Lung Sample
Alexandra Oconnor

Diagnosis
Adenocarcinoma

Accession No.
1918

xT

Date of Birth
1968-03-09

Sex
Female

Physician
Dr. Duane Adams

Institution
Winters LLC

Tumor specimen:
source Lung
CollectedDate 2023-05-01
ReceivedDate 2023-05-01
TumorPercentage 30%

Normal specimen:
source Blood
CollectedDate 2023-05-02
ReceivedDate 2023-05-09

GENOMIC VARIANTS

Somatic - Potentially Actionable — **variant allele fraction**

SMO	c.3113A>G p.A374E Stopgain-LOF	39.39%
HDAC1	c.3113A>G p.Y303H Frameshift-LOF	28.89%

Somatic - Biologically Relevant

U2AF1	c.3113A>G p.R156X Missensevariant(exon2)-GOF	27.05%

Germline - Pathogenic

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

Pertinent Negatives

HDAC2 PKLR SDHA PTEN ABL1

IMMUNOTHERAPY MARKERS

Tumor Mutational Burden

24 m/Mb 92%

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**
IKZF1	c.3113A>G p.N159Y Nonsense-GOF NM_001011645	13.64%
RNF43	c.954G>T p.A169T Nonsense-LOF NM_001011645	17.84%
CHD6	c.4800C>G p.I1600M Nonsense-GOF NM_001011645	9.95%
IKZF1	c.3113A>G p.N159Y Nonsense-LOF NM_001011645	1.41%
HSD3B1	c.1100C>A p.T367= Frameshift-GOF NM_001011645	2.03%
MAP2K2	c.383C>T p.P128L Nonsense-LOF NM_001011645	4.35%
GNAS	c.374A>G p.R201S Nonsense-LOF NM_001011645	9.04%
MYO1G	c.145G>A p.V49M Frameshift-GOF NM_001011645	9.62%

Germline	**Mutation effect**	**Condition**
CTNNB1	c.97T>C p.S45P Stopgain-LOF NM_001011645	technology
TP53	c.638G>A p.V272A Nonsense-LOF NM_001011645	rise

LOW COVERAGE REGIONS

CSF3R BRCA2 ARID2

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

SMO c.3113A>G p.A374E Stopgain-LOF VAF: 39.39%

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.

HDAC1 c.3113A>G p.Y303H Frameshift-LOF VAF: 28.89%

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

U2AF1 c.3113A>G p.R156X Missensevariant(exon2)-GOF VAF: 27.05%

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

2023-04-30