Memorandum - Molecular #39

To: UNC Health System Attending Physicians, Housestaff, Clinical Nurse Coordinators, Department Heads and Supervisors

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Subject: Changes to molecular tests for myeloid malignancies at UNC

Effective February 5, the gene content will be updated for Myeloid Mutation Panels at UNC.

What is changing?

1. The total number of genes tested will increase from 34 to 50, with addition of *BCORL1, CDKN2A, CUX1, DDX41, GATA2, GNAS, JAK3, KMT2A, NF1, PHF6, PIGA, PRPF8, PTEN, RAD21, RRAS2, SH2B3, SMC3.*
2. The 50-gene panel includes currently tested genes, except for *NOTCH1* and *MYD88*, and the additional genes mentioned above. This test is orderable as **Myeloid Mutation Panel (MDS & MPN)** and is appropriate for patients with signs and/or symptoms concerning for a myeloid neoplasm.
3. For patients with acute myeloid leukemia (AML) or concern for AML, the current **Myeloid Mutation Panel (AML) with *FLT3* testing** will expand to 50 genes, as described above. It continues to include separate rapid *FLT3*-ITD/TKD testing and an RNA extract & hold.
4. A 3 gene **Myeloproliferative neoplasm (MPN) hotspot panel (*JAK2, CALR*, *MPL*)** continues to be available for patients being evaluated for a myeloproliferative neoplasm.

Why are these changes occurring?

1. Recent publications emphasize clinical utility of broader gene panels in myeloid neoplasms.
2. Improved diagnostic accuracy in myeloid neoplasms.

Specimen Requirements:

Bone marrow aspirate (1 mL, EDTA), peripheral blood (3mL, EDTA), or formalin-fixed, paraffin-embedded bone marrow clot sections (10 unstained slides, minimum area of marrow = 4mm^2) having at least 30% myeloid cells. The assay is sensitive to 5% variant allele fraction (VAF; 10% clonal cells). This test is NOT appropriate for minimal residual disease monitoring. For patients undergoing repeat testing, previously detected variants are reportable to 3% VAF.

Questions? Email Jonathan Galeotti (Jonathan.Galeotti@unchealth.UNC.edu), or call the UNC Molecular Genetics Lab at **(984) 974-1825.**

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