# UNC Molecular Genetics Laboratory Test Menu

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## 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 myeloid leukemia, to classify acute lymphoblastic leukemia, and to monitor therapeutic efficacy

***BRAF* mutation –** detect *BRAF* V600 variants to classify anaplastic thyroid carcinoma and other malignancies

***FLT3*** - In acute myeloid leukemia, detect *FLT3* internal tandem duplication conferring worse prognosis, and kinase domain mutation impacting response to targeted therapy

**GastroGenus Gastric Cancer Classifier –** In recurrent or metastatic cancer patient tissue, evaluate variants in 27 human genes (listed on website) and assign Epstein-Barr virus status, to explore options for immunotherapy or clinical trial enrollment

***JAK2* mutation –** quantify *JAK2* 1849G>T [V617F] to monitor myeloproliferative neoplasia

***IDH* mutation –** Classify glioma by detecting *IDH1* R132 or *IDH2* R172 mutation

**Immunoglobulin and T cell receptor genes** - Detect rearrangement of the *IGH, IGK* 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 patients on temozolamide

**Microsatellite Instability** – In gastrointestinal or endometrial cancer, assess microsatellite lengths to test for defective DNA mismatch repair which can be acquired or inherited. MSI-high results predict response to immunotherapy and increased likelihood of Lynch syndrome.

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

**Myeloid Mutation Panel –** In blood or marrow, detect hotspot mutations in 35 genes (listed on website) to facilitate diagnosis and management of patients with acute myeloid leukemia, myelodysplastic syndrome, or myeloproliferative neoplasia

**Myeloproliferative Neoplasm Hotspot Panel** – In suspected myeloproliferative neoplasia, detect relevant mutations in *JAK2, CALR,* or *MPL* genes

**Prosigna Breast Cancer Risk of Recurrence Score** – Assess prognosis in early stage ER or PR positive, Her2 negative breast cancer with up to 3 nodes positive.

**Solid Tumor Mutation Panel –** In cancer tissue, detect hotspot mutations in 26 genes to inform therapy selection. Target exons in *BRAF, EGFR, KIT, KRAS, NRAS, PDGFRA, PIK3CA, PTEN, AKT1, STK11, ERBB2* and 14 other genes is listed on our website

***TERT* mutation –** In glioma, refine prognosis based on *TERT* promoter -124C>T or -146C>T

***TP53* mutation –** In chronic lymphocytic leukemia or small lymphocytic lymphoma, refine prognosis based on *TP53* gene variants

## Heritable and Congenital 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 mutation 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

**Congenital CMV test** - Detect cytomegalovirus in Guthrie card (newborn) blood to assess likelihood of congenital infection. Referral to the Pediatric Genetics Clinic is required to access residual blood from NC archives. (For a clinic appointment call (919) 966-4202)

**Factor V & 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

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

**Kidney Heritable Mutation Panel** – Detect mutation in 17 genes associated with Hereditary Nephrotic Syndrome/ Focal Segmental Glomerulosclerosis (FSGS)/ Alport Syndrome

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

**Primary Ciliary Dyskinesia -** Detect mutation in 35 genes (listed on website along with required history/consent forms) associated with ciliary dysfunction in respiratory and genital tracts.

***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 is available for selected gene variants. Call Dr. Booker to discuss options for testing

**DNA or RNA extract and hold** - extract DNA or RNA from specimen and hold for ≥ 1 year

**Validation of assays for use in clinical trials –** Genomic DNA/RNA sequencing, PCR, microarray, & other technologies (see website) are custom-developed to suit each clinical investigation

**UNC Molecular Genetics Laboratory Website:**
http://www.uncmedicalcenter.org/uncmc/professional-education-services/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics/