# Final Report

## Patient

Name: Mitchell Mcdonald
Date of Birth: 1974-10-08
Sex: Female
Case Number: 8436
Diagnosis: Acute myeloid leukemia (AML)

## Specimen Information

Primary Tumor Site: Leukemia
Specimen Site: Spleen
Specimen ID: 5155
Specimen Collected: 2023-09-09
Test Initiated: 2023-09-11

## 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 | 18 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 |
|---|---|---|---|
| MYOD1 | Seq | DNA-Tumor | Stable |
| DPYD | Seq | RNA-Tumor | Mutation not detected |
| PKLR | Seq | DNA-Tumor | Stable |
| BRCA1 | Seq | DNA-Tumor | Mutation not detected |
| MAP2K2 | Seq | RNA-Tumor | Fusion not detected |

| BioMarker | Method | Analyte | Result |
|---|---|---|---|
| FBXW7 | Seq | DNA-Tumor | Mutation not detected |
| FOXL2 | Seq | DNA-Tumor | Stable |
| PIK3CA | Seq | DNA-Tumor | Stable |
| ARID2 | Seq | RNA-Tumor | Mutation not detected |
| ERBB2 | Seq | DNA-Tumor | Fusion not detected |

## Genomic Signatures

| BioMarker | Method | Analyte | Result |
|---|---|---|---|
| Microsatellite instability | Seq | DNA tumor | Low |
| Tumor mutational burden | Seq | DNA tumor | 18 mutations/Mb High |
| Genomic loss of heterozygosity (LOH) | Seq | DNA tumor | High - 18% 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 % |
|---|---|---|---|---|---|---|---|
| IKZF1 | Seq | DNA tumor | Pathogenic | p.R465H | 16 | c.1394G>A | 9.77 |
| ARID2 | Seq | DNA tumor | Benign | p.G12F | 20 | c.38G>T | 23.38 |
| PKLR | Seq | DNA tumor | Likely Benign | p.A374E | 9 | G | 1.78 |
| PRKCA | Seq | DNA tumor | Likely Pathogenic | p.R1012X | 18 | G | 10.38 |
| CSF1R | Seq | DNA tumor | Likely Benign | p.L747_A750del | 2 | c.2155G>A | 4.54 |

## Gene Variants of Unknown Significance

| Gene | Method | Analyte | Variant Interpretation | Protien Alteration | Exon | DNA Alteration | Allele Frequenc |
|---|---|---|---|---|---|---|---|
| HDAC2 | Seq | DNA tumor | Variant of uncertain significance | p.A374E | 17 | 3 | 9.66 |
| MET | Seq | DNA tumor | Variant of uncertain significance | p.N713_A714insKGKGGG | 14 | c.1924A>C | 3.07 |
| TOP2A | Seq | DNA tumor | Variant of uncertain significance | p.Y736fs*4 | 10 | c.2207_2212delinsTAGATTC | 7.12 |
| DDR2 | Seq | DNA tumor | Variant of uncertain significance | p.T367N | 17 | c.1100= | 12.93 |
| MPL | Seq | DNA tumor | Variant of uncertain significance | p.S163P | 8 | c.487T>C | 2.43 |

## Immunohistochemistry Results

| Biomarker | Result |
|---|---|
| AR | Positive\| 1+, 95% |
| MSH6 | Positive\| 3+, 36% |
| PMS2 | Positive\| 3+, 86% |

## Genes Tested with Indeterminate Results by Tumor DNA Sequencing

ARHGAP45 XPC DDR2 FBXW7 FLT3 ALK

## Specimen Information

Specimen ID: 5155

Specimen Collected: 2023-09-09

Specimen Recieved: 2023-09-11

Testing Initiated: 2023-09-11

Gross Description: 5155

**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 |