« فهرست آزمایشات انکولوژی به تفکیک بیماری »

Hematologic Malignancies

A. Common Tests

- **Karyotype:** Bone Marrow Karyotyping and Peripheral Blood Karyotyping (Greater than 10% Blasts)
- **Flow Cytometry:** All CD Markers according to differential diagnosis
- **Immunohistochemistry:** All CD Markers according to differential diagnosis

B. Special Tests by disease

Acute Lymphocytic Leukemia (ALL)

FISH Method

- BCR/ABL - t(9;22)(q34;q11.2)
- ETV6/RUNX1 - t(12;21)
- TCF3 (E2A) - 19p13 , t(1;19)
- KMT2A - 11q23 , t(v;11q23)
- IGH - 14q32 , t(5;14)
- CEP4, CEP10
- C-MYC - 8q24

Molecular Method

- BCR/ABL - t(9;22) Qualitative
- BCR/ABL - t(9;22) Quantitative
- MLL/AFF1 - t(4;11)
- ETV6/RUNX1 (TEL-AML1) - t(12;21)
- TCF3/PBX1 - t(1;19)

Acute Myeloid Leukemia (AML)

FISH Method

- BCR/ABL - t(9;22)(q34;q11.2)
- PML/PARα - t(15;17)
- RUNX1T1/RUNX1 - t(8;21)
- RPN1/MECOM - inv(3)
- CBFB/MYH - inv(16)
- KMT2A - 11q23 , t(9;11)
- ERG1 - del(5)
- D7S486/Cen7 - del(7)
- DEK-NUP214 - t(6;9)

Molecular Method

- AML/ETO - t(8;21)
- NPM1 Mutations
- FLT3 Mutations
- KIT Mutations
- BCR/ABL - t(9;22)
- PML-PARα - t(15-17)
- CEBPA Mutations
- DNMT3A Mutations
- IDH1/2 Mutations
- CBFB/MYH - inv(16)

Myeloproliferative Neoplasms (MPN)

FISH Method

- FIP1L1/PDGFRA - 4q12
- BCR/ABL1 - t(9;22)
- PDGFRB - 5q33.1
- FGFR1 - 8p12

Molecular Method

- MPL Mutations
- KIT Mutations
- SRSF2 Mutations
- U2AF1 Mutations
- JAK2 Mutations
- TP53 Mutation
- SF3B1 Mutations
- EZH2 Mutations
- CALR Mutation
- IDH1/2 Mutations
- ASXL1 Mutations
- DNM3TA Mutations

Hematologic Malignancies

Chronic Lymphocytic Leukemia (CLL)

FISH Method

- del (6q)
- del (11q)
- del (13q)
- del (17p)
- Trisomy 12

Molecular Method

- TP53 Sequencing
- PLCG2 Mutations
- NOTCH1 Mutations
- SF3B1 Mutations
- BIRC3 Mutations
- BTK Mutations

Chronic Myelogenous Leukemia (CML)

FISH Method

- BCR/ABL1 - t(9;22)(q34;q11.2)

Molecular Method

- BCR/ABL - t(9;22) Qualitative
- BCR/ABL - t(9;22) Quantitative
- BCR/ABL1 Mutations for response to TKIs therapy
- JAK2 Mutations

Eosinophilia

FISH Method

- FIP1L1- CHIC2-PDGFRA - 4q12
- FGFR1 - 8p12
- PDGFRB - 5q33
- CBFB - inv(16)

Myelodysplastic Syndrome (MDS)

FISH Method

- EGR1 - del(5)(q31)
- Cen8/MYC
- del(7)
- del(13q)
- del(17p)
- del(20q)

Lymphoma

FISH Method

- Burkitt: MYC - 8q24
- IGH/BCL2 - t(14;18)
- IGH rearrangement - 14q32
- ATM - del(11q)
- BCL2 - 18q21
- IGH/CCND1(BCL1) - t(11;14)
- MALT1 - 18q21
- BCL6 - 3q27
- ALK rearrangement (2p23)

Molecular Method

- BRAF V600E

Multiple Myeloma

FISH Method

- IGH/FGFR3 - t(4;14)
- IGH rearrangement - 14q32
- IGH/CCND3 - t(6;14)
- IGH/CCND1 - t(11;14)
- IGH/MAF - t(14;16)
- IGH/MAFB - t(14;20)
- TP53 - del(17p)
- C-MYC - 8q24
- 1q21 gain
- (+7)
- (+9)
- (+15)
- del(13q)

Thrombophilia Panel

- Prothrombin (Factor II) G20210A
- Factor V (Leiden) G1691A
- Factor V Leiden R2 haplotype (H1299R)
- Factor XIII (FXIII) V34L
- MTHFR (C677T)
- MTHFR (A1298C)
- PAI-1 Serpin E1

Gastrointestinal Cancers

- **KRAS/NRAS Mutations:** for response to anti-EGFR therapy drugs such as *Cetuximab* or *Panitumumab.*
- **BRAF V600E Mutation:** for response to anti-EGFR therapy
- **Microsatellite Instability (MSI) - PCR Method:** Lynch Syndrome (MLH1, MSH2, MSH6, PMS2)
- **Hereditary Non-Polyposis Colorectal Cancer (HNPCC) Mutations:** MLH1, MSH2, MSH6, PMS2
- **Familial Adenomatous Polyposis (FAP Mutations)**
- **Gastrointestinal Stromal Tumor:** for response to TKI therapy
  - C-KIT Mutations : Exons 9, 11, 13
  - PDGFRa Mutations: Exon 18
- **Hereditary Diffuse Gastric Cancer (HDGC) Mutations:** CDH1 Mutations
- **Mismatch Repair (MMR) - IHC Method:** Immunohistochemical Expression of MMR Proteins (MLH1, MSH2, MSH6 and PMS2)
- **Colorectal Cancer Panel - NGS (22 Genes):** APC - ATM - AXIN2 - BMPR1A - CDH1 - CHEK2 - EPCAM - FLCN - GREM1 - MLH1 - MSH2 - MSH3 - MSH6 MUTYH - NTHL1 - PMS2 - POLD1 - POLE - PTEN - SMAD4 - STK11 - TP53

Lung Cancer

- **EGFR Mutations**

  Exons 18, 19, 20, 21 to determine if EGFR-targeted tyrosine kinase inhibitors such as *Gefitinib* and *Erlotinib* may be beneficial for treating the tumor
- **EGFR T790M Mutation**
- **BRAF Mutation:** is associated with responsiveness to combined therapy with *Dabrafenib* and *Trametinib* for the treatment of patients with metastatic NSCLC
- **KRAS Mutations:** is associated with reduced responsiveness to EGFR TKI therapy
- **EML4 - ALK Rearrangement - FISH Method:** for response to *Crizotinib* and *Ceretinib* in metastatic NSCLC
- **ROS1 Rearrangement - FISH Method:** for treatment with the tyrosine kinase inhibitor *Crizotinib*
- **PD-L1 , IHC Method:** to detect PD-L1 protein expression in NSCLC tissue for response to Immunotherapy (*Pembrolizumab)*

Breast Cancer

- ER , PR , HER2, Ki 67 - IHC Method
- HER2-neu (by IHC or FISH Methods)
- MammaPrint *
- BluePrint **
- BRCA1 & BRCA2 Mutations - Gene Sequencing
- BRCA1 & BRCA2 Mutations - MLPA
- Breast and Ovarian Cancer Panel - NGS (18 Genes): ATM - BARD1 - BRCA1 - BRCA2 - BRIP1 - CDH1 - CHEK2 - MUTYH - NBN - NF1 - PALB2 PTEN - RAD51C - RAD51D - SDHB - SDHD - STK11 - TP53

Oncology Drug Toxicity

- **DPD Genotyping:** 5-FU Therapy Candidates
- **Irinotecan Toxicity:** Polymorphism of UGT1A1

* MammaPrint® is a 70–Gene Assay, in vitro diagnostic test service intended to assess a patient's risk for distant metastasis within five years, classifying the patients into high and low recurrence groups.

**BluePrint® is an in vitro diagnostic test intended for molecular subtyping the tumor into Luminal, Basal and HER2-subtypes. Molecular subtyping for determining long-term outcome and how a patient may respond to treatment.

Nervous System Cancers

FISH Method

- 1p19q Co-deletion
- RELA Fusion

Molecular Method

- IDH1/2 Mutations
- TERT Promoter
- H3F3A Mutation
- HIST1H3B Mutation
- BRAF Mutation
- ATRX Mutation

Sarcoma

FISH Method

- EWSR1 Rearrangement - t(11;22)
- FUS1 Rearrangement (16p11)
- SS18 Rearrangement - (18q11)
- DDIT3/CHOP Rearrangement (12q13)
- PDGFB/Col1A1 Rearrangement - t(17;22)
- ALK rearrangement - 2p23
- CDK4 - (12q13)
- MDM2 Amplification
- ETV6 Rearrangement
- FOXO1 - 13q14
- N-MYC Amplification

Molecular Method

- KIT Mutations
- PDGFRA Mutations
- NF1 Mutations
- CDKN2A Muations
- STAT6 Mutation
- NAB2 Mutation
- NCOA2 Mutations
- MYOD1 Mutation
- EED Mutations

array-CGH

- Trisomy 8, 20
- Loss of 5q21

Melanoma

Molecular Method

- CDKN2A Mutation
- TERT Mutation
- BAP1 Mutation
- BRAF Mutation
- KIT Mutation
- MCR1 Mutation
- CDK4 Mutation
- NRAS Mutations

IHC

- PD-L1

array-CGH

- CGH Analysis for chromosomal aberration including chromosome 6,7,9,10

Cancer Screening Panel - NGS

Disorders tested:

Breast cancer - Ovarian cancer - Uterine cancer - Colorectal cancer - Cutaneous melanoma - Gastric cancer - Pancreatic cancer - Prostate cancer
Renal cell cancer - Thyroid cancer

61 Genes by NGS

APC - ATM - AXIN2 - BAP1 - BARD1 - BMPR1A - BRCA1 - BRCA2 - BRIP1 - CDC73 - CDH1 - CDK4 - CDKN2A - CHEK2 - DICER1 – EPCAM FH – FLCN
GREM1 - HOXB13 – KIT – MAX - MEN1 – MET – MITF - MLH1 - MSH2 - MSH3 - MSH6 – MUTYH – NBN - NF1 - NF2 - NTHL1 PALB2 - PDGFRA - PMS2
POLD1 – POLE - PRKAR1A - PTCH1 – PTEN - RAD51C - RAD51D - RB1 – RET - SDHA - SDHAF2 – SDHB – SDHC SDHD - SMAD4 - SMARCA4 - SMARCB1
STK11 - TMEM127 - TP53 - TSC1 - TSC2 - VHL - WT1

تهران، شهرک غرب، میدان صنعت، ضلع غربی ایستگاه مترو صنعت، پلاک ۲
کد پستی: ۱۴۶۶۷-۱۳۷۱۳
۸۸۳۶۳۹۵۵ - ۸۸۳۷۰۸۳۸ فکس: ۸۸۰۸۳۵۷۵
وب سایت: www.irangenepath.com
اینستاگرام: Genetics.Center
ایمیل: lab@irangenepath.com