**Patient** 

Name: Andrew Nelson Date of Birth: 1991-10-15

Sex: Female

Case Number: 1907 Diagnosis: Lymphoma **Specimen Information** 

Primary Tumor Site: Colorectal Specimen Site: Mesentery

Specimen ID: 1496

Specimen Collected: 2023-10-04

Test Initiated: 2023-10-04

# Ordered By

Biomarker	Method	Analyte	Result		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	15 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
MSH2	Seq	RNA-Tumor	Mutation not detected
HDAC2	Seq	DNA-Tumor	Fusion not detected
PRKCA	Seq	DNA-Tumor	Mutation not detected
PR	IHC	Protien	Positive  3+, 34%

# **Genomic Signatures**

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Low
Tumor mutational burden	Seq	DNA tumor	15 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 35% 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 %
TOP2A	Seq	DNA tumor	Benign	p.G1386D	20	3	9.05
CDKN2A	Seq	DNA tumor	Benign	p.T1195S	9	c.3583A>T	18.91
CDKN2A	Seq	DNA tumor	Likely Benign	p.S1982Rfs*22	20	c.5946delT	29.59
CHEK2	Seq	DNA tumor	Pathogenic	p.R282G	7	c.815T>C	14.56
ALDH2	Seq	DNA tumor	Likely Pathogenic	p.N713_A714insKGKGGG	3	c.1924A>C	13.47

# Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
PRKCA	Seq	DNA tumor	Variant of uncertain significance	p.K659E	13	c.1647T>A	23.86
EZH2	Seq	DNA tumor	Variant of uncertain significance	p.P904L	6	c.2711C>T	7.95
B2M	Seq	DNA tumor	Variant of uncertain significance	p.G12A	9	c.35G>C	9.99
HBA2	Seq	DNA tumor	Variant of uncertain significance	p.K409Q	4	c.1225A>C	2.82
HSP90B1	Seq	DNA tumor	Variant of uncertain significance	p.T1195S	9	c.3583A>T	7.64

# Immunohistochemistry Results

Biomarker	Result
MLH1	Positive  1+, 71%
PD-L1(SP142)	Positive  3+, 69%
PR	Positive  1+, 12%
PTEN	Positive  3+, 68%
ER	Negative  2+, 2%

# Genes Tested with Indeterminate Results by Tumor DNA Sequencing

STAT5B HRAS RB1 BRAF STAG2 PRKCA

## **Specimen Information**

Specimen ID: 1496 Specimen Collected: 2023-10-04 Specimen Recieved: 2023-10-04 Testing Initiated: 2023-10-04

Gross Description: 1496

#### **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 Investigation			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 Analys		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		