JAK2 1849G>T [V617F] Mutation in Myeloproliferative Neoplasms

The UNC Molecular Genetics Laboratory performs a molecular test to detect JAK2 1849G>T [V617F] acquired mutation associated with myeloproliferative neoplasms (MPN) including polycythemia vera, essential thrombocytopenia, and primary myelofibrosis.

**Biology of the disease:** Somatic mutation 1849G>T in exon 14 of the Janus kinase 2 gene (JAK2) has been identified as an oncogenic event and a molecular marker of polycythemia vera (PV), essential thrombocythemia (ET), and primary myelofibrosis (PMF). The majority of PV cases (> 95%), and about half of ET and PMF cases harbor the JAK2 1849G>T mutation. Homozygous mutation is often seen in PV and PMF, whereas heterozygous mutation in ET is associated with a low mutant allele burden. The mutation is specific for these (and a very low proportion of other) myeloproliferative or myelodysplastic diseases, meaning that a positive result is indicative of a clonal myeloid neoplasm while a negative result does not exclude any of these diseases.

**Clinical Indications for JAK2 mutation testing:** Testing is recommended in patients suspected of having myeloproliferative neoplasia. This includes uncertain etiology for elevated hemoglobin (e.g. no volume depletion or hypoxia, EPO low or normal), granulocytosis (no active infection), or thrombocytosis (e.g. no hyposplenism, iron deficiency, or inflammation).

**Laboratory Testing for JAK2 mutation:** The preferred sample is 2mL of EDTA anticoagulated blood (lavender-top), which may be refrigerated up to 48 hours before analysis by real-time PCR and probe hybridization followed by melt curve analysis. Results are reported as positive, or as negative to a sensitivity of 5% of DNA in the sample.

**References:**

**Questions?** Call the Molecular Genetics Lab at (919) 966-4408 or Dr. Gulley at 843-4595. E-mail margaret_gulley@med.unc.edu

Website= http://labs.unchealthcare.org/directory/molecular_pathology/index_html