

UNC Molecular Genetics Laboratory Test Menu

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http://labs.unchealthcare.org/directory/molecular_pathology/index_html

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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.

***BCR-ABL1* translocation** - Quantitate p210 or p190 transcripts to aid in the diagnosis of chronic myelogenous leukemia, the prognosis of acute lymphoblastic leukemia, and to monitor therapeutic efficacy.

***ABL1* mutation detection** – Sequence the kinase domain of the *ABL1* gene to predict responsiveness to tyrosine kinase inhibitors in patients with Ph'+ leukemia.

***JAK2* mutation detection** – Detect *JAK2* 1849G>T [V617F] mutation associated with neoplastic myeloproliferative diseases.

***FLT3* and *NPM1* mutation detection** – Detect *FLT3* internal tandem duplication and *NPM1* insertional mutation conferring prognosis in acute myelogenous leukemia of normal karyotype.

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

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

EBV viral load - Quantitate Epstein-Barr virus in plasma, CSF, or tissue to assist in diagnosis and monitoring of EBV-related post-transplant lymphoproliferative disorder, nasopharyngeal carcinoma, or AIDS-related brain lymphoma.

CMV viral load - Quantitate cytomegalovirus in blood to assist in diagnosis and monitoring of CMV disease.

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

BK viral load - Quantitate BK virus in plasma or urine to assist in diagnosis and monitoring of BK viral nephropathy in renal transplant recipients.

***UGT1A1* genotype** – Assess TA sequence repeat length in the promoter of the *UGT1A1* gene to predict toxicity to irinotecan (Camptostar) or to confirm a diagnosis of Gilbert's syndrome.

EGFR mutation- Detect mutations in *EGFR* exons 18, 19, 20 and 21 in lung cancer to predict response to anti-EGFR therapy.

KRAS mutation- Detect mutations in *KRAS* codons 12 and 13 in colon and lung cancers to predict resistance to anti-EGFR therapy.

BRAF mutation- Detect Val600Glu mutation in *BRAF* to predict resistance to anti-EGFR therapy and for prognosis of colon cancer, to essentially rule out Lynch syndrome, and to classify melanoma or papillary thyroid carcinoma.

KIT mutation- In melanoma or gastrointestinal stromal tumor tissue, detect mutations in *KIT* that predict responsiveness to tyrosine kinase inhibitor therapy.

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

Hemochromatosis - Detect *HFE* mutations (63H>D and 282C>Y) associated with inherited predisposition to iron overload in patients with high transferrin saturation (>45%), elevated serum ferritin, hepatic siderosis, or other evidence of iron overload.

BRCA1 & BRCA2 gene tests - Detect heritable mutations in the *BRCA1* or *BRCA2* genes predisposing to breast and ovarian cancer. Prerequisites include a family history and referral from our Cancer Genetics Clinic. (For a clinic appointment call (919) 966-9437.)

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

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.

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

MCAD deficiency - Detect 329K>E mutation of the *ACADM* gene responsible for deficiency of the enzyme medium-chain acyl-coenzyme A dehydrogenase.

Aminoglycoside toxicity - Detect 1555A>G mutation of the *MT-RNR1* gene responsible for non-syndromic hearing loss and predisposition to toxicity from aminoglycoside therapy.

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.

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

Primary Ciliary Dyskinesia - Detect mutations in the *DNAI1* and *DNAH5* genes associated with ciliary dysfunction in the respiratory tract or abnormal sperm motility. (Prior to ordering, visit our website for information for required consent and history forms.)

Custom DNA sequencing – For selected genes, DNA sequencing is done to detect mutations associated with disease. Call Dr. Booker to discuss the possibility of testing the gene or disease of interest, 966-4408.

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