

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:** 20 Apr 2023 1 of 2

## **Sample Information**

Patient Name: 邱欣潔 Gender: Female ID No.: E221667128 History No.: 40622320

**Age:** 49

Ordering Doctor: DOC2095G\_蔡宜芳

Ordering REQ.: D75D3JB Signing in Date: 2023/04/20

**Path No.:** M112-00074 **MP No.:** BR23021

Assay: Oncomine BRCA1/2 Assay

Sample Type: Blood

Date of blood drawing: 2023/04/14

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

Note:

### Sample Cancer Type: Breast Cancer

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#### **Relevant Biomarkers**

No clinically significant biomarkers found in this sample.

#### **Variant Details**

# DNA Sequence Variants

			Allele				
Amino Acid Change	Coding	Locus	Frequency	Transcript	Variant Effect	ClinVar1	Coverage
p.(?)	c26G>A	chr13:32890572	51.76%	NM_000059.3	unknown	Benign	3997
p.(N372H)	c.1114A>C	chr13:32906729	50.81%	NM_000059.3	missense	Benign	2413
p.(K1132=)	c.3396A>G	chr13:32911888	53.95%	NM_000059.3	synonymous	Benign	3281
p.(L1521=)	c.4563A>G	chr13:32913055	100.00%	NM_000059.3	synonymous	Benign	1587
p.(V2171=)	c.6513G>C	chr13:32915005	100.00%	NM_000059.3	synonymous	Benign	2663
p.(S2414=)	c.7242A>G	chr13:32929232	50.14%	NM_000059.3	synonymous	Benign	2591
p.(V2466A)	c.7397T>C	chr13:32929387	99.83%	NM_000059.3	missense	Benign	1809
	p.(?) p.(N372H) p.(K1132=) p.(L1521=) p.(V2171=) p.(S2414=)	p.(?) c26G>A  p.(N372H) c.1114A>C  p.(K1132=) c.3396A>G  p.(L1521=) c.4563A>G  p.(V2171=) c.6513G>C  p.(S2414=) c.7242A>G	p.(?)       c26G>A       chr13:32890572         p.(N372H)       c.1114A>C       chr13:32906729         p.(K1132=)       c.3396A>G       chr13:32911888         p.(L1521=)       c.4563A>G       chr13:32913055         p.(V2171=)       c.6513G>C       chr13:32915005         p.(S2414=)       c.7242A>G       chr13:32929232	Amino Acid Change         Coding         Locus         Frequency           p.(?)         c26G>A         chr13:32890572         51.76%           p.(N372H)         c.1114A>C         chr13:32906729         50.81%           p.(K1132=)         c.3396A>G         chr13:32911888         53.95%           p.(L1521=)         c.4563A>G         chr13:32913055         100.00%           p.(V2171=)         c.6513G>C         chr13:32915005         100.00%           p.(S2414=)         c.7242A>G         chr13:32929232         50.14%	Amino Acid Change         Coding         Locus         Frequency         Transcript           p.(?)         c26G>A         chr13:32890572         51.76%         NM_000059.3           p.(N372H)         c.1114A>C         chr13:32906729         50.81%         NM_000059.3           p.(K1132=)         c.3396A>G         chr13:32911888         53.95%         NM_000059.3           p.(L1521=)         c.4563A>G         chr13:32913055         100.00%         NM_000059.3           p.(V2171=)         c.6513G>C         chr13:32915005         100.00%         NM_000059.3           p.(S2414=)         c.7242A>G         chr13:32929232         50.14%         NM_000059.3	Amino Acid ChangeCodingLocusFrequencyTranscriptVariant Effectp.(?)c26G>Achr13:3289057251.76%NM_000059.3unknownp.(N372H)c.1114A>Cchr13:3290672950.81%NM_000059.3missensep.(K1132=)c.3396A>Gchr13:3291188853.95%NM_000059.3synonymousp.(L1521=)c.4563A>Gchr13:32913055100.00%NM_000059.3synonymousp.(V2171=)c.6513G>Cchr13:32915005100.00%NM_000059.3synonymousp.(S2414=)c.7242A>Gchr13:3292923250.14%NM_000059.3synonymous	Amino Acid Change         Coding         Locus         Frequency         Transcript         Variant Effect         ClinVar1           p.(?)         c26G>A         chr13:32890572         51.76%         NM_000059.3         unknown         Benign           p.(N372H)         c.1114A>C         chr13:32906729         50.81%         NM_000059.3         missense         Benign           p.(K1132=)         c.3396A>G         chr13:32911888         53.95%         NM_000059.3         synonymous         Benign           p.(L1521=)         c.4563A>G         chr13:32913055         100.00%         NM_000059.3         synonymous         Benign           p.(V2171=)         c.6513G>C         chr13:32915005         100.00%         NM_000059.3         synonymous         Benign           p.(S2414=)         c.7242A>G         chr13:32929232         50.14%         NM_000059.3         synonymous         Benign

<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
BRCA1	p.(S1613G)	c.4837A>G	chr17:41223094	50.28%	NM_007294.4	missense	Benign	3994
BRCA1	p.(S1436=)	c.4308T>C	chr17:41234470	48.56%	NM_007294.4	synonymous	Benign	2887
BRCA1	p.(K1183R)	c.3548A>G	chr17:41244000	51.50%	NM_007294.4	missense	Benign	4000
BRCA1	p.(E1038G)	c.3113A>G	chr17:41244435	51.43%	NM_007294.4	missense	Benign	3743
BRCA1	p.(P871L)	c.2612C>T	chr17:41244936	47.33%	NM_007294.4	missense	Benign	4000
BRCA1	p.(L771=)	c.2311T>C	chr17:41245237	51.35%	NM_007294.4	synonymous	Benign	1893
BRCA1	p.(S694=)	c.2082C>T	chr17:41245466	51.54%	NM_007294.4	synonymous	Benign	3999

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