

Patient	Specimen Information	Ordered By
Name: Whitney Garcia	Primary Tumor Site: Thyroid	
Date of Birth: 1974-10-26	Specimen Site: Perithyroidal tissue	
Sex: Female	Specimen ID: 2720	
Case Number: 3622	Specimen Collected: 2024-02-25	
Diagnosis: Medullary thyroid cancer	Test Initiated: 2024-02-25	

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	8 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
ER	IHC	Protien	Positive 1+, 89%
HRAS	Seq	DNA-Tumor	Mutation not detected
DNMT3A	Seq	DNA-Tumor	Mutation not detected
PD-L1(SP142)	IHC	Protien	Negative 1+, 69%
PD-L1(SP142)	IHC	Protien	Positive 1+, 61%

BioMarker	Method	Analyte	Result
CDC73	Seq	DNA-Tumor	Stable
DDR2	Seq	DNA-Tumor	Mutation not detected
KIT	Seq	RNA-Tumor	Mutation not detected
IDH1	Seq	RNA-Tumor	Mutation not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Equivocal
Tumor mutational burden	Seq	DNA tumor	8 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 %
FGFR3	Seq	DNA tumor	Benign	p.W515L	6	c.1543_1544delinsAA	30.29
NOTCH1	Seq	DNA tumor	Pathogenic	p.R465H	5	c.1394G>A	28.74
H3.3	Seq	DNA tumor	Benign	p.P128L	13	c.383C>T	1.14

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
MUTYH	Seq	DNA tumor	Variant of uncertain significance	p.E504K	18	c.1510G>A	22.66
KLF4	Seq	DNA tumor	Variant of uncertain significance	p.N1465S	12	c.4394A>G	6.8
MYO1G	Seq	DNA tumor	Variant of uncertain significance	p.Q709L	14	c.2584A>G	2.27

Immunohistochemistry Results

Biomarker	Result	Biomarker	Result
MSH2	Positive 2+, 60%	AR	Positive 2+, 52%
PTEN	Negative 2+, 60%	PMS2	Negative 1+, 47%
ERBB2	Negative 1+, 65%	MLH1	Negative 1+, 35%
MSH6	Negative 3+, 14%		
PR	Positive 2+, 26%		

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

ERCC2 CTNNB1 CALR ARID2 DDR2

Specimen Information

Specimen ID: 2720Specimen Collected: 2024-02-25

Specimen Recieved: 2024-02-25Testing Initiated: 2024-02-25

Gross Description: 2720

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