

# JAK2 (V617F) Mutation by ddPCR

Content Review: November 2023    Last Update: November 2023

Myeloproliferative neoplasms (MPNs) are a group of blood cancers that cause excess blood cell production in the bone marrow and often in the peripheral blood. Classic *BCR-ABL1*-negative MPNs include polycythemia vera (PV), essential thrombocythemia (ET), and primary myelofibrosis (PMF).<sup>1</sup> The majority of patients with PV, ET, and PMF have *JAK2*, *CALR*, or *MPL* gene variants; thus, genetic testing for *JAK2* is an important tool for the classification and diagnosis of these disorders.<sup>1</sup> A quantitative *JAK2* V617F assay can provide additional information about allele fraction, suggest hemizyosity/homozygosity, and be useful for monitoring ruxolitinib therapy.

For detailed information on the testing strategy for MPNs, including use of genetic testing for variants in the *CALR* and *MPL* genes, refer to the ARUP Consult [Myeloproliferative Neoplasms](#) topic.

## Genetics

### Variants Detected

*JAK2* (V617F) Mutation by ddPCR: point mutation c.1849G>T (V617F) of the *JAK2* gene

Additional variants will be evaluated if reflex testing is ordered; refer to [JAK2 \(V617F\) Mutation by ddPCR, Qualitative with Reflex to \*CALR\* \(Calreticulin\) Exon 9 Mutation Analysis by PCR and \*MPL\* Mutation Detection \(3016839\)](#) and [JAK2 \(V617F\) Mutation by ddPCR, Qualitative with Reflex to \*JAK2\* Exon 12 Mutation Analysis by PCR \(3016840\)](#) on the ARUP Laboratory Test Directory.

## Test Interpretation

### Analytic Specificity

*JAK2* (V617F) Mutation by ddPCR, Quantitative: >99%

*JAK2* (V617F) Mutation by ddPCR, Qualitative: >99%

### Limit of Detection

*JAK2* (V617F) Mutation by ddPCR, Quantitative: 0.2%

*JAK2* (V617F) Mutation by ddPCR, Qualitative: 0.5%

### Limitations

#### JAK2 V617F Mutation by ddPCR Analysis

- Variants in genes other than *JAK2* are not detected.
- Variant alleles of *JAK2* other than V617F (c.1849G>T) are not reported.
- Samples with *JAK2* V617F variants below the limit of reporting may not be detected.
- Results of this test must always be interpreted in the context of morphologic and other relevant data and should not be used alone for a diagnosis of malignancy.

## Featured ARUP Testing

### [JAK2 \(V617F\) Mutation by ddPCR, Quantitative 3003751](#)

**Method:** Droplet Digital PCR (ddPCR)

- Aids in the assessment of suspected MPNs
- Use to quantify the *JAK2* V617F variant in peripheral blood or bone marrow

### [JAK2 \(V617F\) Mutation by ddPCR, Qualitative 3004046](#)

**Method:** Droplet Digital PCR (ddPCR)

- Aids in the workup of suspected MPNs
- Use to quantify the *JAK2* V617F mutation in peripheral blood or bone marrow

Other tests that include *JAK2* (V617F) mutation by ddPCR testing:

### [JAK2 \(V617F\) Mutation by ddPCR, Qualitative With Reflex to \*CALR\* \(Calreticulin\) Exon 9 Mutation Analysis by PCR and \*MPL\* Mutation Detection 3016839](#)

**Method:** Droplet Digital PCR (ddPCR)/Capillary Electrophoresis

### [JAK2 \(V617F\) Mutation by ddPCR, Qualitative With Reflex to \*JAK2\* Exon 12 Mutation Analysis by PCR 3016840](#)

**Method:** Droplet Digital PCR (ddPCR)

Different limitations apply to reflex testing; refer to [JAK2 \(V617F\) Mutation by ddPCR, Qualitative with Reflex to CALR \(Calreticulin\) Exon 9 Mutation Analysis by PCR and MPL Mutation Detection \(3016839\)](#) and [JAK2 \(V617F\) Mutation by ddPCR, Qualitative with Reflex to JAK2 Exon 12 Mutation Analysis by PCR \(3016840\)](#) on the ARUP Laboratory Test Directory.

## References

1. National Comprehensive Cancer Network. [NCCN Clinical Practice Guidelines in Oncology: Myeloproliferative neoplasms](#). Version 3.2023. [Last update: Oct 2023; Accessed: Oct 2023]

ARUP Laboratories is a nonprofit enterprise of the University of Utah and its Department of Pathology, 500 Chipeta Way, Salt Lake City, UT 84108  
(800) 522-2787 | (801) 583-2787 | [aruplab.com](#) | [arupconsult.com](#)