

# Molecular Diagnostics

The Allina Health Laboratory Molecular Diagnostic Laboratory provides high quality molecular biological analyses, serving as a reference laboratory for clinical outreach work and also as a resource for clinical research projects.

The Molecular Laboratory employs numerous molecular biology techniques including qualitative polymerase chain reaction (PCR), quantitative Reverse Transcription PCR (RT-PCR), and Restriction Fragment Length Polymorphism (RFLP) Analyses to examine the human genome for alterations.

## B Cell Gene Rearrangement

Detection of clonal rearrangements of immunoglobulin heavy chain genes that are associated with malignant processes.

## T Cell Gene Rearrangement

Detection of clonal rearrangements of T cell receptor genes that are associated with malignant processes.

## Factor II (Prothrombin) G20210A Mutation Assay

Detection of a point mutation (G to A at position 20210) of the human Factor II gene. This is a pro-thrombotic mutation that has been shown to be involved in venous thrombosis.

## BRAF V600 Mutation Assay

Detection of a point mutation most commonly resulting in a V to E change at codon 600. The mutation status of a tumor may influence drug therapy choices (melanoma, lung and colon) or aid in diagnosis (i.e. Lynch Syndrome).

## EGFR Mutation Assay

Detection of deletions, insertions and mutations in exons 18, 19, 20 and 21 of the EGFR gene. The mutation status of a tumor influences the therapy choices available to the patient.

## Cystic Fibrosis (CF) Assay

Analysis of 23 mutations in the CFTR gene to identify carriers of CF. This assay conforms to the current ACOG recommendations for CF screening. *This assay is not appropriate for CF diagnosis in children or adults presenting with CF related conditions. An appropriate test is available as a send out to Mayo Medical Laboratories (informed consent required).*

### Phone

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## Factor V Leiden Mutation Assay (FVL)

Detection of a point mutation (G to A at position 1691, also known as the Factor V Leiden mutation) of the Factor V gene. This is a pro-thrombotic mutation that is regarded as the most prevalent coagulation abnormality associated with venous thrombosis.

## MTHFR C677T Assay

Detection of a cytosine to thymine substitution at position 677 (C677T) of the methylenetetrahydrofolate reductase (MTHFR) gene. This substitution causes a reduction in enzymatic activity. MTHFR genotyping is considered investigational for nutritional, mental health, or pharmacogenomic assessment.

## Hepatitis C Virus (HCV) Viral Load Assay

Detection and quantitation of the HCV virus in serum.

## Hereditary Hemochromatosis Gene Mutation Assay

Detection of two point mutations (C282Y and H63D) of the human HFE gene. These mutations are associated with iron accumulation and subsequent damage to organs.

## Human Immunodeficiency Virus 1 (HIV-1) Viral Load Assay

Detection and quantitation of the HIV-1 virus in plasma.

## Human Papillomavirus (HPV) Test, High Risk Screen

Detection of high risk HPV viruses in gynecological specimens (14 genotypes). These genotypes have been identified as the principal HPVs detectable in cancers.

## Human Papillomavirus (HPV) Types 16, 18 & High Risk

Genotype analysis of patients positive for HPV. Identifies patients with Type 16 and Type 18 genotypes that may influence treatment.

## Isocitrate Dehydrogenase (IDH) 1 and 2 Assay

Detection of point mutations in codon 132 of IDH1 and 172 of IDH2. This assay is used to help identify brain tumors and provide prognosis for the patient.

## JAK2 V617F Mutation Assay

Detection of point mutation (G to T mutation in exon 12 resulting in the V to F amino acid change at position 617) of the human JAK2 gene. This mutation is associated myeloproliferative disorders.

## FLT3/NPM1 Mutation Assay

Detection of mutations within the *fms*-related tyrosine kinase 3 (FLT3) and nucleophosmin (NPM1) genes, which are associated with several types of leukemias, predominantly acute myeloid leukemia. Two types of mutations are analyzed for FLT3: a point mutation in the kinase domain (D835, TKD) and duplications within the juxtamembrane domain (ITD). A 4 base pair insertion is analyzed for NPM1. The presence of these mutations provide prognosis for the patient.

## Calreticulin Exon 9 Assay

Detection of insertions or deletions in exon 9 of the calreticulin gene. These alterations in the gene are associated with myeloproliferative disorders.

## Microsatellite Instability (MSI) Assay

Analysis of MSI at 5 gene loci. MSI PCR is often performed in conjunction with analysis of DNA mismatch repair enzymes by immunohistochemistry. Deficient DNA repair and MSI are associated with Lynch Syndrome, and they also identify patients eligible for targeted therapies.