



## Sample Information

**Patient Name:** 李雅芬

**Gender:** Female

**ID No.:** F225478637

**History No.:** 49792167

**Age:** 43

**Ordering Doctor:** DOC8170B 陳亭如

**Ordering REQ.:** 0CWLUHT

**Signing in Date:** 2024/1/30

**Path No.:** M113-00029

**MP No.:** BR24010

**Assay:** Oncomine BRCA1/2 Assay

**Sample Type:** Blood

**Date of blood drawing:** 2024/1/25

**Reporting Doctor:** DOC5424G 彭昱璟 (Phone: 8#5424)

**Note:**

## Sample Cancer Type: Breast Cancer

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### Report Highlights

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## Relevant Breast Cancer Variants

| Gene  | Finding       |
|-------|---------------|
| BRCA1 | None detected |
| BRCA2 | None detected |

## Relevant Biomarkers

No clinically significant biomarkers found in this sample.

## Variant Details

### DNA Sequence Variants

| Gene  | Amino Acid Change | Coding    | Locus          | Allele Frequency | Transcript  | Variant Effect | ClinVar <sup>1</sup> | Coverage |
|-------|-------------------|-----------|----------------|------------------|-------------|----------------|----------------------|----------|
| BRCA2 | p.(N372H)         | c.1114A>C | chr13:32906729 | 50.02%           | NM_000059.3 | missense       | Benign               | 4000     |
| BRCA2 | p.(V1269=)        | c.3807T>C | chr13:32912299 | 50.39%           | NM_000059.3 | synonymous     | Benign               | 3995     |

<sup>1</sup> Based on Clinvar version 20200329

Variant Details (continued)

DNA Sequence Variants (continued)

| Gene  | Amino Acid Change | Coding    | Locus          | Allele Frequency | Transcript  | Variant Effect | ClinVar <sup>1</sup>                         | Coverage |
|-------|-------------------|-----------|----------------|------------------|-------------|----------------|--|----------|
| BRCA2 | p.(L1521=)        | c.4563A>G | chr13:32913055 | 99.94%           | NM_000059.3 | synonymous     | Benign                                       | 3170     |
| BRCA2 | p.(V2109I)        | c.6325G>A | chr13:32914817 | 49.84%           | NM_000059.3 | missense       | Conflicting interpretations of pathogenicity | 3058     |
| BRCA2 | p.(V2171=)        | c.6513G>C | chr13:32915005 | 100.00%          | NM_000059.3 | synonymous     | Benign                                       | 3999     |
| BRCA2 | p.(V2466A)        | c.7397T>C | chr13:32929387 | 99.80%           | NM_000059.3 | missense       | Benign                                       | 3998     |
| BRCA2 | p.(P3292L)        | c.9875C>T | chr13:32972525 | 52.43%           | NM_000059.3 | missense       | Benign                                       | 3994     |

<sup>1</sup> Based on Clinvar version 20200329