



JAK2: A new Molecular test for Myeloproliferative disorders

Emory Medical Labs is now offering testing for the V617F mutation of JAK2.

Janus Kinase 2 (JAK2) is a component of the erythropoietin receptor signaling cascade. The V617F mutation makes JAK2 constitutively active. This leads to proliferation of a variety of cells derived from the myeloid lineage include red blood cells, neutrophils, and/or platelets. The mutation is found in greater than 90% of patients with polycythemia vera, half of patients with essential thrombocythemia, and many patients with chronic idiopathic myelofibrosis.

Identification of a V617F mutation in JAK2 defines a subset of patients with Philadelphia chromosome negative myeloproliferative disorders. Although not currently available, new strategies for treating patients with mutated JAK2 are being investigated that directly target the constitutively active JAK2.

Testing for the mutation is an adjunct to the clinical history and routine lab testing for differentiating myeloproliferative disorders from reactive processes. A negative test, however, does not rule out a myeloproliferative disorder.

Testing for V617F of JAK2 is currently offered by the Molecular Diagnostics Laboratory of Emory Medical Labs and is performed once per week. The test is conducted on a peripheral blood specimen collected in a lavender top (EDTA) vacutainer tube and should be sent as whole blood at room temperature.

Please call the Molecular Diagnostics Laboratory at 404-712-0606, for more information