



Molecular Diagnostics Section, Laboratory of Pathology, Center for Cancer Research,  
National Cancer Institute, National Institutes of Health  
Bldg 10 / Room 3S249, (301) 480-8080

### Molecular Diagnostics Section Test Directory

Test Name	Assay Description	Assay Type	Specimen Type Accepted	Category/Use
B Cell Clonality	Immunoglobulin (IGH and IGK) gene rearrangements (PCR/Capillary Electrophoresis)	Qual.	Blood BMA CSF FNA FFPE Tissue	Hematopathology/ Diagnosis
T Cell Clonality	T cell receptor gamma locus (TRG) gene rearrangements (PCR/Capillary Electrophoresis)	Qual.		
EBV (EBNA2)	Lymphoma-associated virus detection (real-time qPCR)	Qual.		
HHV8	Viral detection (PEL, Castleman's disease, Kaposi's Sarcoma (real-time qPCR)	Qual.		
HTLV-I (Pol)	Human T-cell leukemia virus detection (real-time qPCR)	Qual.		
MYD88	MYD88 c.794T>C, p.L265P mutation detection. (droplet-digital PCR high sensitivity assay)	Qual.	Blood BMA FFPE Tissue	
Desmoplastic Round Cell Sarcoma	EWSR1/WT1, t(11;22) translocation detection (real-time qRT-PCR)	Qual.	FFPE Tissue	Sarcoma/ Diagnosis
Ewing's Sarcoma	EWSR1/ERG, t(21;22) EWSR1/FLI1 Type1, t(11;22) EWSR1/FLI1 Type2, t(11;22) translocation detection (real-time qRT-PCR)	Qual.		
Rhabdomyosarcoma Sarcoma	PAX3/FOXO1, t(2;13) PAX7/FOXO1, t(1;13) translocation detection (real-time qRT-PCR)	Qual.		
Synovial Sarcoma	SYT/SSX1, t(X;18) SYT/SSX2, t(X;18) translocation detection (real-time qRT-PCR)	Qual.		

Cancer Gene Mutation-Standard Panel (a multiplex 50 gene panel with next-generation sequencing)	Ion Torrent Cancer Hotspot Panel (CHP2). Targets ~2850 COSMIC hotspot mutations in 50 cancer genes (ABL1, AKT1, ALK, APC, ATM, BRAF, CDH1, CDKN2A, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, FLT3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2, JAK2, JAK3, KDR, KIT, KRAS, MET, MLH1, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RET, SMAD4, SMARCB1, SMO, SRC, STK11, TP53, VHL)	Qual.	FFPE Tissue	Solid adult cancers/Protocol-specific screening assay
Oncomine™ Comprehensive Assay v3 (OCAv3)	Ion Torrent next-generation sequencing (NGS) cancer panel. Detects most clinically relevant mutations, including single nucleotide variants, small insertions and deletions, copy number variants, and gene fusions from 161 unique cancer genes in one streamlined workflow	Qual.	FFPE Tissue	Adult and pediatric solid tumors/Oncology Clinical Research Protocols/Primary screening assay
BRAF V600E Mutation	BRAF c.1799T>A, V600E mutation detection. (droplet-digital PCR high sensitivity assay)	Qual.	Blood BMA CSF FNA FFPE Tissue	Erdheim-Chester disease, Papillary thyroid carcinoma, Hairy cell leukemia /Diagnosis or Oncology Clinical Research Protocol
EGFR Germline Mutation	EGFR germline mutation detection of T790M (droplet-digital PCR high sensitivity assay)	Qual.	Blood	NSCLC Genetic Study, protocol # 11-C-0096
MGMT Methylation	Methylation analysis of 12 CpG sites in the MGMT promoter region. (pyrosequencing)	Qual.	FFPE Tissue	Glioblastoma
Primary CNS Tumor Panel	Ion Torrent NGS custom amplicon panel. Targets 56 genes for detection of ~7,000 of SNVs/small Indels, CNVs, 64 known cancer associated fusion transcripts	Qual.		CNS tumor (various)/ Neuro-oncology Clinical Research Protocol
EGFRVIII RT-PCR	Detection of EGFR variant III expression (real-time qRT-PCR)	Qual.		Glioblastoma

### **Instructions for NIH Clinical Center CRIS orders of Molecular Pathology**

1. The Molecular Diagnostics Section of the Laboratory of Pathology (LP) offers a menu of diagnostic tests that are used in the diagnosis and treatment of cancer, and immune disease-related disorders. The test order can be found in the CRIS under Anatomic Pathology orders.
2. To order a molecular diagnostic test first enter the **CRIS ordering system**.
3. Select the **Anatomical Pathology** subsection.
4. Select **Molecular Pathology** test orders.
5. Select the appropriate test(s) that you wish performed. The Molecular Diagnostics Section printer only prints the tests you have selected. **It is critical that you select the correct tests to avoid delays.**
6. Please use the “**Special Instructions**” free text field as necessary. If you request an additional test for previously molecular reported case, please select the appropriate test(s) and write in the molecular report # in special instruction.
7. Review your order and submit it in CRIS. The order will print out at the Molecular Pathology Section printer.
8. If you have any question regarding which test to order, please call the laboratory for advice (see contact numbers below). This will save time and effort in the long run!

### **Specimen and Handling Requirements**

1. **Peripheral Blood:** minimum of 3 cc whole blood collected in light blue citrate tube. Place a second CRIS order (Research Blood) to Outpatient Phlebotomy to activate the blood draw. Deliver 10/Room 3S247 (open 7:30 AM to 4:00 PM)
2. **Bone Marrow Aspirates:** draw 2-3 cc in plain syringe with NO anticoagulant and immediately place into lavender top EDTA tube. Deliver 10/Room 3S247 (open 7:30 AM to 4:00 PM)
3. **All tissue specimens** from in-house surgery and outside materials (surgical specimens, tissue blocks, stained and unstained slides, and outside reports) are to be brought directly to General Surgical Pathology office - 10/Room 2S262 (open 7:30 AM to 4:00 PM). The molecular diagnostics laboratory cannot accession cases.
4. **Paraffin-embedded tissue specimens** should be formalin fixed. **Either tissue blocks (preferred) or unstained slides are acceptable.** The amount of tissue required for oncology tests on solid tumor is variable depending upon both specimen size and tumor content of the specimen. If you submit unstained slides, we request **10 unstained slides for small biopsies and cytology specimens**, or **5 unstained slides for larger excisional biopsies and resection specimens**. Generally, 20% tumor cells within the specimen are required for most mutation tests.

5. When submitting the molecular pathology order for tests on formalin-fixed, paraffin-embedded (FFPE) tissue, there are several possible situations that dictate how the order and materials should be submitted. These are listed below. Please follow the instructions appropriate for your patient's situation.
  - **The case has been signed out by the NCI, Laboratory of Pathology (LP) and specimen was retained in LP.** In this case, a single molecular pathology CRIS order is required for subsequent molecular pathology tests. Please indicate the LP surgical or cytopathology case accession number in the Special Instructions field of the request, if known (this number can be obtained from the NCI, LP, pathology report).
  - **A new tissue specimen is being submitted from a procedure performed at the NIH CRC.** In this case, two separate CRIS orders are required -- one for surgical pathology or cytopathology to request routine pathology services, and a second CRIS order for the molecular pathology test needed. Please also indicate in the special instructions box on the surgical pathology or cytopathology CRIS order that molecular testing is being requested on the sample.
  - **New submitted material from an outside hospital on a patient already admitted to the NIH CRC.** In this case bring the specimens (tumor block, or stained slides, unstained slides) with the outside pathology report to the Surgical Pathology or Cytopathology office, as appropriate. Two separate CRIS orders are required -- one for surgical pathology or cytopathology, and a second CRIS order for molecular pathology. Please indicate in the special instructions box on the surgical pathology or cytopathology CRIS order that molecular testing is being requested on the sample.
  - **New material from an outside hospital on a patient not yet admitted to the NIH CRC (CRIS ordering is not available).** In this case bring the specimens (tumor block, or stained, and unstained slides) with the outside pathology report to the Surgical Pathology or Cytopathology office as appropriate, and fill out the hand-written requisition form for surgical/cytopathology examination and specify the molecular test required. The resident or fellow responsible for the case will submit the molecular requisition.
  
6. Due to College of American Pathology (CAP) regulations, the laboratory personnel cannot provide the test results to physicians without permission. If you have an urgent case, please contact the Molecular Diagnostics Section. Dr. Raffeld, Dr. Patel or another pathologist will discuss the results with you.

For more detailed specimen collection and handling requirement, please go web site:

<https://ccrod.cancer.gov/confluence/display/CCRLP/Molecular+Pathology>

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