

Date of Birth  
1953-07-11

Sex  
Female

Physician  
Dr. Kathryn Frost

Institution  
Hunter, Williams and Walker

Tumor specimen:  
source Pancreatic  
CollectedDate 2023-05-22  
ReceivedDate 2023-05-23  
TumorPercentage 60%

Normal specimen:  
source Blood  
CollectedDate 2023-05-23  
ReceivedDate 2023-05-24

GENOMIC VARIANTS

Somatic - Potentially Actionable		variant allele fraction	
<b>MET</b>	c.3209T>G p.Y1230C Nonsense-GOF	16.4%	<div></div>
Somatic - Biologically Relevant			
<b>CHEK2</b>	c.1283C>T p.L1205V Nonsense-GOF	10.81%	<div></div>
<b>PIK3CA</b>	c.1624G>C p.S428F Stopgain-LOF	4.22%	<div></div>

Germline - Pathogenic

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

Pertinent Negatives

**ARID2**

IMMUNOTHERAPY MARKERS

Tumor Mutational Burden	Microsatellite Instability Status		
<b>9 m/Mb</b> 5%	<b>Stable</b>	Equivocal	High

FDA-APPROVED THERAPIES, Current Diagnosis

KRAS G12C Inhibitors	<b>Sotorasib</b>	NCCN, Consensus, Non-Small Cell Lung Cancer MSK OncoKB, Level 1 KRASp.G12C G12C-GOF
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FDA-APPROVED THERAPIES, Other Indications

KRAS G12C Inhibitors	<b>Sotorasib</b>	NCCN, Consensus, Non-Small Cell Lung Cancer MSK OncoKB, Level 1 KRASp.G12C G12C-GOF
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ADDITIONAL INDICATORS

Unfavorable Prognosis	NCCN, Consensus, Non-Small Cell Lung Cancer KRASp.G12C Gain-of-function
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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
MAP2K2	c.383C>T p.P128L Missensevariant(exon2)-GOF NM_001011645	10.37% 
NT5C2	c.1100G>A p.R367Q Frameshift-GOF NM_001011645	11.12% 
PHF6	c.3113A>G p.R225X Nonsense-GOF NM_001011645	6.78% 
HSP90B1	c.3113A>G p.I66T Spliceregionvariant-LOF NM_001011645	6.16% 
FBXW7	c.1394G>A p.R465H Missensevariant(exon2)-GOF NM_001011645	3.01% 
CD74	c.3113A>G p.P98S Stopgain-LOF NM_001011645	3.42% 
FGFR3	c.1118A>G p.R248C Spliceregionvariant-LOF NM_001011645	8.43% 
BRCA2	c.5946delT p.S1982Rfs*22 Frameshift-GOF NM_001011645	7.83% 
DNMT3A	c.2711C>T p.P904L Stopgain-LOF NM_001011645	1.5% 
PTPN11	c.1508G>C p.G503A Missensevariant(exon2)-GOF NM_001011645	5.07% 
MTOR	c.5662T>C p.F1888L Frameshift-LOF NM_001011645	7.91% 
MTOR	c.5662T>C p.F1888L Missensevariant(exon2)-GOF NM_001011645	4.59% 

LOW COVERAGE REGIONS

HBB XPC HBA2

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

MET

c.3209T>G p.Y1230C Nonsense-GOF

VAF: 16.4%

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.

SOMATIC VARIANT DETAILS - BIOLOGICALLY RELEVANT

CHEK2

c.1283C>T p.L1205V Nonsense-GOF

VAF: 10.81%

NFE2L2 acts as a transcription factor for proteins that contain an antioxidant response element (ARE) within their promoter sequence. Genes that contain ARE are involved in injury and inflammation response. Activating mutations and overexpression of NFE2L2 are associated with cancer progression.

PIK3CA

c.1624G>C p.S428F Stopgain-LOF

VAF: 4.22%

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.

CLINICAL HISTORY

Diagnosed on  
2023-05-21