Patient

Name: Timothy Fischer Date of Birth: 1998-11-21

Sex: Male

Case Number: 3346

Diagnosis: Papillary thyroid cancer

Specimen Information

Primary Tumor Site: Thyroid Specimen Site: Thyroid gland

Specimen ID: 8303

Specimen Collected: 2023-08-06

Test Initiated: 2023-08-06

Ordered By

Biomarker	Method	Analyte	Result		Therapy association		
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	
ТМВ	seq	DNA tumor	17 m/Mb High	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	Negative 2+, 57%
PR	IHC	Protien	Negative 3+, 17%
H3.3	Seq	DNA-Tumor	Stable

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Low
Tumor mutational burden	Seq	DNA tumor	17 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	Low - 13% 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 %
JAK2	Seq	DNA tumor	Likely Benign	p.P95X	7	G	15.33
CDC73	Seq	DNA tumor	Likely Benign	p.R132V	19	c.395G>A	7.24
BRAF	Seq	DNA tumor	Likely Benign	p.G12S	8	c.34G>A	2.52

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
ABL1	Seq	DNA tumor	Variant of uncertain significance	p.W266*	19	c.798G>A	26.64
MYCN	Seq	DNA tumor	Variant of uncertain significance	p.G1954R	16	c.5664C>A	15.29
NCSTN	Seq	DNA tumor	Variant of uncertain significance	p.A572G	13	3	15.9
RB1	Seq	DNA tumor	Variant of uncertain significance	p.V49M	3	c.145G>A	14.48
CDC73	Seq	DNA tumor	Variant of uncertain significance	p.C134W	6	c.402C>G	5.45

Immunohistochemistry Results

Biomarker	Result
PR	Positive 1+, 62%
AR	Negative 3+, 100%
MSH6	Negative 3+, 40%
PD-L1(SP142)	Positive 2+, 65%
ERBB2	Positive 3+, 82%

Biomarker	Result
MLH1	Positive 3+, 94%
PTEN	Positive 3+, 42%
ER	Positive 2+, 2%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

HSP90B1 ERBB2 HBB DPYD CALR MSH2

Specimen Information

Specimen ID: 8303 Specimen Collected: 2023-08-06 Specimen Recieved: 2023-08-06 Testing Initiated: 2023-08-06

Gross Description: 8303

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			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		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		