Molecular Oncology Tests

**ABL1 mutation** – Sequence the kinase domain of the ABL1 gene to predict response to tyrosine kinase inhibitors in patients with Ph’+ leukemia

**BCR-ABL1 translocation** - Quantify p210 or p190 transcripts for diagnosis of chronic myelogenous leukemia, to classify acute lymphoblastic leukemia, and to monitor therapeutic efficacy

**BRAF Somatic Mutation, Hematologic Malignances** – Detect BRAF codon 600 mutation (in blood or bone marrow) in patients with leukemia, to distinguish hairy cell leukemia from other B-cell neoplasms.

**FLT3 ITD** – Detect FLT3 internal tandem duplication conferring a worse prognosis in acute myeloid leukemia

**FLT3 TKD** - Detect mutation in the FLT3 tyrosine kinase domain associated with response to midostaurin.

**GastroGenus Gastric Cancer Classifier** – In recurrent or metastatic cancer patient tissue, evaluate 28 genes to explore options for clinical trial enrollment.

**Immunoglobulin gene and T cell receptor genes** - Detect rearrangement of the IGH and TRG genes to assess clonality and lineage of B and T cell lesions

**MGMT methylation** – Detect promoter methylation associated with glioblastoma sensitivity to alkylating agents and with prolonged survival of glioma patients on temozolamide

**Microsatellite Instability** – In colon cancer, assess microsatellite length as a predictor of an acquired or inherited defect in a mismatch repair gene

**MLH1 methylation** – Test promoter methylation in the workup of possible Lynch syndrome

**Myeloid Mutation Panel** – Detect somatic mutations in 34 genes (performed on blood or bone marrow) useful for diagnosis, prognosis, and therapy selection in myeloproliferative disorders. Indications include acute myeloid leukemia, myelodysplastic syndrome, or myeloproliferative neoplasia.

**Myeloproliferative Neoplasm (MPN) Hotspot Panel, CALR, JAK2, and MPL** – In peripheral blood or bone marrow, testing for the presence of somatic mutations in these three genes
may assist in diagnosis or exclusion of BCR-ABL1 negative myeloproliferative neoplasms.

**Solid Tumor Mutation Panel** – In cancer tissue, detect mutations in 26 genes by massive parallel sequencing to guide selection of therapy or to explore options for clinical trial enrollment. A full list of targeted exons in BRAF, EGFR, KIT, KRAS, NRAS, PDGFRα, AKT1, PIK3CA, PTEN, AKT1, STK11, ERBB2 and 14 other genes is found in the flyer at: [http://www.uncmedicalcenter.org/uncmc/professional-education-services/mclendon-clinical-laboratories/directory/mol-ophys-genetics/](http://www.uncmedicalcenter.org/uncmc/professional-education-services/mclendon-clinical-laboratories/directory/mol-ophys-genetics/)

**TERT Promoter Mutation** – Test for TERT promoter mutation which, in concert with IDH1/2 mutation status and chromosome 1p/19q FISH results, impacts prognosis in patients with diffuse glioma.

**TP53 Somatic Mutation, Hematologic Malignancies** – Detect TP53 mutation (in peripheral blood or bone marrow) in patients with chronic lymphocytic leukemia and some other mature B-cell lymphomas/leukemias to inform prognosis.

**Heritable Disease Tests**

**Alpha-1-antitrypsin** - Detect mutation in the SERPINA1 gene, including E342K (Z allele) and E264V (S allele), responsible for deficiency of the enzyme alpha-1-antitrypsin

**Connexin 26 and 30** - Detect mutations in the GJB2 gene (exon 2) and selected deletions in GJB6 that alter the connexin 26 and 30 proteins associated with hearing loss

**CYP2C19 variants for clopidogrel therapy** - Detect CYP2C19 gene variants associated with drug efficacy, or bleeding complications, of clopidogrel (Plavix) therapy

**Cystic fibrosis** - test for common mutations in the CFTR gene, offered to women of childbearing age and to patients with signs or symptoms of cystic fibrosis

**Factor V & Factor II Prothrombin gene mutations** - Detect Factor V Leiden and prothrombin (F2 20210G>A) mutations associated with inherited predisposition to venous thrombosis

**Fragile X genotype** - Assess structural alteration of the FMR1 gene associated with Fragile X syndrome of mental retardation, premature ovarian failure, and tremor/ataxia syndrome

**Hereditary Hemochromatosis** - Detect HFE mutation (63H>D & 282C>Y) associated with inherited predisposition to iron overload

**Kidney Genetics Panel** – Detect mutation by next generation sequencing in 17 genes associated with hereditary forms of kidney disease, including Hereditary Nephrotic Syndrome, Focal Segmental Glomerulosclerosis, and Alport Syndrome

**MCAD deficiency** - Detect mutation of the ACADM gene responsible for deficiency of the enzyme medium-chain acyl-coenzyme A dehydrogenase

**Prader-Willi & Angelman Syndromes** - Detect methylated (maternal) and unmethylated (paternal) alleles of the SNRPN gene

**Primary Ciliary Dyskinesia** - Detect mutation by next generation sequencing in 36 genes associated with ciliary dysfunction in the respiratory tract or abnormal sperm motility.

(See website for required consent and history forms.)

**UGT1A1 genotype** – Assess promoter of the UGT1A1 gene to predict toxicity to irinotecan or to confirm a diagnosis of Gilbert’s syndrome
**Additional Test Services**

**DNA Fingerprinting** - In transplant patients, assess marrow engraftment or chimerism via identity testing to quantify the proportion of recipient and donor cells

**Custom DNA sequencing** – DNA sequencing can be done to detect selected gene variants. Call Dr. Booker to discuss options for testing

**DNA or RNA extraction and hold** - extract DNA or RNA from specimen and hold for at least 1 year

**Validation of assays for use in clinical trial use** – Sequencing, microarray, PCR panels, and other technologies are developed to suit the purpose of a given clinical investigation. For a description of services, visit [https://www.uncmedicalcenter.org/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics/](https://www.uncmedicalcenter.org/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics/)