Final Report

Patient

Name: Joseph Villegas
Date of Birth: 1993-08-10
Sex: Female
Case Number: 1641
Diagnosis: Melanoma

Specimen Information

Primary Tumor Site: Skin
Specimen Site: Subcutaneous tissue
Specimen ID: 6823
Specimen Collected: 2024-03-17
Test Initiated: 2024-03-20

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
Mismatch repair status	IHC	Protien	Negative| 2+, 72%
PHF6	Seq	DNA-Tumor	Mutation not detected
ER	IHC	Protien	Positive| 2+, 16%
NTRK1	Seq	RNA-Tumor	Mutation not detected
TP53	Seq	RNA-Tumor	Fusion not detected

BioMarker	Method	Analyte	Result
U2AF1	Seq	DNA-Tumor	Stable

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Equivocal
Tumor mutational burden	Seq	DNA tumor	9 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	Low - 12% 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 %
MYD88	Seq	DNA tumor	Pathogenic	p.N1465S	20	c.6926del	37.49
PTCH1	Seq	DNA tumor	Pathogenic	p.N452D	8	c.1354A>G	34.69
BRAF	Seq	DNA tumor	Likely Pathogenic	p.P98S	7	1	18.25
MYO1G	Seq	DNA tumor	Likely Benign	p.L576P	3	c.1669T>A	13.08
SDHA	Seq	DNA tumor	Pathogenic	p.H598T	13	c.1820G>T	8.6
MPL	Seq	DNA tumor	Likely Pathogenic	p.E542Q	11	c.1624G>A	8.69

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
ARID2	Seq	DNA tumor	Variant of uncertain significance	p.V49M	2	c.145G>A	20.17
CSF1R	Seq	DNA tumor	Variant of uncertain significance	p.G751A	3	c	20.43
PTCH1	Seq	DNA tumor	Variant of uncertain significance	p.R367Q	3	c.1100G>A	16.02
HBB	Seq	DNA tumor	Variant of uncertain significance	p.F332V	6	1	10.89

Immunohistochemistry Results

Biomarker	Result
ER	Negative| 3+, 7%
PD-L1(SP142)	Positive| 1+, 64%
MLH1	Negative| 1+, 66%
PMS2	Negative| 2+, 82%
ERBB2	Negative| 2+, 86%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

MAPK1 HSP90B1 MSH2

Specimen Information

Specimen ID: 6823

Specimen Collected: 2024-03-17

Specimen Recieved: 2024-03-20

Testing Initiated: 2024-03-20

Gross Description: 6823

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