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JAK2 MUTATION ANALYSIS

January 11, 2011

Dear Colleague:

Pathology, Inc. is pleased to announce the availability of a new assay in our molecular test menu: **JAK2 Mutation Analysis**. This assay utilizes polymerase chain reaction (PCR) followed by pyrosequencing for mutation detection and identification.

JAK2

Mutations in the **JAK2** gene can be used in conjunction with bone marrow histology and cytogenetic analysis to assist in the **diagnosis of myeloproliferative neoplasms** (MPN). The JAK2 V617F mutation is found in most patients with polycythemia vera (PV) (65-97%) and in nearly one-half of those with primary myelofibrosis (PMF) (35-57%) and with essential thrombocythemia (ET) (23-57%).

Bone marrow or whole blood is acceptable. Please submit ambient or refrigerated specimens—DO NOT FREEZE. Analysis is performed by PCR/Pyrosequencing and can detect down to 5% mutant in a background of wild-type genomic DNA.

We thank you for choosing Pathology, Inc. and look forward to your continued support. For additional information, please visit our website at www.pathologyinc.com or contact client services at 877.922.7284.

Sincerely,

A handwritten signature in black ink, appearing to read "Alfred Lui".

Alfred Lui, M.D.
Chief Medical Officer

New Tests (Pathology, Inc.) – Effective January 11, 2011

6000 JAK2 Mutation Analysis

Component	Method	Result	Mutations Identified
JAK2	PCR/Pyrosequencing	Normal/Abnormal	Wild-type/Mutation in codon 617

Specimen/Stability	Whole Blood EDTA (4mL) -OR- Whole Blood ACD (4mL) -OR- Bone Marrow ACD -OR- Bone Marrow EDTA; 72 hour stability ambient or refrigerated. DO NOT FREEZE.
Collection Instructions	4mL Whole Blood EDTA or ACD Ambient or Refrigerated; DO NOT FREEZE. Stable for 72 hours.
Sensitivity	The limit of detection is 5% mutant in a background of wild-type genomic DNA
Clinical Utility	<p>The JAK2 V617F (exon 14) mutation analysis can be used in conjunction with bone marrow histology and cytogenetic analysis to assist in the diagnosis of myeloproliferative neoplasms (MPNs). The JAK2 V617F mutation is found in most patients with polycythemia vera (PV) (65-97%) and in nearly one-half of those with primary myelofibrosis (PMF) (35-57%) and with essential thrombocythemia (ET) (23-57%). It is also infrequently present (3%–5%) in myelodysplastic syndrome, chronic myelomonocytic leukemia, and other atypical chronic myeloid disorders.¹ Reactive hematopoietic disorders can be distinguished from PV, ET, and PMF by identifying the JAK2 mutation in BCR/ABL-negative MPNs.² Furthermore, JAK2 mutation has not been reported in Philadelphia chromosome-positive CML.³</p> <p>References:</p> <ol style="list-style-type: none"> 1. Steensma DP, Dewald GW, Lasho TL, et al. The JAK2 V617F activating tyrosine kinase mutation is an infrequent event in both "atypical" myeloproliferative disorders and myelodysplastic syndromes. <i>Blood</i>. 2005;106:1207–1209. 2. Campbell PJ, Scott LM, Buck G, et al. Definition of subtypes of essential thrombocythaemia and relation to polycythaemia vera based on JAK2 V617F mutation status: a prospective study. <i>Lancet</i>. 2005;366:1945-1953. 3. M. Cankovic, L. Whiteley, R. C. Hawley, R. J. Zarbo, and D. Chitale. Clinical Performance of JAK2 V617F Mutation Detection Assays in a Molecular Diagnostics Laboratory: Evaluation of Screening and Quantitation Methods. <i>Am J Clin Pathol</i>, November 1, 2009; 132(5): 713-721.