Final Report

Patient

Name: Alexandra Hawkins
Date of Birth: 1959-05-26
Sex: Female
Case Number: 6167
Diagnosis: Merkel cell carcinoma

Specimen Information

Primary Tumor Site: Skin
Specimen Site: Skin (site of visible lesion)
Specimen ID: 4784
Specimen Collected: 2024-02-04
Test Initiated: 2024-02-06

Ordered By

Biomarker	Method	Analyte	Result	Therapy association		Biomarker level
ER	IHC	protein	Positive | 3+, 100%	BENEFIT	abemaciclib, palbociclib, ribociclib, endocrine, therapy, everolimus	level 2
PR	IHC	protein	Positive | 2+, 95%	BENEFIT	abemaciclib, palbociclib, ribociclib, endocrine therapy	level 2
TMB	seq	DNA tumor	9 m/Mb Low	BENEFIT	pembrolizumab	level 2
ERBB2	IHC	Protien	Negative | 0	LACK OF BENEFT	trastuzumab, ado-trastuzumab emtansine, pertuzumab, fam-trastuzumab deruxtecan-nxki, lapatinib, neratinib, tucatinib	level 1

Cancer-Type Relevant Biomarkers

BioMarker	Method	Analyte	Result
ER	IHC	Protien	Positive| 1+, 88%
ZEB2	Seq	DNA-Tumor	Stable
NRAS	Seq	DNA-Tumor	Fusion not detected
SIX1	Seq	DNA-Tumor	Stable

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	High
Tumor mutational burden	Seq	DNA tumor	9 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	Low - 5% of tested genmoic segments exhibit LOH

Genes Tested with Pathogenic Alterations or likely Pathogenic Alterations

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
KLF1	Seq	DNA tumor	Likely Benign	p.M1R	12	c.2T>G	35.1
BRCA2	Seq	DNA tumor	Likely Pathogenic	p.W515L	18	c.1543_1545delinsAAA	28.2
TEK	Seq	DNA tumor	Pathogenic	p.E27K	10	c.19G>A	16.96
U2AF1	Seq	DNA tumor	Likely Benign	p.V49M	2	c.145G>A	21.39
CDC73	Seq	DNA tumor	Likely Pathogenic	p.S1982Rfs*22	10	c.5946delT	1.33

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
PDGFRB	Seq	DNA tumor	Variant of uncertain significance	p.G751A	5	c	15.68
FGFR3	Seq	DNA tumor	Variant of uncertain significance	p.V777L	12	c.2263_2264delinsCC	10.74
ARID2	Seq	DNA tumor	Variant of uncertain significance	p.A34V	11	3	3.8
RB1	Seq	DNA tumor	Variant of uncertain significance	p.N452D	10	c.1354A>G	9.33
BLM	Seq	DNA tumor	Variant of uncertain significance	p.L857P	5	1	4.6
BRCA2	Seq	DNA tumor	Variant of uncertain significance	p.W515L	7	c.1543_1545delinsAAA	2.51

Immunohistochemistry Results

Biomarker	Result
MLH1	Positive| 3+, 85%
MSH6	Negative| 3+, 64%
ER	Positive| 3+, 53%
AR	Negative| 2+, 99%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

GNAS STAG2 HSP90B1 HDAC1 BRCA2 BRCA2

Specimen Information

Specimen ID: 4784
Specimen Collected: 2024-02-04
Specimen Recieved: 2024-02-06
Testing Initiated: 2024-02-06
Gross Description: 4784

Pathological Diagnosis:

Left breast, central, 12:00, suspicious mass, 12-gauge core needle biopsy: Infiltrating moderately-differentiated mammary carcinoma, grade 2, Nottingham score 6 (architectural grade 3, nuclear grade 2, mitotic figures 1).

Dissection Information:

Molecular testing of this specimen was performed after harvesting of targeted tissues with an approved manual microdissection technique. Candidate slides were examined under a microscope and areas containing tumor cells (and separately normal cells, when necessary for testing) were circled. A laboratory technician harvested targeted tissues for extraction from the marked areas using a dissection microscope.

Clinical Trials Connector

CHEMOTHERAPY CLINICAL TRIALS				
Drug class	**Biomarker**	**Method**	**Analyte**	**Investigational agents**
Anti hormonal therapy	ER	IHC	protein	anastrazole, letrozole, exemestane, fulvestrant, tamoxifen, goserelin, leuprolide
Anti hormonal therapy	PR	IHC	protein	anastrazole, letrozole, exemestane, fulvestrant, tamoxifen, goserelin, leuprolide
Anti inflammatory agents	PIK3CA	NGS	DNA tumor	aspirin

TARGETED THERAPY CLINICAL TRIALS				
Drug class	**Biomarker**	**Method**	**Analyte**	**Investigational agents**
Akt inhibitors	ARID1A	NGS	DNA tumor	AZD5363, MK-2206, ipataserib
immunomodulatory agents	TMB	NGS	DNA tumor	avelumab, atezolizumab, durvalumab, ipilimumab, nivolumab, pembrolizumab
PARP inhibitors	NBN	NGS	DNA tumor	BGB-290, BMN-673, olaparib, rucaparib, talazoparib
Akt/mTor inhibitors	PIK3CA	NGS	DNA tumor	AZD5363, BYL719, MK-2206, ipataserib, everolimus, temsirolimus