Breast Sample
Jason Summers

Diagnosis
Invasive ductal carcinoma

Accession No.
4989

xT

Date of Birth
1995-02-28

Sex
Female

Physician
Dr. Patricia Burgess MD

Institution
Henry LLC

Tumor specimen:
source Breast
CollectedDate 2023-11-28
ReceivedDate 2023-11-28
TumorPercentage 10%

Normal specimen:
source Blood
CollectedDate 2023-11-30
ReceivedDate 2023-11-30

GENOMIC VARIANTS

Somatic - Potentially Actionable		variant allele fraction
BLM	c.2207_2212delinsTAGATTC p.Y736fs*4 Frameshift-GOF	29.51%
PDGFRB	c.3113A>G p.T681I Spliceregionvariant-LOF	7.47%
CHD6	c.4800C>G p.I1600M Spliceregionvariant-LOF	11.65%

Somatic - Biologically Relevant		
ARID2	c.798G>A p.I1600M Nonsense-LOF	15.67%

Germline - Pathogenic

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

Pertinent Negatives

ERCC2 KIT KLF4 FBXW7

IMMUNOTHERAPY MARKERS

Tumor Mutational Burden

46 m/Mb 53%

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
ALDH2	c.1510G>A p.E504K Nonsense-LOF NM_001011645	24.57%
RNF43	c.505G>A p.E318D Missensevariant(exon2)-GOF NM_001011645	15.53%
MYO1G	c.145G>A p.V49M Stopgain-LOF NM_001011645	2.42%
CSF3R	c.3113A>G p.G751A Spliceregionvariant-GOF NM_001011645	4.39%
FGFR2	c.755C>G p.C382R Nonsense-LOF NM_001011645	5.58%
EGFR	c.2239_2256del18 p.L747_P753del Frameshift-LOF NM_001011645	2.96%
ALDH2	c.1510G>A p.E504K Frameshift-LOF NM_001011645	7.78%
CDC73	c.1A>G p.M1V Spliceregionvariant-GOF NM_001011645	5.71%
IDH2	c.419G>A p.R172K Nonsense-LOF NM_001011645	7.38%

LOW COVERAGE REGIONS

BRCA1

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

BLM c.2207_2212delinsTAGATTC p.Y736fs*4 Frameshift-GOF VAF: 29.51%

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.

PDGFRB c.3113A>G p.T681I Spliceregionvariant-LOF VAF: 7.47%

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.

CHD6 c.4800C>G p.I1600M Spliceregionvariant-LOF VAF: 11.65%

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

ARID2 c.798G>A p.I1600M Nonsense-LOF VAF: 15.67%

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

2023-11-26