JAK2 Mutation Analysis

PARTNER IN PATHOLOGY

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OVERVIEW

The Janus kinas 2 (JAK2) gene located on chromosome 9p is a key component of cell growth and differentiation. A mutation in the JAK2 gene (V617F) results in constitutive activation of the JAK2 pathway.[1] JAK2 mutation status is routinely determined when BCR/ABL-negative chronic myeloproliferative neoplasms (MPNs) are suspected.[2] In 2008, JAK2 mutation status was determined to be an important criterion in classifying myeloproliferative neoplasms and the WHO classification scheme was updated accordingly.[1]

MUTATION ANALYSIS

JAK2 V617F (exon 14) mutation analysis can be used in conjunction with bone marrow histology and cytogenetic analysis to assist in the diagnosis of 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%).[2] It is also infrequently present (3%–5%) in myelodysplastic syndrome, chronic myelomonocytic leukemia, and other atypical chronic myeloid disorders.[3]

Reactive hematopoietic disorders can be distinguished from PV, ET, and PMF by identifying the JAK2 mutation in BCR/ABL-negative MPNs.[4] Furthermore, JAK2 mutation has not been reported in Philadelphia chromosome-positive chronic myelogenous leukemia.[2]

METHODOLOGY

Polymerase Chain Reaction/Pyrosequencing. Tumor cell DNA is extracted and PCR-amplified generating a DNA segment spanning exon 14 (codon 617). Pyrosequencing analysis is performed to quantify mutations in this region. The limit of detection is 5% mutant in a background of wild-type genomic DNA. The assay can detect the presence or absence of the V617F mutation.

ORDERING INFORMATION

TEST CODE	TEST NAME
6000	JAK2 Mutation Analysis
Specimen Requirements	Bone Marrow EDTA or ACD OR Whole Blood EDTA (4mL)
Collection	Send at room temperature or refrigerated. Stable for 72 hours. DO NOT FREEZE.

FOR MORE INFORMATION,
877.922.7284
call us at our toll-free number

REFERENCES:

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4. 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. Lancet 2005;366:1945-53.

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