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HLID#: **PATIENT NAME:**
PATIENT ID#: **DOB :** **SEX:**
NPI: **ORDERING PHYSICIAN:**
SPECIMEN TYPE: Bone Marrow Aspirate
COLLECTION DATE: **RECEIPT DATE:**
REPORT DATE: **ICD Code:** C92.A0 **UNITS:**
CLINIC ID#:
CPT: 81450
ACCOUNT:

Last Name, First Name HLID#

MOLECULAR ANALYSIS REPORT

Specimen Type: Bone Marrow Aspirate (Client Pathology Number)

Clinical History/Indications: A patient with a clinical history of acute leukemia

AML (NGS) mutation panel results: NEGATIVE

Analysis/Conclusions:

- **No clinically significant, potentially clinically significant or variants of uncertain clinical significance were detected in this specimen.**
- *The presence of variants below the sensitivity limit (established at 2-5%) cannot be ruled out.*

The AML panel targets the following genes:
Genes marked in **bold** have full coding sequence (CDS) coverage; for further information on covered regions, including transcripts visit our website:

*ANKRD26, ASXL1, **BCOR**, **BCORL1**, **CEBPA**, CSF3R, DDX41, **DNMT3A**, **ETV6**, **EZH2**, FLT3 (ITD+TKD), GATA2, IDH1, IDH2, JAK2, KIT, KRAS, NPM1, NRAS, NF1, **PHF6**, PPM1D, PTPN11, **RAD21**, **RUNX1**, SETBP1, SF3B1, SRSF2, **STAG2**, **TET2**, **TP53**, U2AF1, WT1, **ZRSR2***

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Please note:

- **Currently classified benign variants detected by this assay are not reported.**
- Variant classification is based on current versions (date of the report) of the following public databases: NCBI Short Genetic Variations Database (dbSNP) and Clinically Relevant Sequence Variations (ClinVar), as well as literature and published guidelines. Clinical knowledge can change over time and variant classification can change in significance.
- Clinical and histological correlation required.

Method: Next-generation deep amplicon sequencing (NGS) was utilized to simultaneously detect mutations in selected genes and gene regions (coding sequence and splice region) recurrently reported mutated in AML.[1-3] Fastq files were aligned to hg19 version of the human genome and single-nucleotide and small indel calling was performed using the PiVAT software from Pillar Bioscience version: 2023.1.0. Secondary analysis was performed using NextGENe software v2.4.1 -also utilized to visualize reads. Variants were annotated utilizing Variant Studio v3.0.12 with cDNA and amino acid changes, number of reads supporting the variant allele and population allele frequency. The assay is validated to detect deletions up to 52 bp, and insertions to a size of approximately 100 bp. Orthogonal polymerase chain reaction combined with fragment length analysis is performed to detect FLT3 internal tandem duplications >100 bp in size. Gene fusions and intronic gene regions are not targeted by this assay.

References:

1. Pollyea et al. J Natl Compr Canc Netw 2023;21(5):503–513
2. Döhner et al. Blood 2022; 140 (12): 1345–1377
3. Huber, S., Baer, C., Hutter, S. et al. Leukemia. 2023; 37, 1413–1420

Although molecular testing is highly accurate, rarely false-positive and false-negative diagnostic errors may occur. DW/MRL/DX

Electronically signed by: Bettina Burnworth, Ph.D., Associate Director of Molecular Analysis - 01/01/20XX 12:00 PT; Michael H. Kalnoski, MD, FCAP - 01/01/20XX 12:05 PT