JAK2 V617F MUTATION ANALYSIS

BACKGROUND:
The presence of the somatic V617F mutation (c.1849G>T) in the Janus Kinase 2 (JAK2) gene serves as a clonal marker and a WHO major criterion for diagnosis of myeloproliferative neoplasms including polycythemia vera (PV), essential thrombocythemia (ET), and primary myelofibrosis (PMF). The V617F variant is found in approximately 95% of patients with PV, and 50% of patients with ET and PMF. In addition JAK2 V617F is found in up to 60% of patients who fall within the provisional WHO category of refractory anemia with ring sideroblasts associated with marked thrombocytosis (RARS-T), and in some patients within the WHO category of myeloproliferative neoplasms, unclassifiable.

JAK2 is located on chromosome 9q24 and encodes for “Tyrosine-protein kinase JAK2” (JAK-2), an intracellular tyrosine kinase associated with the cytoplasmic domains of type 1 and type 2 cytokine receptors. The V617F mutation results in constitutive activation of JAK-2 and its downstream cell signaling pathways.

JAK2 V617F MUTATION ANALYSIS:
JAK2 V617F Mutation Analysis can be ordered separately or as part of our myeloproliferative neoplasm suite of tests. When ordered as part of a reflex panel, if PV is suspected, patients in whom JAK2 V617F mutations are not detected will automatically undergo JAK2 Exon 12. If ET or PMF are suspected, patients in whom JAK2 V617F mutations are not detected will automatically undergo CALR. If CALR mutations are not detected, they will automatically undergo MPL Exon 10.

REASONS FOR REFERRAL:
• Diagnosis of polycythemia vera, essential thrombocythemia and primary myelofibrosis.

METHOD:
The JAK2 V617F variant is detected by PCR-hybridization probes.

LIMITATIONS:
The lower limit of detection of the assay is approximately 1% (allele burden). The assay is expected to detect >99% of JAK2 V617F variants that are present at a level of 1% or greater with >99% specificity.

REFERENCE INTERVAL:
JAK2 V617F mutations are reported as mutation detected or not detected.
SPECIMEN REQUIREMENTS:
3-5 ml EDTA (lavender top) whole blood or EDTA bone marrow.

SHIPPING REQUIREMENTS:
Place the room temperature specimen and requisition in plastic bags, seal and insert in a Styrofoam container. Seal the Styrofoam container, place in a sturdy cardboard box and tape securely. Ship the package in compliance with your overnight carrier guidelines. Address package to:

Client Services/Molecular Oncology Laboratory
BloodCenter of Wisconsin
638 N. 18th Street
Milwaukee, WI 53233
800-245-3117, ext. 6250

TURNAROUND TIME: 5-7 days

CPT CODES: 81270

REFLEX ORDERING:
MPN (Myeloproliferative Neoplasms) Reflex - PV
  JAK2 V617F Mutation Analysis CPT Codes: 81270 Turnaround Time: 5-7 days
  JAK2 Exon 12 Mutation Analysis (if indicated) CPT Codes: 81403 Turnaround Time: additional 5-10 days
MPN (Myeloproliferative Neoplasms) Reflex - ET/PMF
  JAK2 V617F Mutation Analysis CPT Codes: 81270 Turnaround Time: 5-7 days
  CALR Mutation Analysis (if indicated) CPT Codes: 81479 Turnaround Time: additional 5-7 days
  MPL Exon 10 Mutation Analysis (if indicated) CPT Codes: 81403 Turnaround Time: additional 5-10 days

REFERENCES: