Final Report

Patient

Name: Craig Matthews
Date of Birth: 1991-02-22
Sex: Male
Case Number: 1070
Diagnosis: Squamous cell carcinoma

Specimen Information

Primary Tumor Site: Skin
Specimen Site: Regional lymph nodes
Specimen ID: 9632
Specimen Collected: 2024-03-31
Test Initiated: 2024-03-31

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	18 m/Mb High	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
U2AF1	Seq	RNA-Tumor	Stable
HBB	Seq	DNA-Tumor	Fusion not detected
HDAC2	Seq	RNA-Tumor	Mutation not detected
NT5C2	Seq	RNA-Tumor	Mutation not detected
SDHA	Seq	DNA-Tumor	Mutation not detected

BioMarker	Method	Analyte	Result
PR	IHC	Protien	Positive| 2+, 31%
Mismatch repair status	IHC	Protien	Negative| 1+, 89%

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	High
Tumor mutational burden	Seq	DNA tumor	18 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 38% 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 %
CSF1R	Seq	DNA tumor	Likely Benign	p.D842V	14	c.2525A>T	30.65
PDGFRA	Seq	DNA tumor	Benign	p.E23Vfs*17	9	c.68_69delAG	13.08
NTRK1	Seq	DNA tumor	Likely Pathogenic	p.R139H	16	c.416G>A	28.34
CD74	Seq	DNA tumor	Benign	p.I1600M	12	c.4800C>G	22.68

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
MYCN	Seq	DNA tumor	Variant of uncertain significance	p.T1195S	12	c.3583A>T	29.34
PTPN11	Seq	DNA tumor	Variant of uncertain significance	p.A34V	16	c	13.95
DPYD	Seq	DNA tumor	Variant of uncertain significance	p.N515H	17	A	14.9
U2AF1	Seq	DNA tumor	Variant of uncertain significance	p.T862I	16	c.2089G>T	9.41
EZH2	Seq	DNA tumor	Variant of uncertain significance	p.P98S	7	1	7.23

Immunohistochemistry Results

Biomarker	Result
MLH1	Positive| 2+, 27%
MSH2	Negative| 2+, 4%
MSH6	Negative| 1+, 63%
PR	Positive| 1+, 6%
PD-L1(SP142)	Positive| 3+, 95%

Biomarker	Result
PMS2	Negative| 3+, 90%
ER	Positive| 1+, 50%
PTEN	Negative| 3+, 85%
ERBB2	Positive| 1+, 71%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

ABL1 CTNNB1 SIX1 NF1 FGFR4

Specimen Information

Specimen ID: 9632

Specimen Collected: 2024-03-31

Specimen Recieved: 2024-03-31

Testing Initiated: 2024-03-31

Gross Description: 9632

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