Leukemia Sample
Jose Petty

Diagnosis
Acute lymphocytic leukemia (ALL)

Accession No.
9846

xT

Date of Birth
1969-10-10

Sex
Male

Physician
Dr. Carla Bird

Institution
Lee-Ramirez

Tumor specimen:
source Leukemia
CollectedDate 2023-08-10
ReceivedDate 2023-08-13
TumorPercentage 37%

Normal specimen:
source Blood
CollectedDate 2023-08-12
ReceivedDate 2023-08-13

GENOMIC VARIANTS

Somatic - Potentially Actionable		**variant allele fraction**
PRKCA	c.1387G>C p.D463H Spliceregionvariant-GOF	24.12%
JAK2	c.2792A>G p.M929I Nonsense-GOF	27.28%
Somatic - Biologically Relevant		
FGFR1	c.1966A>G p.K656E Nonsense-GOF	25.83%

Germline - Pathogenic

No Germline - Pathogenic variants were found in the limited set of genes on which we report.

Pertinent Negatives

FGFR2 IDH1 TOP2A

IMMUNOTHERAPY MARKERS

Tumor Mutational Burden

9 m/Mb 5%

Microsatellite Instability Status

Stable | Equivocal | High

FDA-APPROVED THERAPIES, Current Diagnosis

KRAS G12C Inhibitors	**Sotorasib**	NCCN, Consensus, Non-Small Cell Lung Cancer MSK OncoKB, Level 1 KRASp.G12C G12C-GOF

FDA-APPROVED THERAPIES, Other Indications

KRAS G12C Inhibitors	**Sotorasib**	NCCN, Consensus, Non-Small Cell Lung Cancer MSK OncoKB, Level 1 KRASp.G12C G12C-GOF

ADDITIONAL INDICATORS

Unfavorable Prognosis	NCCN, Consensus, Non-Small Cell Lung Cancer KRASp.G12C Gain-of-function

CLINICAL TRIALS

A Study of VS-6766 v. VS-6766 + Defactinib in Recurrent G12V, Other KRAS and BRAF Non-Small Cell Lung Cancer	**Phase 2** City, state - x mi **KRAS mutation**
A Phase 1/2 Study of MRTX849 in Patients With Cancer Having a KRAS G12C Mutation KRYSTAL-1	**Phase 1/2** City, state - x mi **KRAS mutation** **STK11 mutation**
First-in-human Study of DRP-104 (Sirpiglenastat) as Single Agent and in Combination With Atezolizumab in Patients With Advanced Solid Tumors. (NCT04471415)	**Phase 1/2** City, state - x mi **NFE2L2 mutation** **STK11 mutation**

VARIANTS OF UNKNOWN SIGNIFICANCE

Somatic	**Mutation effect**	**Variant allele fraction**
HSP90B1	c.3113A>G p.I66T Spliceregionvariant-LOF NM_001011645	8.77%
BLM	c.2207_2212delinsTAGATTC p.Y736fs*4 Nonsense-GOF NM_001011645	15.44%
PHF6	c.3113A>G p.R225X Nonsense-LOF NM_001011645	8.08%
MYCN	c.131_132delinsTT p.P44L Frameshift-GOF NM_001011645	1.52%
KLF1	c.892G>C p.M39L Missensevariant(exon2)-GOF NM_001011645	5.52%
PTCH1	c.3583A>T p.T1195S Frameshift-GOF NM_001011645	3.41%
Germline	**Mutation effect**	**Condition**
NOTCH1	c.7398_7399insAGGGACCG p.G1196D Nonsense-GOF NM_001011645	realize
KLF1	c.892G>C p.M39L Stopgain-LOF NM_001011645	deal

LOW COVERAGE REGIONS

SDHB

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

PRKCA c.1387G>C p.D463H Spliceregionvariant-GOF VAF: 24.12%

FAT1 encodes a transmembrane protein involved in tumor suppressor signaling. FAT1 protein can regulate transcriptional activity by sequestering beta-catenin, thereby preventing it from entering the nucleus. Loss of function mutations and copy number loss of FAT1 are associated with cancer progression.

JAK2 c.2792A>G p.M929I Nonsense-GOF VAF: 27.28%

TP53 encodes a protein that is a transcription factor that regulates the expression of genes involved in cell cycle arrest, apoptosis, and DNA repair. TP53 is a tumor suppressor gene that is mutated in many cancers. Mutations in TP53 are associated with cancer progression.

SOMATIC VARIANT DETAILS - BIOLOGICALLY RELEVANT

FGFR1 c.1966A>G p.K656E Nonsense-GOF VAF: 25.83%

FAT1 encodes a transmembrane protein involved in tumor suppressor signaling. FAT1 protein can regulate transcriptional activity by sequestering beta-catenin, thereby preventing it from entering the nucleus. Loss of function mutations and copy number loss of FAT1 are associated with cancer progression.

CLINICAL HISTORY

Diagnosed on

2023-08-10