Final Report

Patient

Name: Gregory Perry
Date of Birth: 1970-05-30
Sex: Female
Case Number: 5908
Diagnosis: Lymphoma

Specimen Information

Primary Tumor Site: Colorectal
Specimen Site: Rectum
Specimen ID: 7844
Specimen Collected: 2023-07-10
Test Initiated: 2023-07-10

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	16 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
PD-L1(SP142)	IHC	Protien	Positive| 3+, 23%
PR	IHC	Protien	Negative| 2+, 2%
AR	IHC	Protien	Negative| 3+, 58%
AR	IHC	Protien	Positive| 1+, 31%
Mismatch repair status	IHC	Protien	Negative| 2+, 2%

BioMarker	Method	Analyte	Result
CALR	Seq	DNA-Tumor	Mutation not detected
MPL	Seq	RNA-Tumor	Fusion not detected
SRSF2	Seq	DNA-Tumor	Stable
FGFR4	Seq	RNA-Tumor	Fusion not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Equivocal
Tumor mutational burden	Seq	DNA tumor	16 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 32% 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 %
MTOR	Seq	DNA tumor	Likely Pathogenic	p.Y1225X	16	c.5438A>G	27.82
PKLR	Seq	DNA tumor	Pathogenic	p.M1R	4	c.2T>G	3.93
CALR	Seq	DNA tumor	Pathogenic	p.F1888L	18	c.5664C>G	8.06
CALR	Seq	DNA tumor	Likely Benign	p.K656E	14	c.1966A>G	6.45

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
MYCN	Seq	DNA tumor	Variant of uncertain significance	p.K385fs*47	6	c.1154_1155insTTGTC	7.02
GNAS	Seq	DNA tumor	Variant of uncertain significance	p.R175C	8	c.396G>C	21.2
CHD6	Seq	DNA tumor	Variant of uncertain significance	p.H1038R	14	c.3113A>G	8.38

Immunohistochemistry Results

Biomarker	Result
PMS2	Positive| 2+, 40%
PD-L1(SP142)	Positive| 3+, 19%
MSH6	Positive| 2+, 21%
PTEN	Negative| 2+, 25%
ERBB2	Negative| 3+, 82%

Biomarker	Result
PR	Negative| 3+, 77%
MSH2	Negative| 1+, 17%
MLH1	Positive| 3+, 33%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

PAX5 NF1 PKLR DPYD EGFR

Final Report

Specimen Information

Specimen ID: 7844

Specimen Collected: 2023-07-10

Specimen Recieved: 2023-07-10

Testing Initiated: 2023-07-10

Gross Description: 7844

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