DATE: June 3rd, 2017  
TO: UCH Medical Staff, Housestaff, Patient Care Centers, and Outpatient Clinics, University Chicago Comprehensive Cancer Center  
FROM: Jeremy P. Segal, MD, PhD  
RE: Launch of NPM1 Next Generation Sequencing (NGS) Assay

Announcement
The Clinical Genomics Laboratory in the Division of Genomic and Molecular Pathology is pleased to announce the launch of a highly sensitive targeted NGS assay for detection of mutations in Exon 11 of NPM1 gene, available as of June 3rd, 2017.

Test information
This Next Generation Sequencing (NGS) assay is intended to detect insertion mutations reported in Exon 11 of NPM1 gene for the assessment of low level residual disease in AML patients. This NGS assay will replace the current fragment analysis based qualitative assay and has a much improved analytical sensitivity (0.2% vs 5%). Acceptable specimens include peripheral blood and bone marrow collected in purple top EDTA tubes. The test procedure involves DNA extraction, DNA quality/quantity assessment, three PCRs for target amplification, molecular barcoding and enrichment, followed by next generation sequencing (NGS) on the Illumina MiSeq sequencer and downstream analysis for detection of 4 nucleotide insertions.

Specimen Requirements
At least 200 µL of peripheral blood (PB) or bone marrow (BM) aspirate collected in EDTA tubes is required, the preferred age is less than 48 hours from the time of collection. Specimens with less than 200 µL may be tested at the discretion of the attending molecular pathologist.

Test ordering
The test can be ordered through Epic using the codes LABAPNPM for PB or LABAPNPMO for BM (Figure 1).

Reporting and Test limitations
The basic report format is similar to the existing NPM1 assay, with identified mutations and variant allele frequencies reported. Assay sensitivity is 0.2% MAF.
Testing Frequency and Turnaround Time
Testing will be performed at least once weekly, Monday through Friday during day shifts only. Expected turnaround time is 5-10 business days following specimen receipt.

Additional Questions
Additional questions may be directed to the Division of Genomic and Molecular Pathology at 773-702-4946 or Dr. Jeremy Segal at 773-702-3674 or Dr. Lauren Ritterhouse at 773-702-8491.