

Department of Pathology and Laboratory Medicine No.201, Sec. 2, Shipai Rd., Beitou District, Taipei City, Taiwan 11217, R.O.C. Tel: 02-2875-7449

Date: 23 Jun 2022

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Sample Information

Patient Name: 詹麗玲 Gender: Female ID No.: K221022491 History No.: 45370352

Age: 54

Ordering Doctor: DOC2095G 蔡宜芳 Ordering REQ.: D6NP6D1 Signing in Date: 2022/06/23

Path No.: S111-99614 **MP No.:** BR22037

Assay: Oncomine BRCA1/2 Assay

Sample Type: Blood

Date of blood drawing: 2022/06/15

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

Note:

Sample Cancer Type: Breast Cancer

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Variant Details

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Relevant Biomarkers

No clinically significant biomarkers found in this sample.

Variant Details

DNA Sequence Variants

| | | | | Allele | | | | _ |
|-------|-------------------|-----------|----------------|-----------|-------------|----------------|---------|----------|
| Gene | Amino Acid Change | Coding | Locus | Frequency | Transcript | Variant Effect | ClinVar | Coverage |
| BRCA2 | p.(N289H) | c.865A>C | chr13:32906480 | 48.99% | NM_000059.3 | missense | Benign | 2621 |
| BRCA2 | p.(S455=) | c.1365A>G | chr13:32906980 | 42.69% | NM_000059.3 | synonymous | Benign | 3999 |
| BRCA2 | p.(H743=) | c.2229T>C | chr13:32910721 | 49.36% | NM_000059.3 | synonymous | Benign | 3999 |
| BRCA2 | p.(N991D) | c.2971A>G | chr13:32911463 | 50.71% | NM_000059.3 | missense | Benign | 2406 |
| BRCA2 | p.(V1269=) | c.3807T>C | chr13:32912299 | 51.49% | NM_000059.3 | synonymous | Benign | 3991 |
| BRCA2 | p.(L1521=) | c.4563A>G | chr13:32913055 | 99.87% | NM_000059.3 | synonymous | Benign | 3156 |
| BRCA2 | p.(V2171=) | c.6513G>C | chr13:32915005 | 100.00% | NM_000059.3 | synonymous | Benign | 3999 |

¹ Based on Clinvar version 20200329

Variant Details (continued)

DNA Sequence Variants (continued)

| Gene | Amino Acid Change | Coding | Locus | Allele Frequency | Transcript | Variant Effect | ClinVar ¹ | Coverage |
|-------|-------------------|-----------|----------------|---------------------|-------------|----------------|----------------------|----------|
| BRCA2 | p.(V2466A) | c.7397T>C | chr13:32929387 | 99.82% | NM_000059.3 | missense | Benign | 3413 |
| BRCA1 | p.(S1613G) | c.4837A>G | chr17:41223094 | 49.74% | NM_007294.4 | missense | Benign | 3983 |
| BRCA1 | p.(S1436=) | c.4308T>C | chr17:41234470 | 49.71% | NM_007294.4 | synonymous | Benign | 3997 |
| BRCA1 | p.(K1183R) | c.3548A>G | chr17:41244000 | 51.30% | NM_007294.4 | missense | Benign | 4000 |
| BRCA1 | p.(E1038G) | c.3113A>G | chr17:41244435 | 51.91% | NM_007294.4 | missense | Benign | 3997 |
| BRCA1 | p.(P871L) | c.2612C>T | chr17:41244936 | 48.49% | NM_007294.4 | missense | Benign | 3997 |
| BRCA1 | p.(L771=) | c.2311T>C | chr17:41245237 | 51.07% | NM_007294.4 | synonymous | Benign | 3726 |
| BRCA1 | p.(S694=) | c.2082C>T | chr17:41245466 | 49.55% | NM_007294.4 | synonymous | Benign | 4000 |

¹ Based on Clinvar version 20200329

Date: 23 Jun 2022

Signatures

Testing Personnel:

Laboratory Supervisor:

Pathologist: