# Final Report

## Patient

Name: Christina Williams
Date of Birth: 1966-05-29
Sex: Female
Case Number: 2025
Diagnosis: Adenocarcinoma

## Specimen Information

Primary Tumor Site: Lung
Specimen Site: Lung lobes (upper
Specimen ID: 7239
Specimen Collected: 2023-10-29
Test Initiated: 2023-10-30

## 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 | 7 m/Mb Low | 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 |
|---|---|---|---|
| KLF1 | Seq | RNA-Tumor | Stable |
| AR | IHC | Protien | Positive\| 2+, 33% |
| PD-L1(SP142) | IHC | Protien | Negative\| 2+, 82% |
| IDH2 | Seq | DNA-Tumor | Stable |

## Genomic Signatures

| BioMarker | Method | Analyte | Result |
|---|---|---|---|
| Microsatellite instability | Seq | DNA tumor | High |
| Tumor mutational burden | Seq | DNA tumor | 7 mutations/Mb Low |
| Genomic loss of heterozygosity (LOH) | Seq | DNA tumor | High - 40% 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 % |
|---|---|---|---|---|---|---|---|
| ALK | Seq | DNA tumor | Pathogenic | p.G1954R | 5 | c.5664C>G | 32.11 |
| FGFR1 | Seq | DNA tumor | Likely Benign | p.I1600M | 12 | c.4800C>G | 31.45 |
| SF3B1 | Seq | DNA tumor | Pathogenic | p.E322K | 16 | c.964G>A | 9.73 |
| ARHGAP45 | Seq | DNA tumor | Benign | p.I1600M | 18 | c.4800C>G | 24.34 |
| HBA2 | Seq | DNA tumor | Pathogenic | p.L576P | 18 | c.1669T>C | 8.35 |

## Gene Variants of Unknown Significance

| Gene | Method | Analyte | Variant Interpretation | Protien Alteration | Exon | DNA Alteration | Allele Frequency % |
|---|---|---|---|---|---|---|---|
| KLF1 | Seq | DNA tumor | Variant of uncertain significance | p.L857P | 7 | 1 | 2.34 |
| NCSTN | Seq | DNA tumor | Variant of uncertain significance | p.N515H | 13 | A | 18.34 |
| MYOD1 | Seq | DNA tumor | Variant of uncertain significance | p.K666X | 17 | c | 16.09 |

## Immunohistochemistry Results

| Biomarker | Result |
|---|---|
| ER | Positive\| 2+, 61% |
| PD-L1(SP142) | Negative\| 3+, 7% |
| MLH1 | Negative\| 1+, 18% |
| AR | Positive\| 2+, 59% |
| MSH6 | Negative\| 1+, 88% |

| Biomarker | Result |
|---|---|
| PR | Negative\| 3+, 50% |
| PTEN | Positive\| 2+, 69% |

## Genes Tested with Indeterminate Results by Tumor DNA Sequencing

MAPK1 CTNNB1 APC ZEB2 PTCH1 HDAC1

## Specimen Information

Specimen ID: 7239

Specimen Collected: 2023-10-29

Specimen Recieved: 2023-10-30

Testing Initiated: 2023-10-30

Gross Description: 7239

**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 |