Final Report

Patient

Name: Jeffery Reeves
Date of Birth: 1967-08-25
Sex: Female
Case Number: 2497
Diagnosis: Ductal carcinoma in situ (DCIS)

Specimen Information

Primary Tumor Site: Breast
Specimen Site: Breast tissue
Specimen ID: 8951
Specimen Collected: 2024-04-18
Test Initiated: 2024-04-24

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	14 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
KLF4	Seq	RNA-Tumor	Mutation not detected
HBB	Seq	RNA-Tumor	Stable
PR	IHC	Protien	Positive| 3+, 72%
KIT	Seq	RNA-Tumor	Stable
BRCA2	Seq	RNA-Tumor	Fusion not detected

BioMarker	Method	Analyte	Result
Mismatch repair status	IHC	Protien	Negative| 2+, 100%
PD-L1(SP142)	IHC	Protien	Negative| 1+, 75%

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	High
Tumor mutational burden	Seq	DNA tumor	14 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 34% 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 %
SDHD	Seq	DNA tumor	Likely Benign	p.G183S	4	c.964G>A	6.99
CHEK2	Seq	DNA tumor	Pathogenic	p.R367Q	3	c.1100G>A	32.65
CSF1R	Seq	DNA tumor	Pathogenic	p.I543V	20	c.1905+1G>A	2.2

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
PDGFRA	Seq	DNA tumor	Variant of uncertain significance	p.A298P	10	c.892G>C	8.53
GNAS	Seq	DNA tumor	Variant of uncertain significance	p.G12V	10	c.181C>A	15.71
SRSF2	Seq	DNA tumor	Variant of uncertain significance	p.A169T	8	c.954G>T	7.18
STAT5B	Seq	DNA tumor	Variant of uncertain significance	p.L857P	11	>	12.41
CSF3R	Seq	DNA tumor	Variant of uncertain significance	p.R140Q	7	c.418C>T	4.55
SMO	Seq	DNA tumor	Variant of uncertain significance	p.M929I	11	c.2792A>G	2.58

Immunohistochemistry Results

Biomarker	Result
PTEN	Positive| 1+, 46%
MSH6	Positive| 1+, 56%
PMS2	Negative| 3+, 24%
PR	Positive| 2+, 73%
MSH2	Negative| 3+, 18%

Biomarker	Result
ER	Positive| 1+, 43%
AR	Negative| 1+, 96%
MLH1	Positive| 2+, 61%
ERBB2	Negative| 3+, 7%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

MYOD1 MSH2 BRCA2 FGFR2 CTNNB1 SMO

Specimen Information

Specimen ID: 8951

Specimen Collected: 2024-04-18

Specimen Recieved: 2024-04-24

Testing Initiated: 2024-04-24

Gross Description: 8951

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