

Patient

Specimen Information

Ordered By

Name: Cindy Roberts

Date of Birth: 2001-04-26

Sex: Female

Case Number: 2158

Diagnosis: Chronic lymphocytic leukemia (CLL)

Primary Tumor Site: Leukemia

Specimen Site: Spleen

Specimen ID: 3789

Specimen Collected: 2024-02-11

Test Initiated: 2024-02-15

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	5 m/Mb Low	BENEFIT	pembrolizumab	level 2
ERBB2	IHC	Protien	Negative 0	LACK OF BENEFIT	trastuzumab, ado-trastuzumab emtansine, pertuzumab, fam-trastuzumab deruxtecan-nxki, lapatinib, neratinib, tucatinib	level 1

Cancer-Type Relevant Biomarkers

BioMarker	Method	Analyte	Result
SDHA	Seq	DNA-Tumor	Fusion not detected
CDC73	Seq	DNA-Tumor	Mutation not detected
PR	IHC	Protien	Negative 3+, 44%
KRAS	Seq	RNA-Tumor	Stable

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Equivocal
Tumor mutational burden	Seq	DNA tumor	5 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	Low - 1% 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 %
ARHGAP45	Seq	DNA tumor	Likely Pathogenic	p.R742W	17	c.1912A>T	27.6
HDAC1	Seq	DNA tumor	Likely Benign	p.P44L	13	c.131_132delinsTT	18.0
PAX5	Seq	DNA tumor	Benign	p.G466E	15	c.1798_1799delinsAA	5.75
CSF1R	Seq	DNA tumor	Benign	p.P98S	6	G	5.29
DNMT3A	Seq	DNA tumor	Likely Pathogenic	p.Q227E	16	c.478C>T	9.58
KLF4	Seq	DNA tumor	Likely Pathogenic	p.Y303H	3	>	2.57

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
HBA2	Seq	DNA tumor	Variant of uncertain significance	p.E322K	11	c.964G>A	16.78
FGFR1	Seq	DNA tumor	Variant of uncertain significance	p.A374E	17	3	16.97
GNAS	Seq	DNA tumor	Variant of uncertain significance	p.R367Q	6	c.1100G>A	6.16
NF1	Seq	DNA tumor	Variant of uncertain significance	p.L130Q	12	c.247C>T	9.77
PIK3CA	Seq	DNA tumor	Variant of uncertain significance	p.K385fs*47	4	c.1092_1143del52	8.65

Immunohistochemistry Results

Biomarker	Result	Biomarker	Result
ERBB2	Positive 2+, 79%	MLH1	Negative 2+, 42%
MSH6	Positive 2+, 95%	MSH2	Negative 2+, 30%
PR	Negative 2+, 73%	AR	Negative 1+, 57%
PD-L1(SP142)	Positive 3+, 71%	PMS2	Negative 3+, 37%
ER	Positive 3+, 37%	PTEN	Negative 1+, 74%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

FBXW7 APC HDAC2 HBA2 NRAS

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Specimen ID: 3789

Specimen Collected: 2024-02-11

Specimen Recieved: 2024-02-15

Testing Initiated: 2024-02-15

Gross Description: 3789

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	anastrozole, letrozole, exemestane, fulvestrant, tamoxifen, goserelin, leuprolide
Anti hormonal therapy	PR	IHC	protein	anastrozole, 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