Final Report

Patient

Name: Denise Williams
Date of Birth: 1965-02-12
Sex: Female
Case Number: 3806
Diagnosis: Lobular carcinoma in situ (LCIS)

Specimen Information

Primary Tumor Site: Breast
Specimen Site: Lymph nodes (axillary
Specimen ID: 2983
Specimen Collected: 2023-11-03
Test Initiated: 2023-11-06

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	7 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
TOP2A	Seq	RNA-Tumor	Stable
PTEN	Seq	DNA-Tumor	Fusion not detected
PD-L1(SP142)	IHC	Protien	Positive| 2+, 90%
AKT1	Seq	RNA-Tumor	Stable
FGFR2	Seq	DNA-Tumor	Mutation not detected

BioMarker	Method	Analyte	Result
DPYD	Seq	DNA-Tumor	Fusion not detected
BTK	Seq	RNA-Tumor	Stable
MSH2	Seq	DNA-Tumor	Stable
CCND3	Seq	RNA-Tumor	Mutation not detected
BTK	Seq	DNA-Tumor	Mutation not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	High
Tumor mutational burden	Seq	DNA tumor	7 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 24% 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 %
MYOD1	Seq	DNA tumor	Likely Benign	p.N452D	20	c.1354A>G	38.84
PTCH1	Seq	DNA tumor	Likely Pathogenic	p.*143Qext*31	11	c.427T>C	9.19
GNAS	Seq	DNA tumor	Benign	p.S346P	11	3	28.57
ARID2	Seq	DNA tumor	Likely Pathogenic	p.R479H	18	c.1436G>A	19.52
NRAS	Seq	DNA tumor	Pathogenic	p.C481X	5	.	11.18

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
XPC	Seq	DNA tumor	Variant of uncertain significance	p.L1196M	11	c.3522C>G	16.17
APC	Seq	DNA tumor	Variant of uncertain significance	p.R140W	2	c.515G>T	9.22
U2AF1	Seq	DNA tumor	Variant of uncertain significance	p.A374E	9	G	14.2

Immunohistochemistry Results

Biomarker	Result
PR	Positive| 3+, 62%
MSH6	Negative| 2+, 49%
PTEN	Negative| 1+, 95%
AR	Negative| 1+, 29%
PD-L1(SP142)	Positive| 1+, 52%

Biomarker	Result
MSH2	Positive| 3+, 1%
PMS2	Negative| 1+, 40%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

BRAF STAT5B CD74 PRKCA SIX1

Specimen Information

Specimen ID: 2983

Specimen Collected: 2023-11-03

Specimen Recieved: 2023-11-06

Testing Initiated: 2023-11-06

Gross Description: 2983

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