Cancer-related Mutation Analysis

Next Generation Gene Sequencing for Myeloid Testing

Assay Summary

IU Health Molecular Pathology Laboratory now offers high throughput sequencing for hot spot mutations found in clinically relevant cancer genes. In addition to a general panel of 54 genes, selected panels have been developed for a more tailored application in specific cancers. Comparing to single gene assay, these panels offer a more comprehensive and economic way to assess prognosis and/or treatment options for cancer patients at the initial diagnosis or at the relapse.

Orderable Name: Use IU Health Molecular Pathology requisition; Call 317.491.6417 for requisition. Panels include:

AML Mutations by NGS
ASXL1, CEBPA, DNMT3A, ETV6/TEL, FLT3, HRAS, IDH1, IDH2, KIT, KRAS, MLL, NPM1, NRAS, PHF6, RUNX1, TET2, TP53, WT1

MDS Mutations by NGS
ASXL1, ATRX, BCOR, BCORL1, ETV6/TEL, DNMT3A, EZH2, GNAS, IDH1, IDH2, RUNX1, SF3B1, SRSF2, TET2, TP53, U2AF1, ZRSR2

CML Mutations by NGS
ABL1

MPN Mutations by NGS
ASXL1, BRAF, CALR, CSF3R, EZH2, IKZF1, JAK2, JAK3, KDM6A, KIT, MPL, PDGRA, SETBP1, TET2

CMML Mutations by NGS
ASXL1, CBL, CBLB, CBLC, EZH2, RUNX1, TET2, TP53, SRSF2

JMML Mutations by NGS
CBL, CBLB, CBLC, HRAS, KRAS, NRAS, PTPN11

ALL Mutations by NGS
ABL1, CSF3R, FBXW7, IKZF1, JAK3, KDM6A, NOTCH1

CLL Mutations by NGS
MYD88, NOTCH1, SF3B1, TP53

Lymphoma/Myeloma Mutations by NGS
BRAF, CDKN2A, CSF3R, FBXW7, HRAS, KRAS, MYD88, NOTCH1, NRAS, SF3B1,TP53

Hematopoietic Neoplasms Mutations by NGS
ABL1, ASXL1, ATRX, BCOR, BCORL1, BRAF, CALR, CBL, CBLB, CBLC,CDKN2A, CEBPA, CSF3R, CUX1, DNMT3A, ETV6/TEL, EZH2, FBXW7, FLT3, GATA1, GATA2, GNAS, HRAS, IDH1, IDH2, IKZF1,JAK2, JAK3, KDM6A, KIT, KRAS, MLL, MPL, MYD88, NOTCH1, NPM1, NRAS, PDGFRA, PHF6, PTEN, PTPN11, RAD21, RUNX1,SETBP1, SF3B1, SMC1A, SMC3, SRSF2, STAG2, TET2, TP53, U2AF1, WT1, ZRSR2

Clinical Utility: This test is useful for the assessment of prognosis and/or treatment options for cancer patients at the initial diagnosis or at the relapse.

Clinical Information: The TruSight Myeloid Panel is a highly multiplexed targeted resequencing assay that provides a comprehensive assessment of 54 genes (tumor suppressor genes and oncogenic hotspots) in one test. This assay enables highly sensitive mutation detection within important genes including FLT3, NPM1, CEBPA, IDH1, IDH2, KIT, JAK2, CALR and MPL. Mutations in these genes are linked to hematological malignancies, focusing on leukemia and myeloproliferative disorders. Many of these genes that are tested have targeted therapies available. This test is useful for the assessment of prognosis and/or treatment options for individuals with cancers at initial diagnosis or at replase1.

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Method: The assay uses optimized oligonucleotide probes for sequencing mutational hotspots in 141 kilobases (kb) of target genomic sequence. Fifty-four genes are targeted with 568 amplicons in a highly multiplexed next generation sequencing reaction.

Specificity: Assay sequences targeted hotspots in these genes: ABL1, ASXL1, ATRX, BCOR, BCORL1, BRAF, CALR, CBL, CBLB, CBLC,CDKN2A, CEBPA, CSF3R, CUX1, DNMT3A, ETV6/TEL, EZH2, FBXW7, FLT3, GATA1, GATA2, GNAS, HRAS, IDH1, IDH2, IKZF1,JAK2, JAK3, KDM6A, KIT, KRAS, MLL, MPL, MYD88, NOTCH1, NPM1, NRAS, PDGFRA, PHF6, PTEN, PTPN11, RAD21, RUNX1,SETBP1, SF3B1, SMC1A, SMC3, SRSF2, STAG2, TET2, TP53, U2AF1, WT1, ZRSR2

Reference Range:

- Mutation not detected. No clinically actionable mutation was found.
- Mutation detected. Results will specify which clinically actionable mutation was found

Performing Laboratory: IUHPL Molecular Pathology

Performance Schedule: Once a week; TAT 7-10 working days

CPT Code: 81450

Specimen Requirements: -1 mL Peripheral Blood in EDTA tube, 1 mL Bone Marrow in EDTA tube, FFPE tissue (Formalin fixative only), cell block FNAs

Specimen Stability and Shipping: Transport peripheral blood and bone marrow refrigerated. Transport/Storage of slides at room temperature.

Causes for Rejection: Peripheral blood or bone marrow collected in Heparin. Excess necrosis for slides. Inadequate percentage tumor; poor DNA quality.