

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: 06 Oct 2022 1 of 3

## Sample Information

Patient Name: 楊雅琴 Gender: Female ID No.: Q223633012 History No.: 48780978

**Age:** 35

Ordering Doctor: DOC2394E 許瓈文 Ordering REQ.: OCAMDDY **Signing in Date: 2022/10/06** 

**Path No.:** S111-97928 MP No.: BR22056

Assay: Oncomine BRCA1/2 Assay

Sample Type: Blood

Date of blood drawing: 2022/09/26

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

Note:

## Sample Cancer Type: Breast Cancer

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## **Relevant Biomarkers**

**DNA Sequence Variants** 

No clinically significant biomarkers found in this sample.

c.3396A>G

c.4563A>G

### **Variant Details**

### Allele **Amino Acid Change** Variant Effect ClinVar1 Gene Coding Locus Frequency Transcript Coverage BRCA2 p.(?) c.-26G>A chr13:32890572 54.09% NM\_000059.3 unknown Benign 3997 BRCA2 p.(N289H) c.865A>C chr13:32906480 49.67% NM 000059.3 missense Benign 1198 BRCA2 p.(S455=) c.1365A>G chr13:32906980 50.96% NM\_000059.3 synonymous Benign 3999 BRCA2 p.(H743=) c.2229T>C chr13:32910721 52.43% NM\_000059.3 Benign 3998 synonymous BRCA2 p.(N991D) c.2971A>G chr13:32911463 56.06% NM\_000059.3 missense Benign 132

50.89% NM\_000059.3

99.95% NM\_000059.3

synonymous

synonymous

Benign

Benign

3995

3995

p.(K1132=)

BRCA2

BRCA2

chr13:32911888

chr13:32913055

p.(L1521=) 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 <sup>1</sup>	Coverage
BRCA2	p.(V2171=)	c.6513G>C	chr13:32915005	100.00%	NM_000059.3	synonymous	Benign	2847
BRCA2	p.(S2414=)	c.7242A>G	chr13:32929232	48.13%	NM_000059.3	synonymous	Benign	4000
BRCA2	p.(V2466A)	c.7397T>C	chr13:32929387	99.85%	NM_000059.3	missense	Benign	3998
BRCA1	p.(S1613G)	c.4837A>G	chr17:41223094	47.36%	NM_007294.4	missense	Benign	3970
BRCA1	p.(S1436=)	c.4308T>C	chr17:41234470	49.37%	NM_007294.4	synonymous	Benign	2157
BRCA1	p.(K1183R)	c.3548A>G	chr17:41244000	50.80%	NM_007294.4	missense	Benign	4000
BRCA1	p.(E1038G)	c.3113A>G	chr17:41244435	53.51%	NM_007294.4	missense	Benign	3999
BRCA1	p.(P871L)	c.2612C>T	chr17:41244936	50.15%	NM_007294.4	missense	Benign	3998
BRCA1	p.(L771=)	c.2311T>C	chr17:41245237	50.00%	NM_007294.4	synonymous	Benign	3998
BRCA1	p.(S694=)	c.2082C>T	chr17:41245466	48.86%	NM_007294.4	synonymous	Benign	3997

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

Date: 06 Oct 2022

# **Signatures**

**Testing Personnel:** 

**Laboratory Supervisor:** 

Pathologist: