Final Report

Patient

Name: John Ortiz
Date of Birth: 1924-04-21
Sex: Female
Case Number: 9466
Diagnosis: Diffuse large B-cell lymphoma

Specimen Information

Primary Tumor Site: Lymphoma
Specimen Site: Bone marrow
Specimen ID: 5353
Specimen Collected: 2023-11-18
Test Initiated: 2023-11-21

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	9 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
ERCC2	Seq	DNA-Tumor	Stable
IDH1	Seq	DNA-Tumor	Fusion not detected
MUTYH	Seq	DNA-Tumor	Stable
MYOD1	Seq	DNA-Tumor	Stable
PD-L1(SP142)	IHC	Protien	Positive| 1+, 37%

BioMarker	Method	Analyte	Result
HDAC2	Seq	RNA-Tumor	Mutation not detected
ER	IHC	Protien	Negative| 3+, 31%
JAK2	Seq	RNA-Tumor	Mutation not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Low
Tumor mutational burden	Seq	DNA tumor	9 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	Low - 5% 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 %
SF3B1	Seq	DNA tumor	Pathogenic	p.P128L	12	c.383C>T	39.19
HDAC1	Seq	DNA tumor	Likely Pathogenic	p.W266*	16	c.798G>A	7.58
MAP2K2	Seq	DNA tumor	Benign	p.I157T	5	c.470T>C	19.98
NRAS	Seq	DNA tumor	Likely Benign	p.N159Y	8	>	8.18
EGFR	Seq	DNA tumor	Pathogenic	p.Y736fs*4	3	c.2207_2212delinsTAGATTC	16.21

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
ABL1	Seq	DNA tumor	Variant of uncertain significance	p.Y646X	7	c.1937A>G	11.89
KRAS	Seq	DNA tumor	Variant of uncertain significance	p.G1386D	14	1	20.06
ALDH2	Seq	DNA tumor	Variant of uncertain significance	p.T681I	9	>	18.88

Immunohistochemistry Results

Biomarker	Result
ER	Positive| 1+, 15%
AR	Negative| 3+, 76%
PD-L1(SP142)	Negative| 1+, 2%
MSH2	Positive| 2+, 23%
PMS2	Negative| 1+, 11%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

ARHGAP45 ABL1 MSH2 ZEB2 IDH1

Specimen Information

Specimen ID: 5353
Specimen Collected: 2023-11-18
Specimen Recieved: 2023-11-21
Testing Initiated: 2023-11-21
Gross Description: 5353

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