Lymphoma Sample
Lisa Robinson

Diagnosis
Hodgkin lymphoma

Accession No.
8439

xT

Date of Birth
1970-04-29

Sex
Male

Physician
Dr. Lisa Garrett

Institution
Williams, Sherman and Carr

Tumor specimen:
source Lymphoma
CollectedDate 2023-07-30
ReceivedDate 2023-08-01
TumorPercentage 15%

Normal specimen:
source Blood
CollectedDate 2023-08-05
ReceivedDate 2023-08-05

GENOMIC VARIANTS

Somatic - Potentially Actionable		variant allele fraction
FOXL2	c.402C>G p.C134W Spliceregionvariant-LOF	5.64%
AKT1	c.49G>A p.E17K Missensevariant(exon2)-GOF	7.82%

Somatic - Biologically Relevant		
CSF3R	c.3113A>G p.G751A Nonsense-GOF	23.83%
CTNNB1	c.94G>A p.D32G Stopgain-LOF	4.13%

Germline - Pathogenic

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

Pertinent Negatives

MUTYH SRSF2 EZH2 DDR2

IMMUNOTHERAPY MARKERS

Tumor Mutational Burden

4 m/Mb 45%

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
MYOD1	c.365T>G p.L122R Spliceregionvariant-LOF NM_001011645	7.83%
PDGFRB	c.3113A>G p.T681I Spliceregionvariant-GOF NM_001011645	16.05%
ARID2	c.798G>A p.W266* Spliceregionvariant-LOF NM_001011645	6.51%
MSH2	c.1906G>C p.A636P Nonsense-GOF NM_001011645	5.65%
TP53	c.523C>T p.R213L Frameshift-LOF NM_001011645	9.0%
KIT	c.1676T>A p.Y846C Nonsense-GOF NM_001011645	5.54%
NRAS	c.181_182delinsTT p.G13X Frameshift-LOF NM_001011645	5.36%
TOP2A	c.3113A>G p.G1386D Nonsense-LOF NM_001011645	4.57%
FGFR2	c.758C>G p.N549H Stopgain-LOF NM_001011645	1.96%
NCSTN	c.3113A>G p.A572G Spliceregionvariant-LOF NM_001011645	7.42%
PIK3CA	c.1633G>A p.H1047Q Spliceregionvariant-LOF NM_001011645	8.87%
STAT5B	c.1924A>C p.N713_A714insKGKGGG Stopgain-LOF NM_001011645	9.42%

LOW COVERAGE REGIONS

FOXL2 PDGFRA

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

FOXL2 c.402C>G p.C134W Spliceregionvariant-LOF VAF: 5.64%

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.

AKT1 c.49G>A p.E17K Missensevariant(exon2)-GOF VAF: 7.82%

PTEN encodes a phosphatase that acts as a tumor suppressor by negatively regulating the PI3K-AKT-mTOR pathway. Loss of function mutations, copy number loss, and underexpression of PTEN are associated with cancer progression.

SOMATIC VARIANT DETAILS - BIOLOGICALLY RELEVANT

CSF3R c.3113A>G p.G751A Nonsense-GOF VAF: 23.83%

BCL11B encodes a C2H2-type zinc finger protein that functions as a transcriptional repressor and plays a role in T-cell development and survival. Loss of function mutations, copy number loss, and fusions resulting in the underexpression of BCL11B are associated with cancer progression.

CTNNB1 c.94G>A p.D32G Stopgain-LOF VAF: 4.13%

PTEN encodes a phosphatase that acts as a tumor suppressor by negatively regulating the PI3K-AKT-mTOR pathway. Loss of function mutations, copy number loss, and underexpression of PTEN are associated with cancer progression.

CLINICAL HISTORY

Diagnosed on

2023-07-29