

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) (C677T,AIZ98C)

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

Molecular Microbiology

Affirm (for vaginitis)

BK virus (FISH)

BK virus PCR, quantitative

Continued on back

* Validated, soon to be implemented

** Performed at LabPMM

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 pneumophila (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)
 Respiratory Virus Multiplex 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 genotyping (S and Z alleles)*
 Bacterial Sequence-Based Identification
BRAF for Hairy Cell Leukemia
COL3A1 exon sequencing
 Constitutional Gene Panel, v1
 FISH for Biliary Tract Neoplasia
 Fragile X syndrome (*FMR1*) DNA analysis (PCR)*
 HLA B5701, HLA B1502
 HLADQ2 and HLADQ8 for Celiac disease
KIT AML mutation analysis
KIT D816V PCR
KIT mutational analysis GIST & Melanoma*
MLH1 methylation
MPL mutation analysis
 Mycobacterial Identification/Resistant Determinants (NGS)
 NOD2 exon sequencing
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
SERPINA1 sequencing
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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