Final Report

Patient

Name: Alison Jones
Date of Birth: 1941-07-04
Sex: Male
Case Number: 4751
Diagnosis: Lobular carcinoma in situ (LCIS)

Specimen Information

Primary Tumor Site: Breast
Specimen Site: Lymph nodes (axillary
Specimen ID: 3372
Specimen Collected: 2024-03-30
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	11 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
CHD6	Seq	RNA-Tumor	Stable
PD-L1(SP142)	IHC	Protien	Positive| 1+, 29%
CSF3R	Seq	RNA-Tumor	Stable
PD-L1(SP142)	IHC	Protien	Positive| 3+, 6%
PR	IHC	Protien	Positive| 1+, 7%

BioMarker	Method	Analyte	Result
Mismatch repair status	IHC	Protien	Negative| 3+, 49%
PR	IHC	Protien	Negative| 2+, 21%
PIK3CA	Seq	DNA-Tumor	Stable
ER	IHC	Protien	Negative| 3+, 62%
Mismatch repair status	IHC	Protien	Positive| 2+, 89%

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	High
Tumor mutational burden	Seq	DNA tumor	11 mutations/Mb Low
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 %
DPYD	Seq	DNA tumor	Pathogenic	p.A374E	7	>	27.28
PIK3CA	Seq	DNA tumor	Pathogenic	p.N159Y	14	3	11.28
CD74	Seq	DNA tumor	Pathogenic	p.C481X	11	1	21.45
PDGFRB	Seq	DNA tumor	Pathogenic	p.G12D	14	c.35_36inv	1.29

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
CALR	Seq	DNA tumor	Variant of uncertain significance	p.N515H	14	A	8.82
SDHB	Seq	DNA tumor	Variant of uncertain significance	p.L122R	5	c.365T>G	24.79
JAK3	Seq	DNA tumor	Variant of uncertain significance	p.P904L	16	c.2711C>T	1.11
ZEB2	Seq	DNA tumor	Variant of uncertain significance	p.K860I	19	c.2238_2252del15	2.88
CDC73	Seq	DNA tumor	Variant of uncertain significance	p.W266*	3	c.798G>A	2.08

Immunohistochemistry Results

Biomarker	Result
AR	Negative| 2+, 4%
PTEN	Negative| 1+, 74%
PR	Positive| 3+, 85%
MSH2	Positive| 3+, 69%
MLH1	Negative| 1+, 7%

Biomarker	Result
ERBB2	Negative| 2+, 12%
PMS2	Positive| 1+, 40%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

JAK2 DICER1 PIK3CA SDHD

Specimen Information

Specimen ID: 3372

Specimen Collected: 2024-03-30

Specimen Recieved: 2024-03-31

Testing Initiated: 2024-03-31

Gross Description: 3372

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