JAK2 (V617F) Mutation by ddPCR

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Myeloproliferative neoplasms (MPNs) are a group of blood cancers that cause excess blood cell production in the bone marrow and often in the peripheral blood. Classic *BCR-ABL1*-negative MPNs include polycythemia vera (PV), essential thrombocythemia (ET), and primary myelofibrosis (PMF).[1] The majority of patients with PV, ET, and PMF have *JAK2*, *CALR*, or *MPL* gene variants; thus, genetic testing for *JAK2* is an important tool for the classification and diagnosis of these disorders.[1] A quantitative *JAK2* V617F assay can provide additional information about allele fraction, suggest hemizygosity/homozygosity, and be useful for monitoring ruxolitinib therapy.

For detailed information on the testing strategy for MPNs, including use of genetic testing for variants in the *CALR* and *MPL* genes, refer to the ARUP Consult Myeloproliferative Neoplasms topic.

Genetics

Variants Detected

JAK2 (V617F) Mutation by ddPCR: point mutation c.1849G>T (V617F) of the *JAK2* gene

Additional variants will be evaluated if reflex testing is ordered; refer to *JAK2* (V617F) Mutation by ddPCR, Qualitative with Reflex to *CALR* (Calreticulin) Exon 9 Mutation Analysis by PCR and *MPL* Mutation Detection (3016839) and *JAK2* (V617F) Mutation by ddPCR, Qualitative with Reflex to *JAK2* Exon 12 Mutation Analysis by PCR (3016840) on the ARUP Laboratory Test Directory.

Test Interpretation

Analytic Specificity

JAK2 (V617F) Mutation by ddPCR, Quantitative: >99%

JAK2 (V617F) Mutation by ddPCR, Qualitative: >99%

Limit of Detection

JAK2 (V617F) Mutation by ddPCR, Quantitative: 0.2%

JAK2 (V617F) Mutation by ddPCR, Qualitative: 0.5%

Limitations

JAK2 V617F Mutation by ddPCR Analysis

- Variants in genes other than *JAK2* are not detected.
- Variant alleles of *JAK2* other than V617F (c.1849G>T) are not reported.
- Samples with *JAK2* V617F variants below the limit of reporting may not be detected.
- Results of this test must always be interpreted in the context of morphologic and other relevant data and should not be used alone for a diagnosis of malignancy.

Featured ARUP Testing

JAK2 (V617F) Mutation by ddPCR, Quantitative 3003751

Method: Droplet Digital PCR (ddPCR)

- Aids in the assessment of suspected MPNs
- Use to quantify the *JAK2* V617F variant in peripheral blood or bone marrow

JAK2 (V617F) Mutation by ddPCR, Qualitative 3004046

Method: Droplet Digital PCR (ddPCR)

- Aids in the workup of suspected MPNs
- Use to detect the *JAK2* V617F mutation in peripheral blood or bone marrow

Other tests that include *JAK2* (V617F) mutation by ddPCR testing:

JAK2 (V617F) Mutation by ddPCR, Qualitative With Reflex to CALR (Calreticulin) Exon 9 Mutation Analysis by PCR and MPL Mutation Detection 3016839

Method: Droplet Digital PCR (ddPCR)/Capillary Electrophoresis

JAK2 (V617F) Mutation by ddPCR, Qualitative With Reflex to JAK2 Exon 12 Mutation Analysis by PCR 3016840

Method: Droplet Digital PCR (ddPCR)

Different limitations apply to reflex testing; refer to *JAK2* (V617F) Mutation by ddPCR, Qualitative with Reflex to *CALR* (Calreticulin) Exon 9 Mutation Analysis by PCR and *MPL* Mutation Detection (3016839) and *JAK2* (V617F) Mutation by ddPCR, Qualitative with Reflex to *JAK2* Exon 12 Mutation Analysis by PCR (3016840) on the ARUP Laboratory Test Directory.

References

1. National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology: Myeloproliferative neoplasms. Version 3.2023. Last update Oct 2023; accessed Oct 2023.

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