Final Report

Patient

Name: Jessica Thompson
Date of Birth: 1979-12-15
Sex: Female
Case Number: 3496
Diagnosis: Triple-negative breast cancer

Specimen Information

Primary Tumor Site: Breast
Specimen Site: Lymph nodes (axillary
Specimen ID: 7192
Specimen Collected: 2023-04-27
Test Initiated: 2023-04-28

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	15 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
TLR8	Seq	RNA-Tumor	Stable
FGFR2	Seq	DNA-Tumor	Mutation not detected
APC	Seq	RNA-Tumor	Mutation not detected
MPL	Seq	RNA-Tumor	Stable
Mismatch repair status	IHC	Protien	Negative| 2+, 75%

BioMarker	Method	Analyte	Result
PR	IHC	Protien	Negative| 1+, 65%
PD-L1(SP142)	IHC	Protien	Negative| 1+, 94%
MAPK1	Seq	RNA-Tumor	Mutation not detected
AR	IHC	Protien	Positive| 2+, 89%

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Equivocal
Tumor mutational burden	Seq	DNA tumor	15 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 28% 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 %
MAP2K2	Seq	DNA tumor	Benign	p.S2467Gfs*11	18	c.7399del	24.15
HSP90B1	Seq	DNA tumor	Likely Benign	p.S163P	4	c.487T>C	29.66
IDH1	Seq	DNA tumor	Benign	p.V49M	11	c.145G>A	27.68
CSF1R	Seq	DNA tumor	Likely Pathogenic	p.W515K	6	c.1544G>T	24.54
SF3B1	Seq	DNA tumor	Pathogenic	p.E504K	15	c.1510G>A	4.29
PTCH1	Seq	DNA tumor	Pathogenic	p.C481X	9	c	2.89

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
CD74	Seq	DNA tumor	Variant of uncertain significance	p.K324M	10	c.3209T>G	5.62
FGFR1	Seq	DNA tumor	Variant of uncertain significance	p.R201H	11	c.680A>T	23.39
MET	Seq	DNA tumor	Variant of uncertain significance	p.S2309Cfs*10	15	c.4394A>G	13.44
SIX1	Seq	DNA tumor	Variant of uncertain significance	p.S2467Rfs*13	5	c.5153T>C	7.32

Immunohistochemistry Results

Biomarker	Result
MSH6	Positive| 3+, 94%
PTEN	Positive| 3+, 47%
PMS2	Negative| 2+, 97%
MSH2	Negative| 2+, 6%
ER	Negative| 2+, 82%

Biomarker	Result
PD-L1(SP142)	Positive| 2+, 71%
MLH1	Negative| 1+, 38%
ERBB2	Positive| 1+, 87%
AR	Positive| 2+, 40%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

FGFR1 CCND3 CHD6 CHD6 MYCN

Specimen Information

Specimen ID: 7192
Specimen Collected: 2023-04-27
Specimen Recieved: 2023-04-28
Testing Initiated: 2023-04-28
Gross Description: 7192

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