Final Report

Patient

Name: John Parker
Date of Birth: 1960-08-17
Sex: Female
Case Number: 6467
Diagnosis: Medullary thyroid cancer

Specimen Information

Primary Tumor Site: Thyroid
Specimen Site: Thyroid gland
Specimen ID: 8001
Specimen Collected: 2024-04-09
Test Initiated: 2024-04-09

Ordered By

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	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
CHD6	Seq	DNA-Tumor	Stable
PTPN11	Seq	DNA-Tumor	Stable
NRAS	Seq	RNA-Tumor	Fusion not detected
HBA2	Seq	RNA-Tumor	Fusion not detected
CALR	Seq	RNA-Tumor	Stable

BioMarker	Method	Analyte	Result
Mismatch repair status	IHC	Protien	Negative| 2+, 64%
ER	IHC	Protien	Negative| 1+, 10%
AR	IHC	Protien	Negative| 1+, 83%

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	High
Tumor mutational burden	Seq	DNA tumor	17 mutations/Mb High
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 39% 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 %
EZH2	Seq	DNA tumor	Benign	p.L239R	12	c.716T>G	3.69
CSF3R	Seq	DNA tumor	Likely Pathogenic	p.N713_A714insKGKGGG	20	c.1924A>C	1.0
PTCH1	Seq	DNA tumor	Likely Pathogenic	p.E17K	2	c.49G>A	26.93
PRKCA	Seq	DNA tumor	Likely Pathogenic	p.S346P	13	G	23.11
JAK2	Seq	DNA tumor	Pathogenic	p.F332V	2	G	12.13
RNF43	Seq	DNA tumor	Benign	p.V842I	6	c.2325_2326ins12	9.83

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
HSD3B1	Seq	DNA tumor	Variant of uncertain significance	p.L367fs*46	2	c.1092_1143del52	25.53
EGFR	Seq	DNA tumor	Variant of uncertain significance	p.E122K	9	c.19G>A	3.96
H3.3	Seq	DNA tumor	Variant of uncertain significance	p.S163P	12	c.487T>C	19.22
FOXL2	Seq	DNA tumor	Variant of uncertain significance	p.D463H	10	c.1387G>C	14.75

Immunohistochemistry Results

Biomarker	Result
ER	Negative| 1+, 96%
ERBB2	Positive| 3+, 6%
PTEN	Negative| 3+, 42%
MLH1	Negative| 3+, 10%
MSH6	Negative| 3+, 86%

Biomarker	Result
MSH2	Negative| 1+, 32%
PR	Negative| 1+, 7%
AR	Negative| 1+, 76%

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

HBA2 EGFR MSH2 NTRK1 MYD88

Specimen Information

Specimen ID: 8001
Specimen Collected: 2024-04-09
Specimen Recieved: 2024-04-09
Testing Initiated: 2024-04-09
Gross Description: 8001

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	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**	**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