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). 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. 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.
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