Myeloid Malignancies Mutation Panel by Next Generation Sequencing

Indications for Ordering

Assess for single gene mutations, including substitutions and insertions and deletions that may have diagnostic, prognostic, and/or therapeutic significance in

- Acute myeloid leukemia
- Myelodysplastic syndromes (MDS)
- Myeloproliferative neoplasms (MPN)
- MDS/MPN overlap disorders such as chronic myelomonocytic leukemia

Test Description

Myeloid Malignancies Mutation Panel by Next Generation Sequencing

- Next generation sequencing (NGS) library construction from genomic DNA
- Enrichment for regions of interest by hybridization
- Massively parallel sequencing

Tests to Consider

Primary tests

Myeloid Malignancies Mutation Panel by Next Generation Sequencing 2011117

Myeloid Malignancies Somatic Mutation and Copy Number Analysis Panel 2012182

Related tests

- *CEBPA* Mutation Detection 2004247
- *NPM1* Mutation by PCR and Fragment Analysis 0040174
- *IDH1* and *IDH2* Mutation Analysis, exon 4 2006444
- *WT1* Mutation Detection by Sequencing 2005766
- *KIT* Mutations in AML by Fragment Analysis and Sequencing 2002437

Disease Overview

Diagnostic issues

- Genetic targets contained in panels are relevant across the spectrum of myeloid malignancies
- Identification of one or more clonal genetic abnormalities may aid in establishing the diagnosis of a neoplasm
- Identification of certain mutations or patterns of mutations may aid in diagnostic subclassification

Prognostic and treatment issues

- Certain mutations or patterns of mutations may have prognostic significance
- Certain mutations may allow for the use of targeted therapies

Genetics

Genes – *ASXL1, ASXL2, BCOR, BCORL1, BRAF, CALR, CBL, CEBPA, CSF3R, DNMT1, DNMT3A, EED, ELANE, ETNK1, ETV6, EZH2, FAM5C, FLT3, GATA1, GATA2, HNRNPK, IDH1, IDH2, JAK2, JAK3, KDM6A, KIT, KRAS, LUC7L2, MAP2K1, MLL, MPL, NOTCH1, NPM1, NRAS, NSD1, PHF6, PRPF40B, PRPF8, PTPN11, RAD21, RUNX1, SETBP1, SF1, SF3A1, SF3B1, SMC1A, SMC3, SRSF2, STAG2, SUZ12, TET2, TP53, U2AF1, U2AF2, WT1, ZRSR2*

Mutations

A full list of targeted regions within these genes can be found at the ARUP website – Myeloid Panel Coordinates (www.aruplab.com/myeloid-panel-coordinates)

Test Interpretation

Results

- Positive – a somatic mutation in one of the 57 tested genes was detected
  - Clinical relevance (diagnosis, prognosis, therapy) will be correlated, if known
- Negative result – no mutations were detected in the sequenced genes

Limitations

- Mutations may be present below the limit of detection
- Not intended to detect minimal residual disease