

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

Chromosomal Microarray

- Constitutional
- Bone marrow cancer

Constitutional *FISH*:

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

Reflex Tests on Bone Marrow Chromosome Studies

- Bone marrow cancer chromosome microarray with normal, suboptimal, or cultures are no growth
- MDS *FISH* with suboptimal or cultures are no growth
- AML *FISH* with suboptimal or cultures are no growth

Molecular Genetics and Genomics

- Alpha-1-antitrypsin targeted genotyping
- Cystic Fibrosis NGS
- Factor V Leiden (PCR)
- Fragile X syndrome (*FMR1*) DNA analysis (PCR)
- HFE* [Hereditary Hemochromatosis] (PCR)
- MTHFR* (PCR) (C677T,A1Z98C)
- Progenitor cell engraftment monitoring (STR;PCR)
- Prothrombin G20210A (PCR)
- Spinal Muscular Atrophy (SMA)

TPMT Genotyping

Molecular Hematopathology

- Alpha thalassemia (*HBA1/2*) gene deletions
- B-cell clonality (*IGH* + *IGK* BIOMED-2 PCR)
- BCR-ABL Qualitative Multiplex RT-PCR
- CALR (Calreticulin) Exon 9 Mutation Analysis
- Tier 2 B-Cell Clonality (BIOMED-2 PCR)
- CEBPA* mutation analysis
- T-cell clonality (TCRB + TCRG BIOMED-2 PCR)
- IGH* PCR (BIOMED-2 Primers)
- IGK* PCR (BIOMED-2 Primers)
- 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
- FLT3* Gene Mutation
- JAK2* V617F mutation
- JAK2* exons 12-15 sequencing

Fluorescence in-situ hybridization

- 1p & 1q
- 5q
- 7q
- 20q/+8
- ALK*
- BCL2*
- BCL6*
- BCR/ABL1*
- CBFB/MYH11* (inv 16)
- CEP 9/15
- CLL (13q,11q, 17p,+12)
- ETV6/RUNX1 (TEL/AML1)*
- FGFR1*
- IGH*
- IGH/BCL2*
- IGH/CCND1*
- IGH/MYC*
- MALT1(18q21)*
- MLL (KMT2A)*

Continued on back

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 and 10

Molecular Oncology*MGMT* promoter methylation

MLH-1 Promoter Hypermethylation

Microsatellite Instability (MSI)

Solid Tumor Cancer Hotspots (NGS)

Lung Cancer Hotspot NGS (EGFR, KRAS, BRAF, MET)

Melanoma Cancer Hotspot NGS (BRAF, KRAS, KIT)

Colon Cancer Hotspot NGS (KRAS, NRAS, BRAF)

IDH1&2 Gene Analysis

BRAF Single Gene Analysis

EGFR Single Gene Analysis

IDH1 Single Gene Analysis

IDH2 Single Gene Analysis

KIT Single Gene Analysis

KRAS Single Gene Analysis

NRAS Single Gene Analysis

Fluorescence in-situ hybridization

1p & 19q

ALK NSCLC FFPET*ALK* NSCLC ThinPrep*DDIT3 (CHOP)**EGFR (gliomas)**EWSR1**FISH* for bladder CA recurrence*FISH* for cutaneous melanoma*FISH* for MYC Angiosarcoma*FOXO1A (FKHR)**FUS**HER2 (ERBB2) with CEP17**HER2 (ERBB2) with D17S122**MDM2**RET**ROS1**SS18 (SYT)**WWTR1/CAMTA1*