Lymphoma Sample
Bryan Bishop

Diagnosis
Hodgkin lymphoma

Accession No.
8371

xT

Date of Birth
1985-09-14

Sex
Female

Physician
Dr. Laura Ballard

Institution
Mcintosh, Garcia and Davis

Tumor specimen:
source Lymphoma
CollectedDate 2023-07-16
ReceivedDate 2023-07-17
TumorPercentage 26%

Normal specimen:
source Blood
CollectedDate 2023-07-23
ReceivedDate 2023-07-23

GENOMIC VARIANTS

Somatic - Potentially Actionable		variant allele fraction
MPL	c.1543_1545delinsAAA p.W515L Stopgain-LOF	10.36%
TP53	c.742C>T p.K132N Spliceregionvariant-LOF	7.97%
ABL1	c.3113A>G p.A34V Spliceregionvariant-GOF	27.51%
BRCA2	c.5946delT p.S1982Rfs*22 Missensevariant(exon2)-GOF	1.82%
Somatic - Biologically Relevant		
MSH2	c.1906G>C p.S1982Rfs*22 Spliceregionvariant-LOF	17.94%
BLM	c.2207_2212delinsTAGATTC p.S1982Rfs*22 Spliceregionvariant-LOF	5.16%

Germline - Pathogenic

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

Pertinent Negatives

APC CCND3 CALR

IMMUNOTHERAPY MARKERS

Tumor Mutational Burden

18 m/Mb 23%

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**
CALR	c.1154_1155insTTGTC p.K385fs*47 Missensevariant(exon2)-GOF NM_001011645	11.11%
FGFR4	c.1162G>A p.G388R Frameshift-LOF NM_001011645	1.93%
EGFR	c.2310_2311ins9 p.D770_N771insD Frameshift-LOF NM_001011645	10.31%
PKLR	c.1436G>A p.R479H Nonsense-LOF NM_001011645	3.76%
MYD88	c.794T>C p.L265P Nonsense-GOF NM_001011645	3.63%
DICER1	c.5428G>C p.E1705V Frameshift-LOF NM_001011645	3.55%
BRCA1	c.5265_5266insC p.Q1756Pfs Stopgain-LOF NM_001011645	3.22%
MTOR	c.5664C>A p.G1954R Nonsense-LOF NM_001011645	7.15%
Germline	**Mutation effect**	**Condition**
CHD6	c.4800C>G p.I1600M Spliceregionvariant-LOF NM_001011645	drive

LOW COVERAGE REGIONS

CDKN2A

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

MPL c.1543_1545delinsAAA p.W515L Stopgain-LOF VAF: 10.36%

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.

TP53 c.742C>T p.K132N Spliceregionvariant-LOF VAF: 7.97%

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ABL1 c.3113A>G p.A34V Spliceregionvariant-GOF VAF: 27.51%

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.

BRCA2 c.5946delT p.S1982Rfs*22 Missensevariant(exon2)-GOF VAF: 1.82%

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

MSH2 c.1906G>C p.S1982Rfs*22 Spliceregionvariant-LOF VAF: 17.94%

KRAS is a GDP/GTP binding protein that acts as an intracellular signal transducer. KRAS is involved in several pathways involved in cellular proliferation and survival, including the PI3K-AKT-mTOR pathway and the Ras-Raf-MEK-ERK pathway. Activating mutations, copy number gains, and overexpression of KRAS are associated with cancer progression.

BLM c.2207_2212delinsTAGATTC p.S1982Rfs*22 Spliceregionvariant-LOF VAF: 5.16%

KRAS is a GDP/GTP binding protein that acts as an intracellular signal transducer. KRAS is involved in several pathways involved in cellular proliferation and survival, including the PI3K-AKT-mTOR pathway and the Ras-Raf-MEK-ERK pathway. Activating mutations, copy number gains, and overexpression of KRAS are associated with cancer progression.

CLINICAL HISTORY

Diagnosed on

2023-07-15