

Patient	Specimen Information	Ordered By
Name: Nichole Michael	Primary Tumor Site: Thyroid	
Date of Birth: 1959-12-01	Specimen Site: Central and lateral cervical lymph nodes	
Sex: Female	Specimen ID: 5135	
Case Number: 3260	Specimen Collected: 2023-06-23	
Diagnosis: Medullary thyroid cancer	Test Initiated: 2023-06-23	

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
ABL1	Seq	DNA-Tumor	Mutation not detected
MET	Seq	DNA-Tumor	Fusion not detected
PTEN	Seq	DNA-Tumor	Mutation not detected
PR	IHC	Protien	Negative 1+, 44%

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Low
Tumor mutational burden	Seq	DNA tumor	8 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	Low - 5% 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 %
NOTCH1	Seq	DNA tumor	Likely Benign	p.S163P	20	c.487T>C	29.23
FGFR1	Seq	DNA tumor	Pathogenic	p.R132V	13	c.395_396inv	34.85
HRAS	Seq	DNA tumor	Likely Pathogenic	p.L597R	8	c.1798G>T	28.86
NRAS	Seq	DNA tumor	Likely Pathogenic	p.S33P	2	c.100G>A	9.78
BTK	Seq	DNA tumor	Benign	p.Q157X	11	c	3.59
ERBB2	Seq	DNA tumor	Pathogenic	p.Q338H	14	c.1014G>C	12.82

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
CDC73	Seq	DNA tumor	Variant of uncertain significance	p.W515L	11	c.1543_1544delinsAA	15.37
CSF3R	Seq	DNA tumor	Variant of uncertain significance	p.H362R	9	c.1085A>G	9.84
FGFR4	Seq	DNA tumor	Variant of uncertain significance	p.R225X	19	3	14.49
ARID2	Seq	DNA tumor	Variant of uncertain significance	p.D1810H	5	c.5428G>C	3.52
CTNNB1	Seq	DNA tumor	Variant of uncertain significance	p.N159Y	15	.	2.23

Immunohistochemistry Results

Biomarker	Result
PTEN	Negative 2+, 50%
PMS2	Positive 1+, 6%
PR	Negative 3+, 58%
MLH1	Positive 3+, 80%
AR	Positive 3+, 38%

Biomarker	Result
MSH6	Positive 3+, 75%
ERBB2	Positive 2+, 15%
MSH2	Positive 3+, 74%
PD-L1(SP142)	Negative 1+, 84%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

FGFR4 CDKN2A FOXL2 NT5C2

Specimen Information

Specimen ID: 5135
Specimen Recieved: 2023-06-23
Gross Description: 5135

Specimen Collected: 2023-06-23
Testing Initiated: 2023-06-23

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