





MEMORANDUM #21

TO: UNC Health Care System Attending Physicians, Housestaff, Nursing Coordinators,
Department Heads and Supervisors

FROM:  Margaret L. Gulley MD, Director of Molecular Pathology Programs
 Herbert C. Whinna MD, PhD, Medical Director, McLendon Clinical Laboratories

SUBJECT: JAK2 V617F Mutation Test in Myeloproliferative Neoplasia

DATE: March 22, 2017

Effective April 3, 2017, the Molecular Genetics Laboratory offers a *JAK2* V617F mutation test.

Clinical Indications for *JAK2* V617F Mutation Testing:

JAK2 mutation testing is useful in the workup of patients suspected of having *BCR-ABL1* negative myeloproliferative neoplasia (MPN). The *JAK2* 1849G>T [Val617Phe, V617F] missense mutation in exon 14 is present in ~95% of polycythemia vera and in about half of essential thrombocythemia (ET) and primary myelofibrosis (PMF).

A negative result for *JAK2* V617F mutation does not exclude a diagnosis of MPN. When high clinical suspicion for MPN remains, the *Myeloid Mutation Panel-MPN* is recommended to more comprehensively test for mutations in 5 genes strongly associated with MPN (*JAK2* exons 12 & 14, *MPL*, *CALR*, *SETBP1* and *CSF3R*). Additional information on the *Myeloid Mutation Panel-MPN* panel is available on the McLendon Labs website: <http://www.uncmedicalcenter.org/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics>

Specimen Requirements: The preferred sample is 3mL of EDTA anticoagulated blood (lavender-top) which may be refrigerated up to 72 hours before analysis by real-time PCR and allele-specific hybridization followed by melt curve analysis on a LightCycler. Bone marrow aspirate (1mL, EDTA) is also acceptable. Results are reported as positive, or as negative to a sensitivity of 5% of DNA in the sample.

Reference:

Vainchenker W and Kravolics R. *Blood*. 2017. 129 (6): 667-679.

If you have questions please call the UNC Molecular Genetics Lab at (984) 974-1825 or Dr. Margaret Gulley at (919) 843-4595; E-mail: margaret_gulley@med.unc.edu

Website: <http://www.uncmedicalcenter.org/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics/>