133001 PID NO: P542200278141 Age: 21.0 Year(s) Sex: Male	Reference: Dr.VINAY KUMAR CHOPRA Sample Collected At: Dr Vinay Kumar Chopra Kos Diagnostic Lab, 6349/i, Nicholson Road, Ambala Cantt, Hry 133001. Processing Location:- Metropolis Healthcare Ltd,unit No409-416,4th Floor,commercial Building-1,kohinoor Mall,mumbai-70	VID: 220054000256658 Registered On: 09/02/2023 04:24 PM Collected On: 09/02/2023 4:25PM Reported On: 17/02/2023 06:18 PM
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Test Name : JAK-2 Mutation (CMPD) Exon 12 Mutation

Specimen : Blood

Results

JAK2 EXON 12 MUTATION	Not Detected
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Comments :JAK2 V617F mutation analysis, if not performed already, is advised.

Method : PCR - Sequencing

Jak2 Exon 12 mutations are detected by a combination of PCR amplification and direct sequencing of coding and junctional region of the exon. The sequence is compared with the reference sequence NM_004972 for the analysis of mutations. Presence of point mutations, insertion, deletion, indels indicates a positive result. Absence of any mutation indicates a negative result.

The lower limit of detection of the assay is approximately 20%. The assay is expected to detect >99% of somatic variants that are present at an allelic burden of ~20% or greater.

Clinical Background:

- Somatic mutations in exon 12 of Jak2 Janus Kinase 2 gene (Jak2) are found in about 2-3% patients with polycythemia vera (PV).
- Jak2 Exon 12 mutations represent clonal markers in PV, and serve as a WHO major criterion for the diagnosis of this disorder. Mutations are predominantly clustered between codons 537-544.
- Exon 12 mutation status is associated with erythrocytosis and atypical bone marrow morphology.
- JAK2 Exon 12 Mutation Analysis may be used to differentiate reactive conditions from malignant erythrocytosis. JAK2 exon 12 mutations appear to be specific to polycythemia vera (PV) or idiopathic erythrocytosis.
- Patients who test negative for JAK2V617F mutation can be tested for JAK2 exon 12 mutation to classify further or rule out PV.

Note:

This test has been developed and its performance is validated at Metropolis Healthcare Ltd.

Limitation of the Assay:

Presence of PCR inhibitors in the sample may prevent DNA amplification. Paradoxical results may arise due selection of inappropriate specimens and contamination during specimen collection.

References:

- Lakey et al., 2010, Am J Clin Pathol 133: 942-948
- Scott et al., 2011, Am J of Hematology, Vol. 86:8, 668-676
- Bench et al., 2013, Br J Haematol. 2013 Jan;160(1):25-34



NOTE:
This Sample was outsourced



KOS Diagnostic Lab
(A Unit of KOS Healthcare)



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-- End of Report --



Tests marked with NABL symbol are accredited by NABL vide Certificate no MC-2139; Validity till 01-06-2024

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