MEMORANDUM # 25

TO: UNC Hospitals Attending Physicians and Faculty Practice Physicians, Housestaff, Clinical Nurse Coordinators, Department Heads and Supervisors

FROM: Karen Weck MD, Director, Molecular Genetics Laboratory
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SUBJECT: ***JAK2* V617F mutation test in myeloproliferative neoplasia**

DATE: February 27, 2019

Effective February 5, 2019, the Molecular Genetics Laboratory offers a quantitative *JAK2* V617F mutation test. This testing replaces the previous qualitative *JAK2* V617F mutation assay.

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* c.1849G>T [p.Val617Phe, V617F] missense mutation in exon 14 is present in ~95% of polycythemia vera cases and in about half of essential thrombocythemia (ET) and primary myelofibrosis (PMF) cases. In addition, this quantitative assay may be used to monitor *JAK2* V617F mutation burden after therapy in some MPN patients.

Importantly, a negative result does not exclude a diagnosis of myeloid neoplasia. When high clinical suspicion for MPN remains, a *Myeloid Mutation Panel* (MPN) is recommended to more comprehensively test for mutations in genes strongly associated with myeloid neoplasia, including less common *JAK2* exon 12 mutations. Additional information on the *Myeloid Mutation Panel* is available on the McLendon Labs website: https://www.uncmedicalcenter.org/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics/

Specimen Requirements: The preferred sample is 3mL of EDTA anticoagulated blood or 1mL of bone marrow (lavender-top), which may be refrigerated up to 72 hours before analysis by droplet digital PCR. Results are reported as positive or negative to a sensitivity of 0.1% variant allele fraction. All positive samples will be reported with a quantitative value.

Reference:
Waterhouse, M., et al. Annals of Hematology, 95(5), 739–744, 2016. PMID: 30056580

Questions? Call the UNC Molecular Genetics Lab at **(984) 974-1825.**

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