Lymphoma Sample
Sarah Hall

Diagnosis
Hodgkin lymphoma

Accession No.
5067

xT

Date of Birth
1968-05-23

Sex
Male

Physician
Dr. Paul Thompson

Institution
Perez Group

Tumor specimen:
source Lymphoma
CollectedDate 2024-01-01
ReceivedDate 2024-01-01
TumorPercentage 72%

Normal specimen:
source Blood
CollectedDate 2024-01-06
ReceivedDate 2024-01-09

GENOMIC VARIANTS

Somatic - Potentially Actionable		variant allele fraction
MYD88	c.794T>C p.L265P Nonsense-LOF	9.58%
BRCA2	c.5946delT p.S1982Rfs*22 Stopgain-LOF	15.84%
MYCN	c.131C>T p.P44L Missensevariant(exon2)-GOF	19.83%

Somatic - Biologically Relevant		
JAK3	c.3113A>G p.L857P Frameshift-GOF	13.22%
ABL1	c.3113A>G p.L857P Frameshift-LOF	11.9%

Germline - Pathogenic

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

Pertinent Negatives

HDAC1 PTPN11 ERBB2

IMMUNOTHERAPY MARKERS

Tumor Mutational Burden

26 m/Mb 50%

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**
NCSTN	c.3113A>G p.A572G Nonsense-LOF NM_001011645	9.21%
HBA2	c.427T>C p.*143Qext*31 Frameshift-GOF NM_001011645	10.38%
FGFR1	c.1966A>G p.K656E Frameshift-GOF NM_001011645	5.53%
HDAC1	c.3113A>G p.Y303H Frameshift-GOF NM_001011645	5.11%
KLF1	c.892G>C p.M39L Stopgain-LOF NM_001011645	3.02%
BRCA1	c.68_69delAG p.E23Vfs*17 Missensevariant(exon2)-GOF NM_001011645	5.62%
MET	c.3688T>C p.Y1230H Nonsense-LOF NM_001011645	2.51%
KLF1	c.892G>C p.A298P Nonsense-LOF NM_001011645	4.25%

Germline	**Mutation effect**	**Condition**
MTOR	c.5664C>A p.F1888L Missensevariant(exon2)-GOF NM_001011645	doctor
ZEB2	c.3113A>G p.H1038R Missensevariant(exon2)-GOF NM_001011645	decade

LOW COVERAGE REGIONS

ALK MAPK1

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

MYD88 c.794T>C p.L265P Nonsense-LOF VAF: 9.58%

RBM10 encodes a protein that contains a RNA-binding motif and interacts with RNA homopolymers, and is thought to function in regulating alternative splicing. Loss of function mutations and copy number loss of RBM10 are associated with cancer progression.

BRCA2 c.5946delT p.S1982Rfs*22 Stopgain-LOF VAF: 15.84%

ARID2 encodes a protein that is a subunit of the SWI/SNF chromatin remodeling complex SWI/SNF-B or PBAF. This complex functions in ligand-dependent transcriptional activation. Loss of function mutations and copy number loss of ARID2 are associated with cancer progression.

MYCN c.131C>T p.P44L Missensevariant(exon2)-GOF VAF: 19.83%

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.

SOMATIC VARIANT DETAILS - BIOLOGICALLY RELEVANT

JAK3 c.3113A>G p.L857P Frameshift-GOF VAF: 13.22%

RBM10 encodes a protein that contains a RNA-binding motif and interacts with RNA homopolymers, and is thought to function in regulating alternative splicing. Loss of function mutations and copy number loss of RBM10 are associated with cancer progression.

ABL1 c.3113A>G p.L857P Frameshift-LOF VAF: 11.9%

RBM10 encodes a protein that contains a RNA-binding motif and interacts with RNA homopolymers, and is thought to function in regulating alternative splicing. Loss of function mutations and copy number loss of RBM10 are associated with cancer progression.

CLINICAL HISTORY

Diagnosed on

2023-12-27