

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: 29 Dec 2021 1 of 3

Sample Information

Patient Name: 鄭雯怡 Gender: Female ID No.: A225231853 History No.: 34324259

Age: 40

Ordering Doctor: DOC2402E 毛士豪 Ordering REQ.: 0BQHFZA Signing in Date: 2021/12/29

Path No.: S110-94998 **MP No.:** BR21061

Assay: Oncomine BRCA Assay

Sample Type: Blood

Date of blood drawing: 2021/12/23

Reporting Doctor: DOC5452C 周德盈 (Phone: 8#5452)

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

Amino Acid Change	Coding	Locus	Allele Frequency	Transcript	Variant Effect	ClinVar ¹	Coverage
p.(N289H)	c.865A>C	chr13:32906480	53.71%	NM_000059.3	missense	Benign	996
p.(N372H)	c.1114A>C	chr13:32906729	49.39%	NM_000059.3	missense	Benign	3999
p.(S455=)	c.1365A>G	chr13:32906980	49.00%	NM_000059.3	synonymous	Benign	3996
p.(H743=)	c.2229T>C	chr13:32910721	50.71%	NM_000059.3	synonymous	Benign	3995
p.(N991D)	c.2971A>G	chr13:32911463	51.45%	NM_000059.3	missense	Benign	3998
p.(L1521=)	c.4563A>G	chr13:32913055	99.94%	NM_000059.3	synonymous	Benign	3479
p.(V2171=)	c.6513G>C	chr13:32915005	99.95%	NM_000059.3	synonymous	Benign	3998
	p.(N289H) p.(N372H) p.(S455=) p.(H743=) p.(N991D) p.(L1521=)	p.(N289H) c.865A>C p.(N372H) c.1114A>C p.(S455=) c.1365A>G p.(H743=) c.2229T>C p.(N991D) c.2971A>G p.(L1521=) c.4563A>G	p.(N289H) c.865A>C chr13:32906480 p.(N372H) c.1114A>C chr13:32906729 p.(S455=) c.1365A>G chr13:32906980 p.(H743=) c.2229T>C chr13:32910721 p.(N991D) c.2971A>G chr13:32911463 p.(L1521=) c.4563A>G chr13:32913055	Amino Acid ChangeCodingLocusFrequencyp.(N289H)c.865A>Cchr13:3290648053.71%p.(N372H)c.1114A>Cchr13:3290672949.39%p.(S455=)c.1365A>Gchr13:3290698049.00%p.(H743=)c.2229T>Cchr13:3291072150.71%p.(N991D)c.2971A>Gchr13:3291146351.45%p.(L1521=)c.4563A>Gchr13:3291305599.94%	Amino Acid ChangeCodingLocusFrequencyTranscriptp.(N289H)c.865A>Cchr13:3290648053.71%NM_000059.3p.(N372H)c.1114A>Cchr13:3290672949.39%NM_000059.3p.(S455=)c.1365A>Gchr13:3290698049.00%NM_000059.3p.(H743=)c.2229T>Cchr13:3291072150.71%NM_000059.3p.(N991D)c.2971A>Gchr13:3291146351.45%NM_000059.3p.(L1521=)c.4563A>Gchr13:3291305599.94%NM_000059.3	Amino Acid ChangeCodingLocusFrequencyTranscriptVariant Effectp.(N289H)c.865A>Cchr13:3290648053.71%NM_000059.3missensep.(N372H)c.1114A>Cchr13:3290672949.39%NM_000059.3missensep.(S455=)c.1365A>Gchr13:3290698049.00%NM_000059.3synonymousp.(H743=)c.2229T>Cchr13:3291072150.71%NM_000059.3synonymousp.(N991D)c.2971A>Gchr13:3291146351.45%NM_000059.3missensep.(L1521=)c.4563A>Gchr13:3291305599.94%NM_000059.3synonymous	Amino Acid Change Coding Locus Frequency Transcript Variant Effect ClinVar1 p.(N289H) c.865A>C chr13:32906480 53.71% NM_000059.3 missense Benign p.(N372H) c.1114A>C chr13:32906729 49.39% NM_000059.3 missense Benign p.(S455=) c.1365A>G chr13:32906980 49.00% NM_000059.3 synonymous Benign p.(H743=) c.2229T>C chr13:32910721 50.71% NM_000059.3 synonymous Benign p.(N991D) c.2971A>G chr13:32911463 51.45% NM_000059.3 synonymous Benign p.(L1521=) c.4563A>G chr13:32913055 99.94% NM_000059.3 synonymous Benign

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	99.85%	NM_000059.3	missense	Benign	3999
BRCA1	p.(S1613G)	c.4837A>G	chr17:41223094	50.75%	NM_007294.4	missense	Benign	3988
BRCA1	p.(S1436=)	c.4308T>C	chr17:41234470	47.10%	NM_007294.4	synonymous	Benign	4000
BRCA1	p.(K1183R)	c.3548A>G	chr17:41244000	51.63%	NM_007294.4	missense	Benign	4000
BRCA1	p.(E1038G)	c.3113A>G	chr17:41244435	48.60%	NM_007294.4	missense	Benign	3998
BRCA1	p.(P871L)	c.2612C>T	chr17:41244936	49.57%	NM_007294.4	missense	Benign	3998
BRCA1	p.(Y856H)	c.2566T>C	chr17:41244982	52.08%	NM_007294.4	missense	Benign	3998
BRCA1	p.(L771=)	c.2311T>C	chr17:41245237	49.90%	NM_007294.4	synonymous	Benign	4000
BRCA1	p.(S694=)	c.2082C>T	chr17:41245466	48.69%	NM_007294.4	synonymous	Benign	3999
BRCA1	p.(S265=)	c.795T>C	chr17:41246753	48.65%	NM_007294.4	synonymous	Benign	3996

¹ Based on Clinvar version 20200329

Date: 29 Dec 2021

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Signatures

Testing Personnel:

Laboratory Supervisor:

Pathologist: