Acute Myeloid Leukemia (AML) Panel

Personal Information	Specimen Information	Test Information
Name: John Doe	Sample ID: 20210818-971-0000	Test reported: 2021-09-01
Date of Birth:	Medical record No:	Ordering physician:
Sex: Male	Date received: 2021-08-18	Institution: Hospital A

Cancer Type R/O AML

RESULT SUMMARY

	Tier 1	Tier 2	Tier 3
Variant	0	0	1
Gene	-	-	ETV6

TEST RESULT

Tier 1 : Variants of Strong Clinical Significance 0

No	Gene	DNA	Protein	VAF(%)	Depth(X)	COSMIC ID
No variant						

INTERPRETATION

A variant corresponding to Tier 1 (strong clinical significance) was not found.

Tier 2 : Variants of Potential Clinical Significance 0

No	Gene	DNA	Protein	VAF(%)	Depth(X)	COSMIC ID
No variant						

INTERPRETATION

A variant corresponding to Tier 2 (potential clinical significance) was not found.

Tier 3 : Variants of Unknown Clinical Significance 1

No	Gene	DNA	Protein	VAF(%)	Depth(X)	COSMIC ID
1	ETV6	c.496G>A	p.Val166Met	48	974	COSM6985046

INTERPRETATION

The c.496G>A (p.Val166Met) variant of the ETV6 gene has previously been reported in solid cancer(N=1, COSM6985046) but its clinical significance is unclear as it is unknown for the oncogenicity.

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TEST INFORMATION

1.TEST METHOD

Target Region	49 genes
Tested Panel	Acute Myeloid Leukemia (AML) Panel
Target Enrichment Method	Hybridization with oligonucleotide probes (Hema v2.0)
Massively Parallel Sequencing	MiSeqDX (150 bp x 2 paired-ends)
Bioinformatic Pipeline	BI_Hema_v2.0 (Alignment: BWA, Variant calling: VarScan2_GATK)
Reference Genome	GRCh37/hg19

2.QC DATA

Sample(DNA) QC	Pass	Mean Coverage of Depth(X)	1025X
Library QC	Pass	% of Target Bases ≥ 50X	99.9%
Sequencing QC	Pass		

3.TEST LIMITATIONS

This test was performed using sequencing analysis, and can detect SNP and small-indel variants within the analyzed region, but not structural variations such as copy number variation (CNV) and gene rearrangement.

The limit of detection for SNV and small-indel variants is approximately 5%.

The detected variants in this test are not re-confirmed by Sanger sequencing, ddPCR or other confirmation methods.

This test does not distinguish between germline and somatic variants. If the variant allele frequency of the mutation is close to 50% or 100%, the possibility of germline variant cannot be excluded.

The variants detected in this test are classified into four (tier 1~4) according to the 2017 JMD guideline (J Mol Diagn 2017;19:313-327), and tier 4 variants are not reported.

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4. CLASSIFICATIONS

Somatic Variants are classified into four stages according to the evidence level and clinical significance of the mutation.
Tier 4 is not reported.

Tier1	Strong clinical significance	Level A or B evidence
Tier2	Potential clinical significance	Level C or D evidence
Tier3	Unknown clinical significance	Not observed at a significant allele frequency in the general or specific subpopulation databases, or no convincing published evidence of cancer association.
Tier4	Benign or likely benign	Observed at significant allele frequency in the general databases. No existing published evidence of cancer association.

EVIDENCE LEVEL

LevelA	Biomarkers related to predicting therapeutic response or resistance to FDA-approved therapies in specific cancer types or biomarkers included in professional guidelines as being related to therapeutic response or resistance to drugs, diagnosis or prognosis of cancer.
LevelB	A biomarker with a consensus among experts in a well-designed study that is associated with the treatment response or resistance to a drug, the diagnosis or prognosis in a specific cancer type.
LevelC	Predictive biomarkers of therapeutic response or resistance to FDA-approved drugs in other cancer types or biomarkers eligible for clinical trial participation, and biomarkers reported to be associated with cancer diagnosis or prognosis in several small studies.
LevelD	Biomarkers with preclinical trials or small studies or several case reports.

REFERENCES

- COSMIC(http://cancer.sanger.ac.uk)
- c-bioportal(http://www.cbioportal.org)
- Cancer Hotspots(http://cancerhotspots.org)
- OncoKB(http://oncokb.org)
- My Cancer Genome(https://www.mycancergenome.org/)
- The Clinical Knowledgebase(https://ckb.jax.org/)
- WHO classification of tumours of haematopoietic and lymphoid tissues(revised 4th edition)
- NCCN guidelines®

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5. GENE INFORMATION

ESSENTIAL GENE LIST

Gene	Target Exon No.	Reference Transcript	Gene	Target Exon No.	Reference Transcript	Gene	Target Exon No.	Reference Transcript
CEBPA	All coding exons	NM_004364	FLT3	14-20	NM_004119	IDH1	4	NM_005896
IDH2	4	NM_002168	JAK2	12, 14	NM_004972	KIT	8-14, 17-18	NM_000222
NPM1	10-11	NM_002520	RUNX1	All coding exons	NM_001754	TP53	All coding exons	NM_000546

ADDITIONAL GENE LIST

Gene	Target Exon No.	Reference Transcript	Gene	Target Exon No.	Reference Transcript	Gene	Target Exon No.	Reference Transcript
ANKRD26	exon1-UTR upstream.1K	NM_014915	ASXL1	12	NM_015338	ATRX	8-10, 17-31	NM_000489
BCOR	All coding exons	NM_001123385	BCORL1	All coding exons	NM_021946	BRAF	15	NM_004333
CALR	All coding exons	NM_004343	CBL	8-9	NM_005188	CBLB	9-10	NM_170662
CSF3R	14, 17	NM_156039	DDX41	All coding exons	NM_016222	DNMT3A	All coding exons	NM_022552
ETV6	All coding exons	NM_001987	EZH2	All coding exons	NM_004456	GATA1	All coding exons	NM_002049
GATA2	All coding exons	NM_001145661	HRAS	2-3	NM_005343	JAK3	13	NM_000215
KDM6A	All coding exons	NM_001291415	KRAS	2-4	NM_004985	MPL	10	NM_005373
NOTCH1	26-28, 34	NM_017617	NRAS	2-4	NM_002524	PDGFRA	12, 14, 18	NM_006206
PHF6	All coding exons	NM_001015877	PPM1D	6	NM_003620	PTPN11	3-4, 12-13	NM_002834
RAD21	All coding exons	NM_006265	SETBP1	4 (799 ~ 965 codons)	NM_015559	SF3B1	6-8, 12-17	NM_012433
SMC1A	2, 11, 16-17	NM_006306	SMC3	All coding exons	NM_005445	SRSF2	1	NM_003016
STAG1	All coding exons	NM_005862	STAG2	All coding exons	NM_001042749	STAT3	20-21	NM_003150
TET2	All coding exons	NM_001127208	U2AF1	2, 6	NM_006758	WT1	2-10	NM_024426
ZRSR2	All coding exons	NM_005089						

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