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## **NEW JAK2 MUTATION TEST FOR MYELOPROLIFERATIVE DISORDERS (MPD)**

### **Polycythemia Vera (PV) Essential Thrombocythemia (ET) Idiopathic Myelofibrosis (IMF)**

#### **Most patients with myeloproliferative disorders carry a V617F JAK2 mutation:**

- 74 - 97% of patients with polycythemia vera (PV)
- 33 - 57% of patients with essential thrombocythemia (ET)
- 35 - 50% of patients with myelofibrosis (IMF)
- Also present infrequently (3-5%) in myelodysplastic syndrome (MDS) & CMML
- JAK2 V617F mutation (auto-inhibitory domain) activates the kinase

#### **Clinical Utility of Direct Diagnostic Test for the V617F JAK2 mutation:**

- To confirm a diagnosis of PV, ET, and IMF
- To confirm a clonal hematopoietic stem cell disorder
- To confirm MPD in cases with high erythrocyte, leukocyte, or platelet counts
- May obviate the need for slow Endogenous Erythroid Colony (EEC) growth test
- JAK2 targeted therapies may soon be available

#### **Testing Information:**

- allele-specific PCR for mutant and wild type alleles
- Detection sensitivity ~1% (i.e., mutant clones constituting >1% of leukocytes will be detected)
- Reviewed and reported by board-certified Pathologist (within 1 week)

#### **Sample Requirements:**

- 5-10 mL of blood or bone marrow — yellow (ACD) or purple (EDTA) tube (unspun)
- Deliver to lab at shipping address above within 24 hours of collection, if sample can not arrive within 24 hours, refrigerate until sample can be transported, then transport at room temperature.
- For shipping, maintain sample at room temperature or refrigerator temperature - do not freeze

For more information about this and other tests offered by the OHSU Molecular Diagnostic Center go to [www.ohsulabs.com](http://www.ohsulabs.com) or call the phone numbers listed above.

#### References:

James et al. Nature 434: 1144-48, 2005.  
Baxter et al. The Lancet: 365: 1054-61, 2005.  
Kralovics et al. N Engl J Med 352: 1779-90, 2005.  
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