**UPDATE**

JAK2 V617F Mutation Testing Improves Sensitivity and Turnaround Time

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**BENEFITS**

- Improved, quantitative test results
- Detection limit reduced to 0.1%
- Turnaround time reduced to 24–48 hours
- Monitoring graph faxed each time sample is submitted

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**WHAT’S NEW?**

Effective Wednesday, December 10, PeaceHealth Laboratories will offer the quantitative JAK2 V617F mutation test with improved turnaround time and sensitivity. The new test replaces the previously-offered qualitative JAK2 Mutation (85334) test.

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**OVERVIEW**

The JAK2 V617F test detects the percentage of G>T transversion in the Janus kinase 2 (JAK2) gene resulting in a Valine-to-Phenylalanine substitution at position 617 of JAK2 (JAK2 V617F). The mutation is an acquired, somatic mutation present in the majority of patients with myeloproliferative cancer (myeloproliferative neoplasms), i.e. nearly 100% of patients with polycythemia vera and in about 50% of patients with essential thrombocytosis and primary myelofibrosis.

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**BACKGROUND**

The JAK2 V617F somatic mutation is found in nucleated myeloid cells and erythrocytic precursors of patients with nonchronic myeloid leukemia myeloproliferative syndromes, including polycythemia vera (>90%), essential thrombocytopenia (~50%) and chronic idiopathic myelofibrosis (35–60%).

This mutation is rarely present in myelodysplastic syndromes (≤5%), chronic myelomonocytic leukemia (<5%) or acute myeloid leukemia (≤2%), which may reflect a low but detectable mutation frequency among otherwise healthy persons.

It has not been reported in chronic myeloid leukemia, acute lymphoblastic or chronic lymphocytic leukemia, or lymphomas.

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**INDICATIONS FOR USE**

This test is most helpful to confirm a diagnosis of polycythemia vera in patients with increased red cell mass and a low serum erythropoietin level; it has a reasonably high negative predictive value for polycythemia vera to rule out this diagnosis.

The JAK2 V617F mutation test can also be used to monitor the effectiveness of therapies for the treatment of myeloproliferative cancer.

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The presence of JAK2 V617F is also useful to diagnose essential thrombocythemia or chronic idiopathic myelofibrosis when other clinically similar conditions have been ruled out (e.g., reactive thrombocytosis). However, the negative predictive values are too low to rule out these clinical entities. Importantly, the presence of the JAK2 V617F mutation does not obviate the need to perform bone marrow biopsy for any of these myeloproliferative syndromes.\(^5\)

Total genomic DNA is extracted from peripheral blood or bone marrow and tested for the presence of the V617F JAK2 mutation by allele-specific polymerase chain reaction amplification with primers specific for the wild type and mutant JAK2 V617F allele.

### Test Comparison

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<tr>
<td>Unit Code</td>
<td>85334</td>
<td>58132</td>
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<tr>
<td>Test Name</td>
<td>JAK2 Mutation</td>
<td>JAK2 V617F</td>
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<tr>
<td>Reported Result</td>
<td>Positive or Negative</td>
<td>Percent JAK2 V617F</td>
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<tr>
<td>Detection Limit</td>
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<tr>
<td>Turnaround Time</td>
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<td>24–48 hours</td>
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**TEST RESULTS**

Results are reported as a percentage of JAK2 V617F by using the numerical values of sample Mutant Quantity/(Sample Mutant Quantity + Sample Wild Type Quantity) x 100 (as shown in the above table). This assay has a detection limit of 0.1% and is ~100% specific for the JAK2 V617F allele, as determined by the PeaceHealth Molecular Pathology Laboratory.

A patient monitoring graph is faxed separately in addition to the quantitative results to monitor changes in the percent of cells with the JAK2 V617F mutation over time (particularly if the patient is on therapy).
Patient Name: ZZTest, Ann  
DOB: 1/2/1984

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<th>Test #</th>
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<td>5/3/14</td>
<td>VA358965</td>
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</tbody>
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QUESTIONS?
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ORDERING INFORMATION
58132: JAK2 V617F, Quantitative PCR
Methodology: Quantitative Polymerase Chain Reaction
Performed: Monday and Thursday
Released: Monday and Thursday, PM
CPT Code: 81270

SPECIMEN REQUIREMENTS
Collect: Peripheral blood or bone marrow in 4 mL lavender top tube (EDTA)
Handling: Ambient or refrigerated
Stability: Specimen must arrive at testing laboratory within 72 hours of collection
Transport: Ambient or refrigerated
Rejection Criteria: >72 hours, heparin, frozen
Retention: DNA extract will be stored two months

Note: Additional supplemental testing is available through our reference laboratory for the following tests: CALR Mutation Analysis, JAK2 Exon 12–14 Mutation Analysis and MPL Mutation Analysis.

REFERENCES