

Patient	Specimen Information	Ordered By
Name: Megan Chapman	Primary Tumor Site: Colorectal	
Date of Birth: 1923-12-08	Specimen Site: Mesentery	
Sex: Female	Specimen ID: 3454	
Case Number: 6331	Specimen Collected: 2023-12-03	
Diagnosis: Gastrointestinal stromal tumors (GIST)	Test Initiated: 2023-12-03	

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	7 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
NTRK1	Seq	RNA-Tumor	Mutation not detected
Mismatch repair status	IHC	Protien	Negative 2+, 88%
HSD3B1	Seq	RNA-Tumor	Stable
PKLR	Seq	RNA-Tumor	Fusion not detected
MYOD1	Seq	RNA-Tumor	Stable

BioMarker	Method	Analyte	Result
AR	IHC	Protien	Positive 2+, 33%
FOXL2	Seq	RNA-Tumor	Fusion not detected
Mismatch repair status	IHC	Protien	Positive 1+, 82%
NF1	Seq	DNA-Tumor	Mutation not detected

Genomic Signatures

BioMarker	Method	Analyte	Result
Microsatellite instability	Seq	DNA tumor	Low
Tumor mutational burden	Seq	DNA tumor	7 mutations/Mb Low
Genomic loss of heterozygosity (LOH)	Seq	DNA tumor	High - 24% 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 %
MYD88	Seq	DNA tumor	Pathogenic	p.F332V	3	>	18.53
HDAC1	Seq	DNA tumor	Likely Pathogenic	p.R132L	9	c.395_396inv	28.29
SMO	Seq	DNA tumor	Benign	p.V701X	3	A	14.26

Gene Variants of Unknown Significance

Gene	Method	Analyte	Variant Interpretation	Protien Alteration	Exon	DNA Alteration	Allele Frequency %
IDH2	Seq	DNA tumor	Variant of uncertain significance	p.I157T	19	c.470T>C	3.17
ARID2	Seq	DNA tumor	Variant of uncertain significance	p.S252W	7	c.758C>G	24.16
TLR8	Seq	DNA tumor	Variant of uncertain significance	p.R625X	19	>	18.88

Immunohistochemistry Results

Biomarker	Result	Biomarker	Result
MSH2	Positive 2+, 13%	PTEN	Positive 1+, 99%
MLH1	Negative 1+, 82%		
AR	Negative 1+, 78%		
PMS2	Negative 2+, 55%		
PD-L1(SP142)	Positive 3+, 34%		

Genes Tested with Indeterminate Results by Tumor DNA Sequencing

EZH2 CSF3R XPC FOXL2 BRAF NT5C2

Specimen Information

Specimen ID: 3454

Specimen Collected: 2023-12-03

Specimen Recieved: 2023-12-03

Testing Initiated: 2023-12-03

Gross Description: 3454

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