**Patient** 

Name: Michelle Stanley
Date of Birth: 1937-09-15

Sex: Male

Case Number: 4092 Diagnosis: Lymphoma **Specimen Information** 

Primary Tumor Site: Colorectal

Specimen Site: Rectum Specimen ID: 3849

Specimen Collected: 2023-07-20

Test Initiated: 2023-07-20

## Ordered By

| Biomarker | Method | Analyte   | Result              |                   | Biomarker level   |         |
|-----------|--------|-----------|---------------------|-------------------|---|---------|
| ER        | IHC    | protein   | Positive   3+, 100% | BENEFIT           | abemaciclib, palbociclib, ribociclib,<br>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<br>BENEFT | trastuzumab, ado-trastuzumab<br>emtansine, pertuzumab,<br>fam-trastuzumab deruxtecan-nxki,<br>lapatinib, neratinib, tucatinib | level 1 |

## Cancer-Type Relevant Biomarkers

| BioMarker | Method | Analyte   | Result                |
|-----------|--------|-----------|-----------------------|
| ARID2     | Seq    | RNA-Tumor | Fusion not detected   |
| FBXW7     | Seq    | DNA-Tumor | Fusion not detected   |
| ER        | IHC    | Protien   | Negative  2+, 63%     |
| ALK       | Seq    | RNA-Tumor | Mutation not detected |
| MUTYH     | Seq    | RNA-Tumor | Mutation not detected |

| BioMarker              | Method | Analyte   | Result                |
|------------------------|--------|-----------|-----------------------|
| DNMT3A                 | Seq    | DNA-Tumor | Fusion not detected   |
| PD-L1(SP142)           | IHC    | Protien   | Negative  1+, 73%     |
| RNF43                  | Seq    | DNA-Tumor | Mutation not detected |
| Mismatch repair status | IHC    | Protien   | Negative  1+, 55%     |

# 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 - 6% 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 % |
|-------|--------|-----------|------------------------|--------------------|------|----------------|--------------------|
| DPYD  | Seq    | DNA tumor | Benign                 | p.C481X            | 2    | 3              | 9.29               |
| MYOD1 | Seq    | DNA tumor | Pathogenic             | p.G12X             | 16   | c.436G>A       | 11.46              |
| FBXW7 | Seq    | DNA tumor | Benign                 | p.N515H            | 2    | С              | 19.26              |
| CSF1R | Seq    | DNA tumor | Benign                 | p.A298P            | 15   | c.115A>C       | 20.19              |
| JAK2  | Seq    | DNA tumor | Benign                 | p.A298P            | 16   | c.892G>C       | 7.41               |
| IKZF1 | Seq    | DNA tumor | Benign                 | p.S2309Cfs*10      | 2    | c.4394A>G      | 11.91              |

## Gene Variants of Unknown Significance

| Gene  | Method | Analyte   | Variant Interpretation            | Protien Alteration | Exon | DNA Alteration | Allele Frequency % |
|-------|--------|-----------|-----------------------------------|--------------------|------|----------------|--------------------|
| CALR  | Seq    | DNA tumor | Variant of uncertain significance | p.L857P            | 19   | 3              | 11.62              |
| NCSTN | Seq    | DNA tumor | Variant of uncertain significance | p.P128L            | 17   | c.383C>T       | 22.52              |
| TEK   | Seq    | DNA tumor | Variant of uncertain significance | p.E455fs*7         | 17   | С              | 13.77              |
| NF1   | Seq    | DNA tumor | Variant of uncertain significance | p.1960V            | 9    | c.2624C>A      | 14.99              |
| SDHD  | Seq    | DNA tumor | Variant of uncertain significance | p.D842V            | 17   | c.2525A>T      | 5.97               |

## Immunohistochemistry Results

| Biomarker | Result            |
|-----------|-------------------|
| MSH2      | Negative  3+, 29% |
| PR        | Positive  1+, 69% |
| PMS2      | Positive  1+, 69% |
| ER        | Negative  3+, 14% |
| MSH6      | Negative  2+, 4%  |

| Biomarker    | Result            |
|--------------|-------------------|
| MLH1         | Positive  3+, 97% |
| AR           | Negative  3+, 95% |
| ERBB2        | Negative  3+, 49% |
| PD-L1(SP142) | Negative  3+, 77% |

## Genes Tested with Indeterminate Results by Tumor DNA Sequencing

MAPK1 KRAS CD74 CTNNB1 HDAC1 BTK

## **Specimen Information**

Specimen ID: 3849 Specimen Collected: 2023-07-20 Specimen Recieved: 2023-07-20 Testing Initiated: 2023-07-20

Gross Description: 3849

### **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 Analyt |        |     | 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 Biomai                |        | 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           |  |  |  |