SAMPLE REPORT

Acute Lymphoblastic Leukemia (ALL) Panel

Personal Information	Specimen Information	Test Information
Name:	Sample ID:	Test reported:
Date of Birth:	Medical record No:	Ordering physician:
Sex:	Date received:	Institution:

Cancer Type R/O ALL

RESULT SUMMARY

	Panel Result			Pharmacogene Result	
	Tier 1	Tier 2	Tier 3	NUDT15 Phenotype	TPMT Phenotype
Variant	0	0	3	Normal Metabolizer	Normal Metabolizer
Gene	-	-	DNM2, IDH2, TCF3		

TEST RESULT

Tier 1 : Variants of Strong Clinical Significance 0

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

INTERPRETATION

No tier 1 (Strong clinical importance) mutations were identified.

Tier 2 : Variants of Potential Clinical Significance 0

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

INTERPRETATION

No tier 2 (Potential clinical importance) mutations were identified.

Tier 3 : Variants of Unknown Clinical Significance 3

No	Gene	DNA	Protein	VAF(%)	Depth(X)	COSMIC ID
1	DNM2	c.931G>A	p.Val311Met	50	918	-
2	IDH2	c.673G>A	p.Asp225Asn	49	421	-
3	TCF3	c.1564A>C	p.Lys522Gln	52	848	-

INTERPRETATION

The c.931G>A (p.Val311Met) variatn in DNM2 gene has not been previously reported in hematologic malignancy and its clinical significance is unclear due to unknown oncogenicity of the variant.

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INTERPRETATION

The c.673G>A (p.Asp225Asn) variatn in IDH2 gene has not been previously reported in hematologic malignancy and its clinical significance is unclear due to unknown oncogenicity of the variant.

The c.1564A>C (p.Lys522Gln) variatn in TCF3 gene has not been previously reported in hematologic malignancy and its clinical significance is unclear due to unknown oncogenicity of the variant.

Pharmacogene result: NUDT15

Diplotype	Allele function status	Phenotype	EHR Priority Result
*1/*1	Normal function/Normal function	Normal Metabolizer	Normal/Routine/Low risk

INTERPRETATION

These results imply that the patient carries two alleles with normal function, and the genotyping findings predict that the patient is a NUDT15 Normal Metabolizer. The majority of drugs that are metabolized by NUDT15 do not require any special dosage adjustments. See the CPIC Guidelines at https://cpicpgx.org for more details.

Pharmacogene result: TPMT

Diplotype	Allele function status	Phenotype	EHR Priority Result
*1/*1	Normal function/Normal function	Normal Metabolizer	Normal/Routine/Low risk

INTERPRETATION

These results imply that the patient carries two alleles with normal function, and the genotyping findings predict that the patient is a TPMT Normal Metabolizer. The majority of drugs that are metabolized by TPMT do not require any special dosage adjustments. See the CPIC Guidelines at https://cpicpgx.org for more details.

* Reference: CPIC (Clinical Pharmacogenetics Implementation Consortium) (https://cpicpgx.org/)

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TEST INFORMATION

1. TEST METHOD

Target Region	50 genes
Tested Panel	Acute Lymphoblastic Leukemia (ALL) Panel
Target Enrichment Method	Hybridization with oligonucleotide probes (ALL 1.0)
Massively Parallel Sequencing	MiSeqDX (150 bp x 2 paired-ends)
Bioinformatic Pipeline	BI_ALL 1.0 (Alignment: BWA, Variant calling: VarScan2 & GATK)
Reference Genome	GRCh37/hg19

2. QC DATA

Sample(DNA) QC	Pass	**Mean Coverage of Depth(X)**	912X
Library QC	Pass	**% of Target Bases ≥ 50X**	98.8%
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
IKZF1	All coding exons	NM_006060.5	JAK2	16, 20-21, 24	NM_004972.3	NRAS	2-3	NM_002524.4
RB1	4, 9, 13, 20	NM_000321.2	TP53	All coding exons	NM_000546.5			

ADDITIONAL GENE LIST

Gene	Target Exon No.	Reference Transcript	Gene	Target Exon No.	Reference Transcript	Gene	Target Exon No.	Reference Transcript
ABL1	5-10	NM_005157.5	BRAF	11(G469), 15(600E, K601N)	NM_004333.4	BTG1	2	NM_001731.2
CDKN2A	All coding exons	NM_000077.4	CREBBP	4, 6, 14, 17-19, 21, 26-28, 31	NM_004380.2	CRLF2	6	NM_022148.3
DNM2	All coding exons	NM_001190716.1	DNMT3A	All coding exons	NM_022552.4	EP300	30	NM_001429.3
ETV6	All coding exons	NM_001987.4	EZH2	All coding exons	NM_004456.4	FBXW7	All coding exons	NM_033632.3
FLT3	5, 9, 13-16, 19-21, plus exons 13-15 (FLT3 -ITD)	NM_004119.2	GATA3	4-6	NM_002051.2	IDH1	4 (R132), 7	NM_005896.3
IDH2	All coding exons	NM_002168.3	IL7R	3, 5-6	NM_002185.3	JAK1	10, 13, 14-23	NM_002227.3
JAK3	2, 4, 10-13, 18-19	NM_000215.3	KDM6A	15, 25	NM_021140.2	KMT2A	All coding exons	NM_001197104.1
KMT2D	All coding exons	NM_003482.3	KRAS	2-4	NM_004985.4	LEF1	3-4	NM_016269.4
LMO1	All coding exons	NM_002315.2	MAPK1	4	NM_002745.4	NF1	9-10, 12, 18-19, 21, 23, 25, 28-29, 31, 33-34, 36 -38, 41, 44, 49, 52	NM_000267.3
NOTCH1	All coding exons	NM_017617.4	NT5C2	All coding exons	NM_001134373.2	NUDT15	1-3	NM_018283.3
PAX5	2(G24-V26), 3-5, 7, 9	NM_016734.2	PHF6	All coding exons	NM_001015877.1	PTEN	2, 5, 7	NM_000314.6
PTPN11	3, 8, 13	NM_002834.4	RUNX1	All coding exons	NM_001754.4	SETD2	All coding exons	NM_014159.6
SH2B3	All coding exons	NM_005475.2	STAG2	All coding exons	NM_001042749.1	STAT3	All coding exons	NM_139276.2
STAT5B	All coding exons	NM_012448.3	TBL1XR1	5-7, 9	NM_024665.4	TCF3	All coding exons	NM_001136139.2
TPMT	All coding exons	NM_000367.4	NSD2 (WHSC1)	18	NM_001042424.2	WT1	All coding exons	NM_024426.4