# Final Report

## Patient

Name: Melissa Dean
Date of Birth: 2003-09-27
Sex: Male
Case Number: 7591
Diagnosis: Melanoma

## Specimen Information

Primary Tumor Site: Skin
Specimen Site: Sentinel lymph node
Specimen ID: 5251
Specimen Collected: 2023-10-21
Test Initiated: 2023-10-21

## 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 | 14 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 |
|---|---|---|---|
| ER | IHC | Protien | Positive\| 2+, 77% |
| TP53 | Seq | RNA-Tumor | Stable |
| PR | IHC | Protien | Negative\| 2+, 17% |
| RNF43 | Seq | DNA-Tumor | Stable |
| PD-L1(SP142) | IHC | Protien | Positive\| 1+, 69% |

## Genomic Signatures

| BioMarker | Method | Analyte | Result |
|---|---|---|---|
| Microsatellite instability | Seq | DNA tumor | Low |
| Tumor mutational burden | Seq | DNA tumor | 14 mutations/Mb High |
| Genomic loss of heterozygosity (LOH) | Seq | DNA tumor | High - 31% 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 % |
|---|---|---|---|---|---|---|---|
| BRAF | Seq | DNA tumor | Benign | p.R337H | 17 | c.523C>T | 26.44 |
| TOP2A | Seq | DNA tumor | Benign | p.N452D | 7 | c.1354A>G | 5.73 |
| NTRK1 | Seq | DNA tumor | Likely Benign | p.T1087I | 6 | c.3497T>G | 12.56 |
| JAK2 | Seq | DNA tumor | Benign | p.Y303H | 9 | G | 12.73 |
| PIK3CA | Seq | DNA tumor | Pathogenic | p.Q61R | 8 | c.182A>T | 5.63 |

## Gene Variants of Unknown Significance

| Gene | Method | Analyte | Variant Interpretation | Protien Alteration | Exon | DNA Alteration | Allele Frequency % |
|---|---|---|---|---|---|---|---|
| MSH2 | Seq | DNA tumor | Variant of uncertain significance | p.Q177R | 4 | c.530A>G | 12.45 |
| ARHGAP45 | Seq | DNA tumor | Variant of uncertain significance | p.Y303H | 15 | 3 | 1.81 |
| NTRK1 | Seq | DNA tumor | Variant of uncertain significance | p.D463H | 7 | c.1387G>C | 4.59 |
| IDH1 | Seq | DNA tumor | Variant of uncertain significance | p.A636P | 10 | c.1906G>C | 12.66 |
| ARHGAP45 | Seq | DNA tumor | Variant of uncertain significance | p.G751A | 4 | 3 | 3.83 |
| HRAS | Seq | DNA tumor | Variant of uncertain significance | p.H1047L | 7 | c.1625A>T | 2.74 |

## Immunohistochemistry Results

| Biomarker | Result |
|---|---|
| ERBB2 | Negative\| 1+, 88% |
| PTEN | Negative\| 1+, 57% |
| MSH2 | Negative\| 1+, 91% |

## Genes Tested with Indeterminate Results by Tumor DNA Sequencing

CDC73 IDH2 CHEK2 DDR2

## Specimen Information

Specimen ID: 5251

Specimen Collected: 2023-10-21

Specimen Recieved: 2023-10-21

Testing Initiated: 2023-10-21

Gross Description: 5251

**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 |