ARUP LABORATORIES

THE SOLID TUMOR MUTATION PANEL BY NEXT-GENERATION SEQUENCING

ARUP test code 2007991, detects hotspot mutation in 48 cancer-related genes, including *ABL1, BRAF, EGFR, ERBB2, JAK3, KIT, KRAS, NRAS, PDGFRA, PIK3CA, PTEN,* and *SMAD4*.

TEST HIGHLIGHTS

Specific somatic mutations have been discovered in multiple cancer-related genes; these mutations have diagnostic, therapeutic, and prognostic utility.

Next-generation sequencing dramatically reduces time to data, as it sequentially identifies small fragments of DNA across millions of strands simultaneously, enabling rapid sequencing of large stretches of DNA spanning entire genomes.

Utilizing next-generation sequencing, solid tumor samples can now be sequenced in a matter of hours and fully analyzed within two days.

A personalized mutational profile is useful, as response to targeted therapy is closely associated with the mutation status of the tumor.

For a full list of targeted mutations within the 48 genes, visit: http://www.aruplab.com/NGS-Oncology-Mutations

INDICATIONS FOR ORDERING

- Useful for prognosis and/or treatment of individuals with solid tumor cancers at initial diagnosis or with refractory disease

TEST DESCRIPTION

Test methodology

- DNA isolated from microdissected tumor tissue is amplified for mutational hotspot regions in 48 genes
- Mutation status determined by massively parallel sequencing (next-generation sequencing)

TESTS TO CONSIDER

Primary test

- Solid Tumor Mutation Panel by Next Generation Sequencing 2007991
  - Detects mutations in hotspot regions of 48 cancer-related genes

Related tests

Single-assay mutation detection by sequencing

- *BRAF* Codon 600 Mutation Detection by Pyrosequencing 2002498
- *EGFR* Mutation Detection by Pyrosequencing 2002440
- *KIT* Mutations, Melanoma 2002695
- *KRAS* Mutation Detection 0040248
- *NRAS* Mutation Detection by Pyrosequencing 2003123
- *PIK3CA* Mutation Detection 2004510

Mutation detection–multiple genes or reflex assays

- Gastrointestinal Stromal Tumor Mutation 2002674
  - *KIT* and *PDGFRA* mutation detection

- *IDH1* and *IDH2* Mutation Analysis, exon 4 2006444
- *KRAS* Mutation Detection with Reflex to *BRAF* Codon 600 Mutation Detection 2001932
  - Reflex assay

DISEASE OVERVIEW

Incidence

- All cancers in U.S.—473/100,000
- Deaths from cancer—179/100,000

Treatment issues

- Many of the genes tested have targeted therapies available

GENETICS

Genes–*ABL1, AKT1, ALK, APC, ATM, BRAF, CDH1, CDKN2A, CSF1R, CTNNB1, EGFR, ERBB2, ERBB4, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2, JAK3, KDR, KIT, KRAS, MET, MLH1, MPL, NOTCH1, NPM1, NRAS, PDGFRA, PIK3CA, PTEN, PTPN11, RB1, RET, SMAD4, SMARCB1, SMO, SRC, STK11, TP53, VHL*

Inheritance–somatic mutations

Mutations–a full list of targeted mutations within these genes can be found at www.aruplab.com/NGS-Oncology-Mutations

TEST INTERPRETATION

Analytical sensitivity–5% mutant alleles

Positive result

- Mutation in one or more of the 48 genes was detected
  - Clinical relevance (prognosis or therapy) will be correlated, if known

Negative result

- No mutations were detected

Limitations

- Not intended to detect minimal residual disease

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ARUP is a nonprofit enterprise of the University of Utah and its Department of Pathology.

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