# Final Report

## Patient

Name: Jack Richardson
Date of Birth: 1986-11-06
Sex: Male
Case Number: 5086
Diagnosis: Adenocarcinoma

## Specimen Information

Primary Tumor Site: Colorectal
Specimen Site: Perirectal lymph nodes
Specimen ID: 4773
Specimen Collected: 2024-01-08
Test Initiated: 2024-01-08

## 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 | 13 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 |
|---|---|---|---|
| BRCA2 | Seq | RNA-Tumor | Mutation not detected |
| TOP2A | Seq | DNA-Tumor | Mutation not detected |
| NTRK1 | Seq | RNA-Tumor | Mutation not detected |
| MAPK1 | Seq | RNA-Tumor | Fusion not detected |
| ER | IHC | Protien | Positive\| 2+, 5% |

| BioMarker | Method | Analyte | Result |
|---|---|---|---|
| AR | IHC | Protien | Positive\| 1+, 74% |
| Mismatch repair status | IHC | Protien | Positive\| 1+, 16% |
| ARID2 | Seq | RNA-Tumor | Fusion not detected |
| STAT5B | Seq | DNA-Tumor | Stable |

## Genomic Signatures

| BioMarker | Method | Analyte | Result |
|---|---|---|---|
| Microsatellite instability | Seq | DNA tumor | Equivocal |
| Tumor mutational burden | Seq | DNA tumor | 13 mutations/Mb High |
| Genomic loss of heterozygosity (LOH) | Seq | DNA tumor | High - 33% 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 % |
|---|---|---|---|---|---|---|---|
| FLT3 | Seq | DNA tumor | Benign | p.G12D | 13 | c.35G>T | 28.22 |
| PTCH1 | Seq | DNA tumor | Pathogenic | p.K656E | 19 | c.1966A>G | 17.97 |
| MAP2K2 | Seq | DNA tumor | Likely Pathogenic | p.S249C | 13 | c.1948A>G | 2.2 |
| RB1 | Seq | DNA tumor | Likely Benign | p.K656E | 19 | c.1966A>G | 6.39 |

## Gene Variants of Unknown Significance

| Gene | Method | Analyte | Variant Interpretation | Protien Alteration | Exon | DNA Alteration | Allele Frequency % |
|---|---|---|---|---|---|---|---|
| MTOR | Seq | DNA tumor | Variant of uncertain significance | p.W266* | 7 | c.798G>A | 11.42 |
| HDAC1 | Seq | DNA tumor | Variant of uncertain significance | p.P80R | 17 | c.547G>A | 17.92 |
| ARID2 | Seq | DNA tumor | Variant of uncertain significance | p.Q61X | 11 | c.34G>C | 14.95 |
| ARHGAP45 | Seq | DNA tumor | Variant of uncertain significance | p.C481X | 11 | G | 4.91 |

## Immunohistochemistry Results

| Biomarker | Result |
|---|---|
| PMS2 | Positive\| 3+, 17% |
| MLH1 | Negative\| 2+, 54% |
| ER | Negative\| 2+, 86% |
| PTEN | Positive\| 1+, 73% |
| ERBB2 | Positive\| 2+, 53% |

| Biomarker | Result |
|---|---|
| PR | Positive\| 3+, 55% |
| MSH6 | Negative\| 3+, 8% |

## Genes Tested with Indeterminate Results by Tumor DNA Sequencing

FLT3 JAK3 SDHB ALDH2 PAX5 ARID2

## Specimen Information

Specimen ID: 4773
Specimen Collected: 2024-01-08
Specimen Recieved: 2024-01-08
Testing Initiated: 2024-01-08
Gross Description: 4773

**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 |