BCR-ABL1 Mutation Analysis for Tyrosine Kinase Inhibitor Resistance by Next Generation Sequencing
ARUP test code 2008420

BCR-ABL1 Mutation by NGS Source
Whole Blood

BCR-ABL1 Mutation by NGS Result

**Detected**

The following mutation was detected:

p.Thr315Ile, (c.944C>T); 98.4%

p.Thr315Ile has been reported to confer resistance to BCR-ABL1 tyrosine kinase inhibitors.

This result has been reviewed and approved by Jay Patel, M.D.

**BACKGROUND INFORMATION: BCR-ABL1 Mutation Analysis for Tyrosine Kinase Inhibitor Resistance by Next Generation Sequencing**

**CHARACTERISTICS:** This test is designed to detect mutations in the SH2, SH3 and tyrosine kinase domain of BCR-ABL1 fusions with the major or minor breakpoint that may impart resistance to tyrosine kinase inhibitor (TKI) therapy. The test spans ABL1 codons 46 - 542 and detects essentially all clinically actionable BCR-ABL1 kinase domain mutations, including T315I.

**METHODOLOGY:** Patient RNA was isolated, reverse transcribed into cDNA, and amplified across the BCR-ABL1 breakpoint using primers specific for the BCR and ABL1 genes. A sequencing library was then constructed from the resulting amplicons and sequencing performed on the Illumina NextSeq sequencing platform. Detected mutations were reported with their frequency.

**LIMITATIONS:** The next-generation sequencing technology utilized in this test allows for the detection of substitution mutations present at frequencies as low as 5 percent of the sequenced fusions. The sensitivity of this assay may be limited, and sequencing may not be possible in patient samples containing low tumor burden (ie, low levels of BCR-ABL1 fusion transcript by IS percent or NCN). This assay is not intended to be used for detection or quantification of BCR-ABL1 fusion transcripts. Results of this test must always be interpreted within the patient's clinical context and in conjunction with other relevant data. Results should not be used alone for a diagnosis of malignancy.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement B: aruplab.com/CS

H=High, L=Low, *=Abnormal, C=Critical
### Verified/Reported Dates

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