**The following ({{nvariants}} ) have been detected in this bone marrow specimen.**

**{% for variant in variant\_list %}**

**Gene: {{variant.gene}}**

**Mutation: {{variant.mutation}}**

**Allele Frequency (%):{{variant.allele\_frequency}}**

**Interpretation:** {{variant.interpretation}}

**{% endfor %}**

Intradepartmental consensus was obtained.

**The following 97 genes were tested on this panel and with the exception of the genes listed above; none of these contain any clinically significant mutations**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| ABL1 | BTK | CRLF2 | FOXO1 | JAK1 | MEF2B | PHF6 | RUNX1 | TCF3 |
| AKT1 | CALR | CSF1R | FOXP1 | JAK2 | MPL | PIAS2 | SETBP1 | TET2 |
| ARNTL | CARD11 | CSF3R | GATA1 | JAK3 | MYBL2 | PIK3R2 | SF3B1 | TNFAIP3 |
| ASXL1 | CBL | CUX1 | GATA2 | KDM6A | MYD88 | PLCG2 | SH2B3 | TNFRSF14 |
| ATM | CBLB | DNMT3A | GNA13 | KIF17 | NF1 | PRDM1 | SMC1A | TP53 |
| BCL2 | CCND1 | EP300 | HNRNPK | KIT | NOTCH1 | PRMT5 | SMC3 | U2AF1 |
| BCL6 | CD79B | ETV6 | HRAS | KLHL6 | NOTCH2 | PTEN | SOCS1 | WHSC1 |
| BCOR | CDKN2A | EZH2 | IDH1 | KMT2A | NOTCH3 | PTPN11 | SRSF2 | WT1 |
| BCORL1 | CEBPA | FAM5C | IDH2 | KMT2C | NPM1 | PTPRD | STAG2 | ZRSR2 |
| BIRC3 | CLSTN1 | FBXW7 | IKZF1 | KMT2D | NRAS | PTPRT | STAT3 |  |
| BRAF | CREBBP | FLT3 | IL7R | KRAS | PAX5 | RAD21 | SUZ12 |  |

**Additional Details on Mutation Identified**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Gene** | **Transcript** | **Genome** | **Chrom** | **Start** | **End** | **Ref** | **Variant** |
| {%tr for variant in variant\_tbl\_entries %} |  | | | | | | |
| {{ variant.gene}} | {{variant.transcript}} | {{variant.genome}} | {{variant.chrom}} | {{variant.start}} | {{variant.end}} | {{variant.ref}} | {{variant.variant}} |
| {%tr endfor %} |

**Methodology**

DNA is extracted and hybridized with custom-designed probes to enrich the targeted regions of 97 genes associated with hematologic malignancies. Samples are then sequenced on the Illumina HiSeq 2500 (Illumina, Inc, CA). A custom bioinformatics pipeline aligns the data to human reference genome GRCh37 to call variants. The limit of detection (related in part to depth of coverage, neoplastic cell percentage, and allelic frequency for the mutation) was determined to be 5% allele frequency, at which our assay has sensitivity of 98% and 91%, respectively, to detect single nucleotide variants (SNVs) and insertions/deletions (indels). Mutant allele populations below this detection limit will not be reliably detected by this method. Pseudogenes, highly homologous regions, and repeat regions may interfere with the detection of variants in this assay. This assay targets genes involved in hematologic malignancies. Some of the genes targeted may also cause inherited genetic disorders, variants in these genes will not be reported unless they are determined to contribute to the diagnosis, prognosis, or treatment of hematologic malignancies.

In addition to the mutations/variants reported above, several benign polymorphisms and variants of unknown/uncertain clinical significance have been detected. These variants can be made available upon request

This test was developed and its performance characteristics determined by the Clinical Molecular and Genomic Pathology Laboratory at the University of Kentucky. It has not been cleared or approved by the U.S. Food and Drug Administration. This test, which utilizes analyte specific reagents, does not require FDA approval. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) as qualified to perform high complexity clinical laboratory testing.

This service has been rendered in part by a resident. A pathologist has personally reviewed the test results and has rendered and is responsible for the diagnosis that appears on the report.