

JAK2 V617F

Genetik und Klinik

A recurrent somatic mutation, V617F, affecting the Janus tyrosine kinase 2 (JAK2) gene, has been identified in 2005, leading to a major breakthrough in the understanding, classification, and diagnosis of myeloproliferative neoplasms (MPN). JAK2 is a critical intracellular signaling molecule for a number of cytokines, including erythropoietin.

The JAK2 V617F mutation is detected in >95% of patients with polycythemia vera (PV), 50–60% of patients with essential thrombocythemia (ET), and in 50% of patients with primary myelofibrosis (PMF). JAK2 V617F has been also detected in some rare cases of chronic myelomonocytic leukemia, myelodysplastic syndrome, systemic mastocytosis, and chronic neutrophilic leukemia, but in 0% of CML.

The mutation corresponds to a single-nucleotide change of JAK2 nucleotide 1849 in exon 14, resulting in a unique valine (V) to phenylalanine (F) substitution at position 617 of the protein (JH2 domain). It leads to constitutive activation of JAK2, hematopoietic transformation in vitro, and erythropoietin-independent erythroid colony (EEC) growth in all patients with PV and a large proportion of ET and PMF patients. JAK2 V617F represents a key driver in the transformation of hematopoietic cells in MPN, but the exact pathological mechanisms leading, with the same unique mutation, to such different clinical and biological entities remain to be fully elucidated.

Traditionally, the diagnosis of MPNs was based on clinical, bone marrow histology and cytogenetic criteria. The discovery of a disease-specific molecular marker resulted in both simplification of the process and increased diagnostic accuracy. Detection of the JAK2 V617F mutation is now part of the reference WHO 2008 criteria for the diagnosis of BCR-ABL negative MPN, and presence of this mutation is a major criterion for diagnostic confirmation.

(Reference: ipsogen JAK2 MutaScreen Kit Handbook, 01/2013)

Dienstleistung

Auftrag: JAK2 V617F
Fachbereich: Hämato-Onkologie
Methode: Allelspezifische PCR
Gen(e): JAK2

Untersuchungsmaterial

Probe: Venöses Blut oder Knochenmark **Probengefäss:** EDTA- oder Heparin-Röhrchen
Menge: 5-10ml

Praktische Informationen

Zustellung: Postexpress oder Veloblitz (Stadt Zürich) **Dauer:** 7 Tage
Preis (TP): Bei medizinischer Indikation gemäss Tarif Analysenliste
Bemerkung: -