



Test Catalog

Diagnostic. Prognostic. Predictive. Predisposition.



JAK2 Exon 12-13 Mutation Analysis

Alternative Name

JAK2 Mutation Analysis

Methodology

Molecular

Test Description

RT-PCR and bi-directional sequencing to detect mutations in exons 12-13, corresponding to the majority of the JAK2 pseudokinase domain. Exon deletion mutations are detectable. Testing is performed on plasma for increased sensitivity whenever possible. V617F analysis is recommended before or concurrently with this test. Exon 12-13 Mutation Analysis may be ordered separately, with concurrent V617F testing, by reflex after negative V617F testing, or as part of the [MPN JAK2 V617F with Sequential Reflex to JAK2 Exon 12-13, CALR, and MPL](#). Testing is approved for specimens from the state of New York.

Clinical Significance

While the majority of polycythemia vera (PV) patients carry the V617F mutation (~90%), most of those who are negative carry one of over 40 additional JAK2 mutations in exons 12-15. RNA-based testing in this assay allows detection of deletions not detectable by DNA-based tests. Mutation analysis helps differentiate reactive conditions from malignant erythrocytosis.

Specimen Requirements

- **Peripheral Blood:** 5mL EDTA tube
- **Bone Marrow:** 2mL EDTA tube

Note: Test is RNA-based. Please select **Extract & Hold – RNA** if specimen hold service is desired. If considering the MPN JAK2 V617F with Sequential Reflex to JAK2 Exon 12-13, CALR, and MPL test as an add-on, we recommend adding Extract & Hold – DNA with Extract & Hold – RNA order.

Storage & Transportation

Use cold pack for transport, making sure cold pack is not in direct contact with specimen. Ship same day as drawn whenever possible; specimens <7 days old preferred.

CPT Code(s)*

81279 (as of 01/01/2021); Prior to CPT Code was 81403

New York Approved

Yes

Level of Service

Global

Turnaround Time

7 days

Medical Necessity Resource

[Medical Necessity for NeoTYPE Myeloid Profiles](#)

*The CPT codes provided with our test descriptions are based on AMA guidelines and are for informational purposes only. Correct CPT coding is the sole responsibility of the billing party.

Please direct any questions regarding coding to the payor being billed.

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Committed to research as the means to improve patient care, we provide Pharma Services for pharmaceutical companies, in vitro diagnostic manufacturers, and academic scientist-clinicians. We promote joint publications with our client physicians. NeoGenomics welcomes your inquiries for collaborations. Please contact us for more information.

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