Cleveland Clinic

Cleveland Clinic Laboratories

Molecular Pathology Tests

Molecular Cytogenetics

Chromosome Studies

Postnatal:
- Peripheral blood
- Fibroblasts

Products of Conception:
- CVS (chorionic villi)
- Placenta
- Skin
- Bone

Cancer:
- Bone Marrow
- Leukemic blood
- Lymph node
- Fatty tumors
- Renal tissue

Constitutional Chromosomal Microarray
- Whole Genome microarray (SNP array)

Molecular Genetic Pathology

CYP2C9 and *VKORC1* Warfarin PGX
CYP2C19 Clopidogrel PGX
CYP2D6
CYP2D6 Tamoxifen PGX
Cystic fibrosis [ACOG panel] (PCR)
DNA fingerprinting for identity (STR;PCR)
Factor V Leiden (PCR)
HFE [Hereditary Hemochromatosis] (PCR)
MTHFR (PCR)
PLA1, *PLA2* (PCR)
Progenitor cell engraftment monitoring (STR;PCR)
Prothrombin G20210A (PCR)

Molecular Hematopathology

Alpha thalassemia (*HBA1/2*) gene deletions
B-cell clonality (*IGH* + *IGK* BIOMED-2 PCR)
CEBPA mutation analysis
T-cell clonality (TCRB + TCRG BIOMED-2 PCR)
IGH PCR (BIOMED-2 Primers)
IGK PCR (BIOMED-2 Primers)
TCRB PCR (BIOMED-2 Primers)
TCRG PCR (BIOMED-2 Primers)
BCL2 mbr (PCR)
p210 *BCR/ABL1*(RT-PCR, quantitative)
p190 *BCR/ABL1*(RT-PCR, quantitative)
BCR/ABL Kinase Domain Mutation Analysis
PML/RARA RT-PCR, qualitative
Nucleophosmin gene (*NPM1*) mutation analysis
FLT3 Gene Mutations**
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/MALT1*
- *IGH/MYC*
- *MALT1(18q21)*
- *MLL*
- *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

* Validated, soon to be implemented
** Performed at LabPMM

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Molecular Microbiology

Affirm (for vaginitis)
BK virus (FISH)
BK virus PCR, quantitative
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 (RT-PCR with reverse hybridization)
HCV viral load (quantitative HCV RT-PCR)
HIV DNA PCR
HIV viral load (quantitative HIV RT-PCR)
HPV DNA (high-risk types; hybrid capture)
HSV PCR (for CSF)
Influenza A/Influenza B/RSV (multiplexed RT-PCR)
Legionella pneumophilia (PCR)
Mycobacterium sp. identification (pyrosequencing and probe analysis)
Mycobacterium tuberculosis vs. NTM PCR
Neisseria gonorrhoeae (NAAT)
Nocardia and other aerobic actinomycetes identification (pyrosequencing)
Pneumocystis jiroveci (PCR)
S. aureus and *Candida albicans/C. glabrata* from positive blood cultures identification (FISH)
S. aureus (MRSA/MSSA) PCR (screens from nasal swabs)
Streptococcus Group A, probe analysis
Streptococcus Group B PCR

Molecular Oncology

BRAF mutational analysis
EGFR mutational analysis
KRAS mutational analysis
FISH for bladder CA recurrence
FISH for Cutaneous Melanoma
Fluorescence in-situ hybridization
- *1p*
- *19q*
- *ALK* NSCLC FFPET
- *ALK* NSCLC ThinPrep
- *DDIT3 (CHOP)*
- *EGFR*
- *EWSR1*
- *FOXO1A (FKHR)*
- *FUS*
- *HER2 (ERBB2)*
- *MDM2*
- *SS18 (SYT)*

LOH for *1p* (PCR)
LOH for *19q* (PCR)
MGMT promoter methylation
Microsatellite Instability (MSI)
PCA3

Tests in Development

Acanthamoeba PCR
ACTA2 exon sequencing
Alpha-1 antitrypsin exon sequencing
Alpha-1-antitrypsin genotyping (S and Z alleles)*
BRAF for Hairy Cell Leukemia
CEBPA mutation analysis
COL3A1 exon sequencing
FISH for aneusomy of chromosomes 7 & 17
Fragile X syndrome (*FMR1*) DNA analysis (PCR)*
KIT AML mutation analysis
KIT D816V PCR
KIT mutational analysis GIST & Melanoma*
MLH1 methylation
MPL mutation analysis
PRKAG2 and *LAMP2*
RT PCR for *EWSR1* (gene rearrangement)
RT PCR for *SYT* (gene rearrangement)
Solid Tumor SNP chromosomal array
Spectral Karyotyping
- Bone Marrow
- Blood
- Cell lines

Constitutional FISH:
- X cen on PB
- Y cen on PB
- *SRY* on PB
- *XIST* on PB

SMAD3 exon sequencing
TGFBR1 exon sequencing
TGFBR2 exon sequencing
Transthyretin (*TTR*) exon sequencing*
Vascular Panel (*ACTA2, COL3A1, SMAD3,TGFBR1, TGFBR2*)
VWF exon 28 sequencing*

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** Performed at LabPMM