JAK2 Mutation Detection

**Indication:** Many patients with BCR/ABL negative myeloproliferative neoplasms carry a JAK2 V617F activating mutation in exon 14. The JAK2 V617F mutation has been reported in 95% of patients with polycythemia vera (PV), 60% patients with essential thrombocythemia (ET), 60% of patients with myelofibrosis (IMF), and in 3-5% of patients with myelodysplastic syndrome (MDS) and CMML. Additionally, a small minority of PV patients carry JAK2 exon 12 mutations.

**Testing Method:** Amplicon based targeted next generation sequencing. JAK2 mutation detection can be ordered as an individual test or as part of Myeloproliferative Neoplasm panel. JAK2 exons 12+14 are evaluated.

**Test Parameters:**

- **Diagnostic sensitivity:** This assay is designed to detect known single nucleotide variants only within defined regions. Nucleotide insertions and deletions more than 25bp or outside of the defined regions may not be detected. Gene rearrangements are not detected.
- **Technical sensitivity:** This assay may not detect certain mutations if the proportion of tumor cells in the sample studied is less than 5%.

**Turnaround Time:** 5-7 business days

**Sample Requirements:**

- **Blood - Specimen stability:** Ambient - 72 hours; Refrigerated - 1 week
  - 3 ml peripheral blood in lavender top tube (EDTA)
  - Note: One lavender tube of blood is sufficient for multiple DNA based tests

- **Bone marrow aspirates** (anticoagulated with either heparin or EDTA and, if possible, placed into tissue culture medium) - **Specimen stability:** Refrigerated - 1 week (ship cold)

**CPT Codes:** 81270, G0452