BCM Baylor College of Medicine
BioSequence

MEDICAL GENETICS LABORATORIES

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CANCER GENETICS LABORATORY

SAMPLE REPORT

Cancer Gene Mutation Panel by Next Generation Sequencing

Leukemia Mutation Panel

RESULTS **IDH1 c.394C>T (p.R132C) AND SRSF2 c.284C>T (p.P95L) MUTATIONS DETECTED**

INTERPRETATION:

We were requested to perform next generation sequence analysis for a panel of over 3,200 mutations in 48 key cancer genes associated with leukemia on the leukemic blood sample of this individual using the techniques described in methodology section below.

Our next generation sequencing analysis identified a c.394C>T (p.R132C) mutation of IDH1gene and c.284C>T (p.P95L) mutation of SRSF2 gene in the leukemic blood sample from the patient. Recent studies showed that IDH1 mutations were found in 7% patients with AML and R132 was the most common mutated amino acid in IDH1 gene. IDH1 mutations has been described to be associated with favorable clinical outcome (Patel JP et al., 2012). SRSF2 mutations are commonly seen in myeloid neoplasms with highest frequencies in CMML, followed by AML with MDS features and MDS subtypes RARS/RCMD-RS (Yoshida K et al., 2011). Recent studies have indicated that SRSF2 mutations are predictive for shorter survival (Makishima H, at al, 2011).

Patel JP et al., N Engl J Med. 2012 Mar 22;366(12):1079-89. Epub 2012 Mar 14. Yoshida K et al., Nature. 2011 Sep 11;478(7367):64-9.
Makishima H, et al., Blood. 2012 Apr 5;119(14):3203-10.

Mutation(s)

Gene	Nucleotide Change	Amino Acid Change	Exon	COSMIC ID	Reference(s) / Comments
IDH1	c.394C>T	p.R132C	4	28747	confirmed
SRSF2	c.284C>T	p.P95L	1	146288	confirmed

Benign Variant(s)

Gene	Nucleotide Change	Amino Acid Change	Exon	COSMIC ID	Reference(s) / Comments
PDGFRA	c.2472C	p.V824V	18	22413	db SNP: rs2228230

No Mutation Detected

ABL1	ASXL1	BRAF	CBL	CDKN2A	CEBPA	CREBBP	CRLF2
CSF1R	CTCF	DNM2	DNMT3A	EED	EP300	ETV6	EZH2
FBXW 7	FLT3	GATA1	HRAS	IDH2	IKZF1	IKZF3	IL7R
JAK2	JAK3	KIT	KRAS	MPL	NOTCH1	NPM1	NRAS
PAX5	PDGFRA	PHF6	PTEN	PTPN11	RELN	RUNX1	SF3B1
SUZ12	TAL1	TET2	TP53	U2AF1	WT1		

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Target below 100X

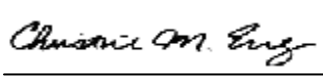
Gene	Target(s)
CDKN	p.M53, p.V51
FLT3	p.K614
RB1	E137,
RUNX	p.A60, p.D75, p.G69, p.H105, p.L56, p.M466, p.M52, p.R76, p.S100
SRSF	p.P95
W T1	p.H69

METHODOLOGY:

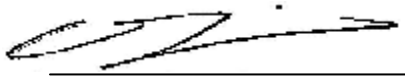
Genomic DNA extracted from this patient's sample was used for multiplex PCR amplification of 287 amplicons, which target over 3,000 mutations in 48 key cancer genes associated with leukemia, with the Ion AmpliSeq™ Kit. Next generation sequencing was performed on the Ion Torrent Personal Genome Machine and analyzed with the Torrent Suite Software. DNA sequences used as references for this panel of genes can be found at http://www.ncbi.nlm.nih.gov/refseq/rsg/. The mutation nomenclature is based on the convention recommended by the Human Genome Variation Society (http://www.hgvs.org/mutnomen/).

This mutation panel is designed to detect targeted mutations only. Other mutations in the 287 amplicons may not be detected. The 48 genes covered are not all sequenced in their entirety. Mutations outside the 287 amplicons will not be detected. The limit of detection is 5% at 500X coverage and 10% at 200X coverage. This technology cannot reliably detect mutations at coverage below 100X. Confirmation of mutations is performed by castPCR™ , pyrosequencing, or Sanger sequencing.

Individuals being studied should understand that rare diagnostic errors may occur. Possible sources of diagnostic errors include sample mix-ups and genotyping errors. Genotyping errors can result from trace contamination of PCR, from rare genetic variants which interfere with analysis, from mosaicism at levels below standard detection, and from other sources.



Christine M. Eng, M.D.
Medical Director



Liu Liu, Ph.D. FACMG
Assistant Laboratory Director

This test was developed and its performance determined by this laboratory. It has not been cleared or approved by U.S. Food and Drug Administration. Since FDA is not required for clinical use of this test, this laboratory has established and validated the test's accuracy and precision, pursuant to the requirement of CLIA '88. This laboratory is licensed and/or accredited under CLIA and CAP. (CAP# 2109314 / CLIA# 45D0660090)