

## Polycythemia Vera, *JAK2* V617F with Reflex to *JAK2* Exon 12-15, Sequencing for Erythrocytosis, Varies

Test ID: PVJAK

**Explanation:** The updated testing algorithm expands the *JAK2* V617F positive reflex range from 0.06%-0.6% to 0.06%-2%, facilitating a more comprehensive testing algorithm. This range extends beyond the International Consensus Classification (ICC) recommendation of  $\leq 1\%$ .

### Current Testing Algorithm

The test starts with a highly sensitive DNA-based *JAK2* V617F test by allele-specific polymerase chain reaction. If the *JAK2* V617F result is negative or very low positive (**0.06%-0.6%**), *JAK2* exon 12-15 Sanger sequencing will be performed on the stored RNA sample. If a *JAK2* V617F mutation (**>0.6%**) is detected, no further testing will be performed.

The Sanger sequencing covers *JAK2* exons 12 through the first 90% of exon 15, which spans the region containing essentially all mutations reported in myeloproliferative neoplasms.

### New Testing Algorithm

The test starts with a highly sensitive DNA-based *JAK2* V617F test by allele-specific polymerase chain reaction. If the *JAK2* V617F result is negative or very low positive (**0.06%-2%**), *JAK2* exon 12-15 Sanger sequencing will be performed on the stored RNA sample. If a *JAK2* V617F mutation (**>2%**) is detected, no further testing will be performed.

The Sanger sequencing covers *JAK2* exons 12 through the first 90% of exon 15, which spans the region containing essentially all mutations reported in myeloproliferative neoplasms.

### Questions

Contact Connie Penz, Laboratory Resource Coordinator at 800-533-1710.