Cancer-related Mutation Analysis

Next Generation Gene Sequencing for Solid Tumors

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 48 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:

Lung cancer panel
AKT1, ALK, BRAF, EGFR, KRAS, MET, NRAS, PIK3CA, PTEN

Colon cancer panel
APC, AKT1, BRAF, KRAS, NRAS, PIK3CA, PTEN, SMAD4

Gastrointestinal stromal tumor (GIST) panel
BRAF, KIT, PDGFRA

Melanoma panel
BRAF, CTNNB1, GNA11, GNAQ, KIT, NRAS

Ovarian cancer panel
BRAF, KRAS, PIK3CA, PTEN

Thyroid cancer panel
KIT, BRAF, RET

Breast cancer panel
AKT1, ERBB2, PIK3CA

Oncology sequencing panel
ABL1, AKT1, ALK, APC, ATM, BRAF, CDH1, CDKN2A, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, FBXW7, FGFR1, FGFR2, FGFR3, FLT3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, JAK2, JAK3, KDR, KIT, KRAS, MET, MLH1, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RET, SMAD4, SMARCB1, SMO, SRC, STK11, TP53, VHL

Clinical Utility: This test is useful for the assessment of prognosis and/or treatment options for individuals with cancers at initial diagnosis or at replase1.

Clinical Information: The Oncology Sequencing Solid Tumor Panel is a highly multiplexed targeted resequencing assay for detecting somatic mutations across hundreds of mutational hotspots in cancer genomes. This assay enables highly sensitive mutation detection within 48 important genes that include BRAF, KRAS, and EGFR. Mutations in these genes are linked to many cancers including melanoma, colorectal, ovarian, GIST and lung cancer. 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.

Indiana University Health

IU Health Pathology Laboratory
350 W 11th Street
Indianapolis, IN 46202-4108
317-491-6000 Fax: 317-491-6001 800-433-0740
www.iuhealth.net/pathologylab

Method: Slides submitted for testing have the tumor tissue macro-dissected from the slides to isolate DNA. Then assay uses optimized oligonucleotide probes for sequencing mutational hotspots in > 35 kilobases (kb) of target genomic sequence. Forty-eight genes are targeted with 212 amplicons in a highly multiplexed next generation sequencing reaction. Positive clinically actionable mutations will be confirmed by independent assays.

Specificity: Assay sequences targeted hotspots in these genes: ABL1, AKT1, ALK, APC, ATM, BRAF, CDH1, CDKN2A, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, FBXW7, FGFR1, FGFR2, FGFR3, FLT3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, JAK2, JAK3, KDR, KIT, KRAS, MET, MLH1, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RET, SMAD4, SMARCB1, SMO, SRC, STK11, TP53, VHL

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: 81445

Specimen Requirements: Preferable primary tumor.
FFPE tissue (Formalin fixative only), cell block FNAs
For tissue resection: 1 H&E and 15 unstained slides
For a biopsy: 1 section on 1 slide for H&E plus 15 unstained slides with 3 sections/ slide

Specimen Stability and Shipping: Transport/Storage of slides at room temperature.

Causes for Rejection: Excess necrosis for slides. Inadequate percentage tumor; poor DNA quality and quantity.

Modified 3-8-2016