Final Report

Patient

Name: Catherine Hernandez
Date of Birth: 1991-12-25
Sex: Female
Case Number: 1859
Diagnosis: Germ cell tumors

Specimen Information

Primary Tumor Site: Ovarian
Specimen Site: Ovaries
Specimen ID: 2895
Specimen Collected: 2024-04-18
Test Initiated: 2024-04-18

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
PR	IHC	Protien	Positive| 1+, 1%
TOP2A	Seq	DNA-Tumor	Stable
MYO1G	Seq	DNA-Tumor	Fusion not detected
ABL1	Seq	DNA-Tumor	Stable
FGFR2	Seq	DNA-Tumor	Mutation not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Equivocal
Tumor mutational burden	Seq	DNA tumor	12 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	Low - 4% 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 %
CTNNB1	Seq	DNA tumor	Pathogenic	p.A169T	15	c.505G>A	22.31
ALDH2	Seq	DNA tumor	Benign	p.R479H	14	c.1436G>A	28.56
ARID2	Seq	DNA tumor	Likely Pathogenic	p.P95X	17	1	2.49
KLF4	Seq	DNA tumor	Benign	p.H598T	20	c.1792C>T	23.64
ALK	Seq	DNA tumor	Likely Pathogenic	p.T367N	20	c.1100C>A	1.08
EGFR	Seq	DNA tumor	Benign	p.W515L	17	c.1544G>T	2.29

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
ERCC2	Seq	DNA tumor	Variant of uncertain significance	p.R1012X	15	1	14.69
TOP2A	Seq	DNA tumor	Variant of uncertain significance	p.Y599C	19	c.1775T>G	2.59
PTEN	Seq	DNA tumor	Variant of uncertain significance	p.A34V	19	A	3.02

Immunohistochemistry Results

Biomarker	Result
ER	Negative| 2+, 3%
PR	Negative| 1+, 82%
ERBB2	Positive| 2+, 52%
PMS2	Positive| 2+, 40%
MLH1	Positive| 1+, 64%

Biomarker	Result
AR	Negative| 3+, 65%
MSH2	Positive| 3+, 62%
PD-L1(SP142)	Negative| 3+, 97%
MSH6	Positive| 1+, 52%
PTEN	Positive| 2+, 62%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

BRCA1 PIK3CA ARHGAP45 PTCH1

Specimen Information

Specimen ID: 2895
Specimen Collected: 2024-04-18
Specimen Recieved: 2024-04-18
Testing Initiated: 2024-04-18
Gross Description: 2895

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