

Patient	Specimen Information	Ordered By
Name: Jessica Villa	Primary Tumor Site: Thyroid	
Date of Birth: 1941-01-18	Specimen Site: Thyroid gland	
Sex: Male	Specimen ID: 1313	
Case Number: 3095	Specimen Collected: 2023-07-27	
Diagnosis: Follicular thyroid cancer	Test Initiated: 2023-07-27	

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	19 m/Mb High	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
DICER1	Seq	RNA-Tumor	Fusion not detected
PTEN	Seq	DNA-Tumor	Fusion not detected
ER	IHC	Protien	Negative 1+, 53%
BRCA1	Seq	DNA-Tumor	Mutation not detected
IDH1	Seq	DNA-Tumor	Fusion not detected

BioMarker	Method	Analyte	Result
MTOR	Seq	DNA-Tumor	Fusion not detected
NT5C2	Seq	RNA-Tumor	Mutation not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Low
Tumor mutational burden	Seq	DNA tumor	19 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 17% 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 %
STAT5B	Seq	DNA tumor	Pathogenic	p.L858G	7	c.2386G>C	22.64
DNMT3A	Seq	DNA tumor	Likely Benign	p.D463H	18	c.1387G>C	20.09
MYCN	Seq	DNA tumor	Pathogenic	p.E322K	2	c.964G>A	8.02
APC	Seq	DNA tumor	Likely Benign	p.F332V	18	A	19.24

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
EZH2	Seq	DNA tumor	Variant of uncertain significance	p.A374E	19	3	28.53
PDGFRA	Seq	DNA tumor	Variant of uncertain significance	p.K385I	13	c.2525A>T	4.25
CSF1R	Seq	DNA tumor	Variant of uncertain significance	p.G35W	9	1	8.18
FGFR2	Seq	DNA tumor	Variant of uncertain significance	p.P95X	2	A	1.29
NTRK1	Seq	DNA tumor	Variant of uncertain significance	p.F691L	9	c.2028C>A	5.69
SRSF2	Seq	DNA tumor	Variant of uncertain significance	p.R479H	14	c.1436G>A	3.65

Immunohistochemistry Results

Biomarker	Result	Biomarker	Result
MLH1	Positive 3+, 84%	PR	Negative 1+, 92%
AR	Positive 1+, 77%	PTEN	Positive 1+, 4%
ERBB2	Negative 2+, 83%	MSH6	Negative 2+, 82%
MSH2	Negative 2+, 71%	PD-L1(SP142)	Positive 2+, 99%
ER	Negative 2+, 46%		

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

GNAS MPL CSF3R BRAF RNF43 HDAC2

Specimen Information

Specimen ID: 1313

Specimen Collected: 2023-07-27

Specimen Recieved: 2023-07-27

Testing Initiated: 2023-07-27

Gross Description: 1313

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