

Patient	Specimen Information	Ordered By
Name: Faith Garcia	Primary Tumor Site: Thyroid	
Date of Birth: 1998-07-19	Specimen Site: Perithyroidal tissue	
Sex: Male	Specimen ID: 7985	
Case Number: 4601	Specimen Collected: 2024-02-11	
Diagnosis: Papillary thyroid cancer	Test Initiated: 2024-02-16	

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 BENEFIT	trastuzumab, ado-trastuzumab emtansine, pertuzumab, fam-trastuzumab deruxtecan-nxki, lapatinib, neratinib, tucatinib	level 1

Cancer-Type Relevant Biomarkers

BioMarker	Method	Analyte	Result
MYO1G	Seq	RNA-Tumor	Stable
PDGFRA	Seq	DNA-Tumor	Mutation not detected
PR	IHC	Protien	Positive  2+, 88%
DPYD	Seq	RNA-Tumor	Mutation not detected
PD-L1(SP142)	IHC	Protien	Negative  1+, 70%

BioMarker	Method	Analyte	Result
TP53	Seq	DNA-Tumor	Stable
AR	IHC	Protien	Negative  3+, 57%
CDC73	Seq	DNA-Tumor	Mutation not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	High
Tumor mutational burden	Seq	DNA tumor	12 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 18% 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 %
ABL1	Seq	DNA tumor	Pathogenic	p.R130G	18	c.545T>G	35.59
CDKN2A	Seq	DNA tumor	Likely Benign	p.R1012X	14	.	17.72
KLF4	Seq	DNA tumor	Likely Pathogenic	p.L239R	2	c.716T>G	12.63
FGFR1	Seq	DNA tumor	Likely Pathogenic	p.G503A	4	c.1508G>C	4.16
SF3B1	Seq	DNA tumor	Benign	p.H362R	12	c.1085A>G	6.17
SDHD	Seq	DNA tumor	Likely Pathogenic	p.N452D	18	c.1354A>G	2.53

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
NT5C2	Seq	DNA tumor	Variant of uncertain significance	p.T367=	4	c.1100C>A	22.19
DPYD	Seq	DNA tumor	Variant of uncertain significance	p.E23Vfs*17	6	c.68_69delAG	23.78
DPYD	Seq	DNA tumor	Variant of uncertain significance	p.M1R	3	c.2T>G	3.25
TLR8	Seq	DNA tumor	Variant of uncertain significance	p.L122R	5	c.365T>G	8.07
PAX5	Seq	DNA tumor	Variant of uncertain significance	p.M1V	8	c.1A>G	4.7
TLR8	Seq	DNA tumor	Variant of uncertain significance	p.Y736fs*4	4	c.2207_2212delinsTAGATTC	1.53

Immunohistochemistry Results

Biomarker	Result
ERBB2	Positive  2+, 49%
ER	Negative  2+, 54%
PTEN	Positive  1+, 37%
PMS2	Negative  3+, 86%
MSH6	Positive  3+, 28%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

IKZF1      EGFR      MPL      AKT1

Specimen Information

Specimen ID: 7985

Specimen Collected: 2024-02-11

Specimen Recieved: 2024-02-16

Testing Initiated: 2024-02-16

Gross Description: 7985

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