Final Report

Patient

Name: Robert Johnson
Date of Birth: 1983-03-09
Sex: Female
Case Number: 6846
Diagnosis: Triple-negative breast cancer

Specimen Information

Primary Tumor Site: Breast
Specimen Site: Lymph nodes (axillary
Specimen ID: 7768
Specimen Collected: 2023-06-21
Test Initiated: 2023-06-24

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
NT5C2	Seq	DNA-Tumor	Mutation not detected
B2M	Seq	RNA-Tumor	Stable
PTPN11	Seq	RNA-Tumor	Stable
FGFR1	Seq	DNA-Tumor	Mutation not detected
STAT5B	Seq	RNA-Tumor	Mutation not detected

BioMarker	Method	Analyte	Result
NRAS	Seq	DNA-Tumor	Fusion not detected
ER	IHC	Protien	Negative| 1+, 56%

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 - 37% 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 %
JAK2	Seq	DNA tumor	Benign	p.N452D	5	c.1354A>G	31.52
FGFR3	Seq	DNA tumor	Benign	p.Q61L	11	c.37G>A	26.97
NF1	Seq	DNA tumor	Likely Benign	p.Y736fs*4	15	c.2207_2212delinsTAGATTC	17.68
SF3B1	Seq	DNA tumor	Likely Pathogenic	p.P98S	7	>	10.75
NRAS	Seq	DNA tumor	Benign	p.F1888L	3	c.5664C>A	4.52

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
NOTCH1	Seq	DNA tumor	Variant of uncertain significance	p.Y303H	18	A	21.07
TP53	Seq	DNA tumor	Variant of uncertain significance	p.A322T	7	c.964G>A	18.07
ZEB2	Seq	DNA tumor	Variant of uncertain significance	p.I1600M	11	c.4800C>G	18.04
FGFR2	Seq	DNA tumor	Variant of uncertain significance	p.D32G	9	c.94G>T	8.53
SDHA	Seq	DNA tumor	Variant of uncertain significance	p.V49M	8	c.145G>A	4.75
BTK	Seq	DNA tumor	Variant of uncertain significance	p.A34V	15	1	3.94

Immunohistochemistry Results

Biomarker	Result
PD-L1(SP142)	Negative| 3+, 35%
AR	Positive| 1+, 94%
ER	Positive| 2+, 99%
MSH2	Positive| 3+, 94%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

IDH1 NF1 SF3B1

Specimen Information

Specimen ID: 7768

Specimen Collected: 2023-06-21

Specimen Recieved: 2023-06-24

Testing Initiated: 2023-06-24

Gross Description: 7768

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