

## Molecular Pathology Tests

### Cytogenetics

#### Chromosome Studies

##### Postnatal:

- Peripheral blood
- Fibroblasts

##### Products of Conception:

- CVS (chorionic villi)
- Fetal tissue
- Placenta
- Skin
- Bone

##### Cancer:

- Bone Marrow
- Leukemic blood
- Lymph node
- Fatty tumors
- Renal tissue
- Solid tumors

##### Constitutional Chromosomal Microarray

- Whole Genome microarray (SNP array)

##### Constitutional FISH:

- X cen on PB
- Y cen on PB
- Yqh on PB
- SRY on PB
- X/ST on PB

### Molecular Genetics and Genomics

Alpha-1-antitrypsin genotyping (S and Z alleles)

Cystic Fibrosis NGS

DNA fingerprinting for identity (STR;PCR)

Factor V Leiden (PCR)

Fragile X syndrome (*FMR1*) DNA analysis (PCR)

*HFE* [Hereditary Hemochromatosis] (PCR)

*MTHFR* (PCR) (C677T,AIZ98C)

NOD2 exon sequencing

Progenitor cell engraftment monitoring (STR;PCR)

Prothrombin G20210A (PCR)

TGFBR2 exon sequencing

### Molecular Hematopathology

Alpha thalassemia (*HBA1/2*) gene deletions

B-cell clonality (*IGH* + *IGK* BIOMED-2 PCR)

BCR-ABL Qualitative Multiplex RT-PCR

*CEBPA* mutation analysis

T-cell clonality (TCRB + TCRG BIOMED-2 PCR)

*IGH* PCR (BIOMED-2 Primers)

*IGK* PCR (BIOMED-2 Primers)

*KIT* AML mutation analysis

*KIT* D816V (PCR)

*MPL* mutation analysis

*MYD88* L265P mutation analysis

*TCRB* PCR (BIOMED-2 Primers)

*TCRG* PCR (BIOMED-2 Primers)

p210 *BCR/ABL1*( RT-PCR, quantitative)

p190 *BCR/ABL1*( RT-PCR, quantitative)

Nucleophosmin gene (*NPM1*) mutation analysis

*JAK2* V617F mutation (PCR)

*JAK2* exons 12-15 sequencing

### Fluorescence in-situ hybridization

5q

*ALK*

*BCL2*

*BCL6*

*BCR/ABL1*

*BIRC3(API2)/MALT1*

*CBFB/MYH11* (inv 16)

CLL (13q,11q, 17p,+12)

*ETV6/RUNX1 (TEL/AML1)*

*FGFR1*

*IGH*

*IGH/BCL2*

*IGH/CCND1*

*IGH/MYC*

*MALT1(18q21)*

*MLL (KMT2A)*

*MYC*

Myelodysplasia (-5/5q, -7/7q,+8,-20q)

Plasma cell myeloma [13q, IGH, TP53, t(11,14), t(4;14), t(14;16)]

*PDGFRA*

*PDGFRB*

*PML/RARA*

*RARA*

*RUNX1/RUNX1T1 (AML1/ETO)*

Trisomy 4, 10 and 17

*Continued on back*

**Molecular Microbiology**

Affirm (for vaginitis)  
BK virus (FISH)  
BK virus PCR, quantitative  
*Bordetella pertussis* (NAAT)  
*Chlamydia trachomatis* (NAAT)  
*Clostridium difficile* PCR  
CMV detection (quantitative CMV PCR)  
Dimorphic fungi (probe analysis)  
EBV detection (quantitative EBV PCR)  
Enterovirus detection (RT-PCR)  
HBV detection (quantitative HBV PCR)  
HCV genotyping  
HCV viral load (quantitative HCV RT-PCR)  
HIV1 qualitative PCR  
HPVDNA (high risk types) – includes  
    Genotyping 16, 18 and other HR types  
HSV PCR (for CSF)  
Influenza A/Influenza B/RSV (multiplexed RT-PCR)  
*Legionella pneumophila* (PCR)  
*Mycobacterium sp.* identification (pyrosequencing and  
    probe analysis)  
*Mycobacterium tuberculosis* vs. NTM PCR  
    (smear positive only)  
*Neisseria gonorrhoeae* (NAAT)  
*Nocardia* and other aerobic actinomycetes identification  
    (pyrosequencing)  
*Pneumocystis jirovecii* (PCR)  
Respiratory Virus Multiplex PCR  
*S. aureus* (MRSA/MSSA) PCR (screens from nasal swabs)  
*Streptococcus* Group A, PCR  
*Streptococcus* Group B PCR  
*Trichomonas vaginalis* PCR

**Molecular Oncology**

*BRAF* for Hairy Cell Leukemia  
*BRAF* mutational analysis  
*EGFR* gene analysis  
*EGFR* mutational analysis (Cell pellet and tissue)  
*FISH* for MYC Angiosarcoma  
*HER2* (ERBB2) with D17S122  
*KRAS* gene analysis  
*MGMT* promoter methylation  
Microsatellite Instability (MSI)  
*RET*  
*ROS1*  
Transthyretin (TTR) exon sequencing  
VWF exon 28 sequencing

**Fluorescence in-situ hybridization**

*1p*  
*19q*  
*ALK* NSCLC FFPE  
*ALK* NSCLC ThinPrep  
*DDIT3* (CHOP)  
*EGFR* (gliomas)  
*EWSR1*  
*FISH* for bladder CA recurrence  
*FOXO1A* (FKHR)  
*FUS*  
*HER2* (ERBB2) with *CEP17*  
*MDM2*  
*SS18* (SYT)