**Patient** 

Name: Monica Morales

Date of Birth: 1942-08-20

Sex: Male

Case Number: 6600 Diagnosis: Lymphoma **Specimen Information** 

Primary Tumor Site: Colorectal

Specimen Site: Rectum Specimen ID: 9850

Specimen Collected: 2023-12-25

Test Initiated: 2023-12-25

### 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
ТМВ	seq	DNA tumor	13 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
PTCH1	Seq RNA-Tumor		Fusion not detected
ER	IHC	Protien	Negative  2+, 23%
PHF6	Seq	DNA-Tumor	Fusion not detected
PD-L1(SP142)	IHC	Protien	Positive  3+, 50%
ARID2	Seq	RNA-Tumor	Fusion not detected

# Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Low
Tumor mutational burden	Seq	DNA tumor	13 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 21% 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 %
BRCA1	Seq	DNA tumor	Likely Benign	p.A374E	10	А	36.65
NCSTN	Seq	DNA tumor	Likely Benign	p.S163P	8	c.487T>C	7.83
STAT5B	Seq	DNA tumor	Likely Benign	p.L239R	16	c.1912A>T	21.27
ALK	Seq	DNA tumor	Likely Pathogenic	p.R139H	9	c.416G>A	8.61
SDHA	Seq	DNA tumor	Pathogenic	p.A1515S	19	3	19.33
HBB	Seq	DNA tumor	Benign	p.T862I	8	c.2524G>A	1.54

### 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.Y303H	7	С	26.71
IDH1	Seq	DNA tumor	Variant of uncertain significance	p.R479H	15	c.1436G>A	8.83
PAX5	Seq	DNA tumor	Variant of uncertain significance	p.G996R	10	c.1849G>T	10.04
KLF1	Seq	DNA tumor	Variant of uncertain significance	p.C134W	6	c.402C>G	11.55

### Immunohistochemistry Results

Biomarker	Result
ER	Positive  3+, 52%
MSH6	Positive  1+, 75%
AR	Positive  3+, 91%
PMS2	Negative  1+, 93%
ERBB2	Negative  2+, 18%

Biomarker	Result
PD-L1(SP142)	Positive  1+, 21%
PTEN	Negative  2+, 70%

### Genes Tested with Indeterminate Results by Tumor DNA Sequencing

RNF43 ALK DICER1 MYD88 AKT1

#### **Specimen Information**

Specimen ID: 9850 Specimen Collected: 2023-12-25 Specimen Recieved: 2023-12-25 Testing Initiated: 2023-12-25

Gross Description: 9850

#### **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		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		