JAK2 Mutation Analysis

JAK2 Mutation Analysis provides targeted detection of mutations in the JAK2 gene using DNA extracted from blood or bone marrow specimens. This test is used for diagnostic, prognostic, and predictive purposes associated with Hematological disorders.

Testing Method and Background

This test utilizes amplicon-based targeted **Next Generation Sequencing (NGS) technology** for detection of mutations in the JAK2 gene (exons 12 + 14), including detection of the common JAK2 V617F mutation using extracted DNA from blood or bone marrow specimens. Many patients with BCR/ABL negative myeloproliferative neoplasms carry a *JAK2* V617F activating mutation in exon 14. The *JAK2* V617F mutation has been reported in 95% of patients with polycythemia vera (PV), 60% patients with essential thrombocythemia (ET), 60% of patients with myelofibrosis (IMF), and in 3-5% of patients with myelodysplastic syndrome (MDS) and CMML. Additionally, a small minority of PV patients carry *JAK2* exon 12 mutations.

Highlights of JAK2 Mutation Analysis

Targeted Region

JAK2: Exons 12 + 14

- **Accurate Results from Low-Quality Samples**
  Workflow with low quantity of input DNA and accurate detection of variants down to 5% mutant allele frequency.
- **Wide-ranging Coverage of Variants**
  Assessment of single-nucleotide variants (SNVs) and small insertions/deletions within multiple target exons

Ordering Information

Get started (non-HFHS): Print a Molecular Hematologic Testing requisition form online at www.HenryFord.com/HFCPD

Get started (HFHS): Order through Epic using test "JAK2 Mutation" (MOL80441 Blood, MOL80442 Bone Marrow)

Specimen requirements:

- Peripheral Blood - 1-3ml in lavender top tube (EDTA) **Specimen stability: Ambient - 72 hours; Refrigerated - 1 week**
- Bone Marrow - 1-3ml, anticoagulated with either heparin or EDTA, **Specimen stability: Refrigerated - 1 week** (ship cold)
- Extracted DNA - from a CLIA-certified Laboratory

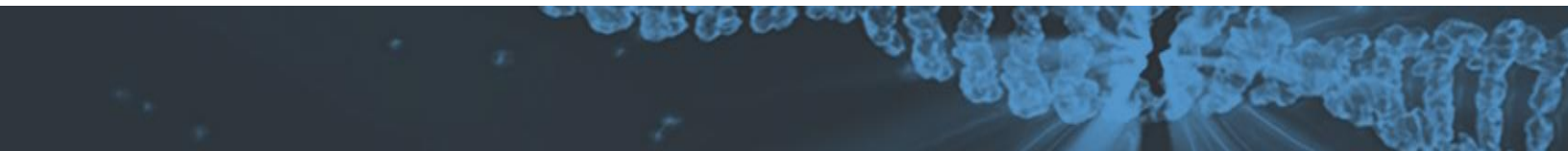
Cause for Rejection: Clotted, hemolyzed, or frozen specimens, improper anticoagulant, tubes not labeled with dual patient identification, non-dedicated tubes.

TAT: 5-10 business days (after Prior Authorization obtained)

Mail test material to:
Henry Ford Center for Precision Diagnostics
Pathology and Laboratory Medicine
Clinic Building, K6, Core Lab, E-655
2799 W. Grand Blvd., Detroit, MI 48202

CPT Codes: 81279, G0452 (88363 or 88381 may apply)

Contact us: Client Services, Account and Billing Set-up, and connect with a Molecular Pathologist at (313) 916-4DNA (4362)

For more information on Comprehensive Molecular Services, visit our website
www.HenryFord.com/HFCPD

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