Final Report

Patient

Name: Kathleen Middleton
Date of Birth: 1939-11-18
Sex: Female
Case Number: 2896
Diagnosis: Chronic lymphocytic leukemia (CLL)

Specimen Information

Primary Tumor Site: Leukemia
Specimen Site: Lymph nodes
Specimen ID: 1632
Specimen Collected: 2023-06-03
Test Initiated: 2023-06-03

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	12 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
ERBB2	Seq	RNA-Tumor	Stable
PD-L1(SP142)	IHC	Protien	Positive| 3+, 79%
CHEK2	Seq	RNA-Tumor	Stable
SF3B1	Seq	DNA-Tumor	Fusion not detected
ARHGAP45	Seq	DNA-Tumor	Stable

BioMarker	Method	Analyte	Result
XPC	Seq	DNA-Tumor	Stable
SDHD	Seq	RNA-Tumor	Mutation not detected
Mismatch repair status	IHC	Protien	Positive| 1+, 68%
MUTYH	Seq	DNA-Tumor	Mutation not detected
CDKN2A	Seq	DNA-Tumor	Mutation not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	High
Tumor mutational burden	Seq	DNA tumor	12 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 38% 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 %
NF1	Seq	DNA tumor	Likely Benign	p.I638F	13	c.2304T>A	6.77
DDR2	Seq	DNA tumor	Benign	p.S346P	2	.	28.33
MTOR	Seq	DNA tumor	Benign	p.R201C	16	c.680A>G	3.46

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.N713_A714insKGKGGG	18	c.1924A>C	16.8
PTEN	Seq	DNA tumor	Variant of uncertain significance	p.G503A	20	c.1508G>C	6.99
HDAC2	Seq	DNA tumor	Variant of uncertain significance	p.*143Qext*31	3	c.427T>C	13.59
DPYD	Seq	DNA tumor	Variant of uncertain significance	p.N713_A714insKGKGGG	19	c.1924A>C	8.38

Immunohistochemistry Results

Biomarker	Result
PD-L1(SP142)	Negative| 3+, 29%
PTEN	Negative| 2+, 90%
ER	Positive| 1+, 28%
ERBB2	Negative| 2+, 11%
PR	Negative| 2+, 42%

Biomarker	Result
AR	Positive| 3+, 69%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

ZEB2 MYOD1 SF3B1 PTPN11

Specimen Information

Specimen ID: 1632

Specimen Collected: 2023-06-03

Specimen Recieved: 2023-06-03

Testing Initiated: 2023-06-03

Gross Description: 1632

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