



MEMO

To: All Regional Medical Laboratory (RML) Clients

From: Dr. Cindi Starkey, M.D., Ph.D., Chief of Molecular Diagnostics.
Sonja Matthews MT (ASCP), SC, Director of Clinical Operations

Date: December 3, 2012

Subject: **JAK2 Mutation Testing by Molecular Method**

RML is pleased to announce we are now offering JAK2 V617F mutation testing by PCR in house. We will be utilizing Ipsogen's (Qiagen) RT PCR test. This is a qualitative molecular test that detects the V617F mutation of the JAK2 gene and can detect as little as 2% tumor cells within a background of normal cells. Turn around time will be improved by performing this assay in house.

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 testing to evaluate for exon 12 mutations may be performed for an additional charge, 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 are positive but very close to the limit of detection (2-6%), will be reported as inconclusive, and repeat testing in 3-4 months would be suggested. Specimen requirement: peripheral blood collected in EDTA, and stored at room temperature for up to one week. This may also be automatically ordered for any new diagnosis of PV or MPN for patients of your practice if desired by calling Client Services at 918-744-2150.

Test Name: **JAK2 MUTATION ANALYSIS**
Order Mnemonic: **JAK2 MUTAT**
Test number: **9100185**
Methodology: Real time PCR (RT-PCR)
Reference Range: Negative for mutation
CPT code: 83891, 83892, 83907, 83912, 83896(x2), 83898 (x2), 83914 (x1) **-or-**
New Tier CPT Code: 81270 Effective Jan 1, 2013

If you have questions, please contact Dr. Cindi Starkey, M.D., Ph.D. Chief of Molecular Diagnostics or Sonja Matthews, MT (ASCP), SC, Director of Clinical Operations at 918-744-2553 or 800-722-8077.