RML
REGIONAL MEDICAL LABORATORY

**Effective 11/05/2012**

The order code: **JAK 2 CELL** [5572375] is being replaced by the RML performed Test Code:

**JAK2 MUTAT** [9100185] JAK 2 MUTATION (V617F) ANALYSIS

The JAK2 V617F mutation is associated with myeloproliferative neoplasms. It is present in at least 95% of cases of polycythemia vera, and as such, is one of the major diagnostic criteria for this process. It is also detected in approximately 50% of cases of primary myelofibrosis and in 40-50% of cases of essential thrombocythemia. Therefore, while a positive test (reported as DETECTED) is consistent with a myeloproliferative neoplasm, it is not specific for the type, requiring correlation with other clinical, laboratory and bone marrow morphology findings for a final diagnosis.
A negative test (resulted as NOT DETECTED) does not rule out a myeloproliferative neoplasm, but it makes a diagnosis of polycythemia vera highly unlikely. If the clinical concern for polycythemia vera is very high and JAK2 V617F mutation testing is negative, additional tests to evaluate for other mutations of the JAK2 exons 12 and 13 may be performed, although these mutations are exceedingly rare. The limit of detection is 2% cells containing the V617F mutation in a background of normal cells.
However, samples that result very close to this limit of detection are considered inconclusive and will be reported as INDETERMINATE, thus repeat testing in 3-4 months would be indicated.
Evaluation for the JAK2 V617F mutation was performed by RT-PCR methodology using the Ipsogen JAK2 MutaScreen kit. This test has not been cleared or approved by the FDA, but the FDA has determined that such clearance is not necessary to use this test for clinical purposes. Regional Medical Laboratory has extensively tested and validated its performance characteristics. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) as qualified to perform high complexity clinical laboratory testing.

REGIONAL MEDICAL LABORATORY
1923 SOUTH UTICA AVENUE
TULSA, OKLAHOMA 74104-6502

# JAK 2 Mutation (V617F) Analysis

**Order Name:** JAK2 MUTAT
Test Number: 9100185

| TEST COMPONENTS | REV DATE: 11/05/2012 |
|---|---|
| **Test Name:** | **Methodology:** |
| JAK 2 Mutation (V617F) Analysis | PCR |

**SPECIMEN REQUIREMENTS**

| | Specimen Volume(min) | Specimen Type | Specimen Container | Transport Environment |
|---|---|---|---|---|
| **Preferred** | **6mL(4mL)** | **Whole Blood** | **EDTA (Lavender Top)** | **Room Temperature** |
| **Alternate** | **6mL(4mL)** | **Bone Marrow** | **EDTA (Lavender Top)** | **Room Temperature** |
| | **6mL(4mL)** | **Bone Marrow** | **Sodium Heparin (Green Top)** | **Room Temperature** |

**Special Instructions:** 6mL(4mL) **Peripheral Whole Blood** in **EDTA** Lavender top tube. Please **keep specimens at room temperature** and do not centrifuge.
- or -
3mL(2mL) **Bone marrow** in **EDTA** Lavender top tube. If part of a Bone marrow work up the JAK 2 Mutation can be performed on **Sodium Heparin** green top tube provided there is adequate volume of aspirate to share with the cytogenetics. Please keep specimens at room temperature and do not centrifuge.

**NOTE: Information regarding draw time and date is required on the sample to ensure the stability of the sample can be maintained. Specimen integrity will be determined at the performing laboratory.**

**GENERAL INFORMATION**

| | |
|---|---|
| **Testing Schedule:** | **Batched Mon-Fri as needed** |
| **Expected TAT:** | 3-7 Days |
| **Clinical Use:** | Myeloproliferative disorders (MPDs) are clonal hematopoietic stem cell malignancies characterized by excessive production of blood cells by hematopoietic precursors. In addition to thrombotic and hemorrhagic complications, leukemic transformation can occur. The main members of MPD are Polycythemia Vera (PV), Essential Thrombocythemia (ET) and Idiopathic Myelofibrosis (MF). The molecular pathogenesis of most MPDs is unknown. This V617F mutation leads to constituitive tyrosine phosphorylation activity that promotes cytokine activity and induces erythrocytosis. The V617F mutation in JAK2 is a dominant gain-of function mutation that contributes to the expansion of the myeloproliferative disorder clone. |
| **Cpt Code(s):** | 83891, 83892, 83907, 83896x2, 83898x2, 83914, 83912 |