Solid Tumor Mutation Panel by Next Generation Sequencing

Indications for Ordering

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

Test Description

- 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

- Aid in therapeutic decisions for solid tumor cancers
- Does not detect translocations

Related tests

Single-assay mutation detection by sequencing

- *BRAF* Codon 600 Mutation Detection by Pyrosequencing 2002498
- *BRAF* V600E Mutation Detection in Circulating Cell-Free DNA by Digital Droplet PCR 2013921
- *EGFR* Mutation Detection by Pyrosequencing 2002440
- *KIT* Mutations, Melanoma 2002695
- *KRAS* Mutation Detection 0040248
- *NRAS* Mutation Detection by Pyrosequencing 2003123
- Colon Cancer Gene Panel, Somatic 2011616

Mutation detection – multiple genes or reflex assays

- Gastrointestinal Stromal Tumor Mutation 2002674
  - Detects activating mutations in *KIT* and *PDGFRA*
- *IDH1* and *IDH2* Mutation Analysis, exon 4 2006444
- *KRAS* Mutation Detection with Reflex to *BRAF* Codon 600 Mutation Detection 2001932

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 the targeted regions of the above genes can be found on the ARUP website – Solid Tumor Mutation Panel by Next Generation Sequencing

Test Interpretation

Results

- Positive
  - Mutation in 1 or more of the 48 genes was detected
    - Clinical relevance (prognosis or therapy) will be correlated, if known
- Negative
  - No mutations were detected

Limitations

Not intended to detect minimal residual disease