Pancreatic Sample
Omar Hanson

Diagnosis
Adenocarcinoma

Accession No.
8629

xT

Date of Birth
1996-10-02

Sex
Female

Physician
Dr. Derek Cooley

Institution
Thomas and Sons

Tumor specimen:
source Pancreatic
CollectedDate 2024-01-26
ReceivedDate 2024-01-26
TumorPercentage 56%

Normal specimen:
source Blood
CollectedDate 2024-01-29
ReceivedDate 2024-01-31

GENOMIC VARIANTS

Somatic - Potentially Actionable		variant allele fraction
ALDH2	c.1510G>A p.E504K Missensevariant(exon2)-GOF	5.46%
STAG2	c.3113A>G p.R1012X Nonsense-LOF	33.74%
AKT1	c.49G>A p.E17K Missensevariant(exon2)-GOF	22.36%
Somatic - Biologically Relevant		
ARID2	c.798G>A p.W266* Nonsense-GOF	23.86%

Germline - Pathogenic

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

Pertinent Negatives

FGFR2 NCSTN ERCC2 IKZF1

IMMUNOTHERAPY MARKERS

Tumor Mutational Burden

23 m/Mb 63%

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**
DDR2	c.716T>G p.I638F Frameshift-GOF NM_001011645	13.45%
EGFR	c.2235_2252del18 p.G719A Spliceregionvariant-LOF NM_001011645	16.87%
DPYD	c.2846A>T p.I543V Spliceregionvariant-LOF NM_001011645	3.81%
ERBB2	c.2264_2278del p.S310F Frameshift-GOF NM_001011645	1.41%
AKT1	c.49G>A p.E17K Missensevariant(exon2)-GOF NM_001011645	5.4%
STAG2	c.3113A>G p.R1012X Stopgain-LOF NM_001011645	4.35%
Germline	**Mutation effect**	**Condition**
FBXW7	c.1394G>A p.R465H Stopgain-LOF NM_001011645	catch

LOW COVERAGE REGIONS

ARID2

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

ALDH2 c.1510G>A p.E504K Missensevariant(exon2)-GOF VAF: 5.46%

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.

STAG2 c.3113A>G p.R1012X Nonsense-LOF VAF: 33.74%

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.

AKT1 c.49G>A p.E17K Missensevariant(exon2)-GOF VAF: 22.36%

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

ARID2 c.798G>A p.W266* Nonsense-GOF VAF: 23.86%

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.

CLINICAL HISTORY

Diagnosed on

2024-01-24