Reported Date: 0000-00-00

axen BRCA

Humanizing Genomics macrogen

PATIENT

Name: Patient_name
Gender: Gender
Birth year: Birth

SPECIMEN

Specimen type: Specimen_type
Speciment ID: Tube_ID
Collected Date: 0000-00-00
Received Date: 0000-00-00

MEDICAL FACILITY

Hospital: Hospital
Ordering Physician: Doctor

PATIENT RESULT

POSITIVE

POSITIVE

Gene	Variant	Clinical Significance
BRCA1	c.1039_1040delCT (p.Leu347fs)	Pathogenic
BRCA2	No clinically significant mutation identified.	

Personal/Family History Summary

Family member	Cancer / Clinical Diagnosis	Age at Diagnosis

Limitations & Disclaimer

This test should detect the variants comprehensively but the variant list may not be completed due to the quality of the specimen or because of the technical limitation such as library preparation and sequencing bias. Clinical interpretation of the variants is based on up-to-date database but the interpretation may change as the database will be continuously updated with new study results. Clinical interpretation based on current database may not fully reflect ethnic diversity in variants and their disease correlation. Provided family history of certain genetic disease and critical clinic information more conclusive clinical interpretation may be offered. This test is neither intended nor validated for diagnosis.

Note

Decisions on care and treatment should be based on the independent medical judgment of the treating physician taking into consideration all available information concerning the patient's condition, including other laboratory test, in accordance with the standard of care in a given community. Decisions regarding care and treatment should not be based on a single test such as this one.

Method

Genomic DNA was used for the generation of the amplicon library using Axen™ BRCA1/2. The amplicon library was sequenced by MiSeqDx sequencer.

Notes for Personalized Management

Lab Director

Electronically Signed by
Sangjoon Hwang

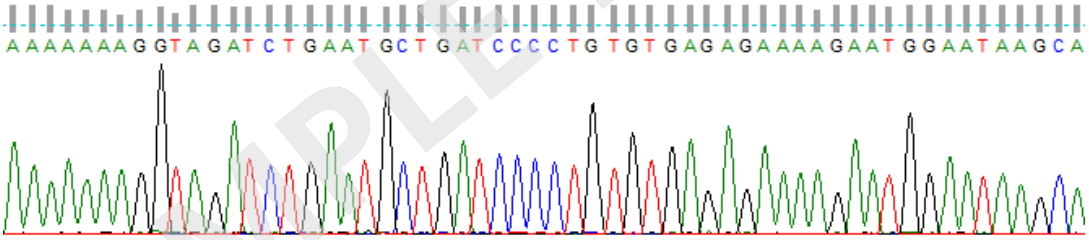
Scientific use of these results requires permission of MACROGEN.
More information is available at www.macrogen.com or ngsclinic@macrogen.com

www.macrogen.com | ngsclinic@macrogen.com | Tel. +82-2-2180-7000 | Fax. +82-2-2180-7100

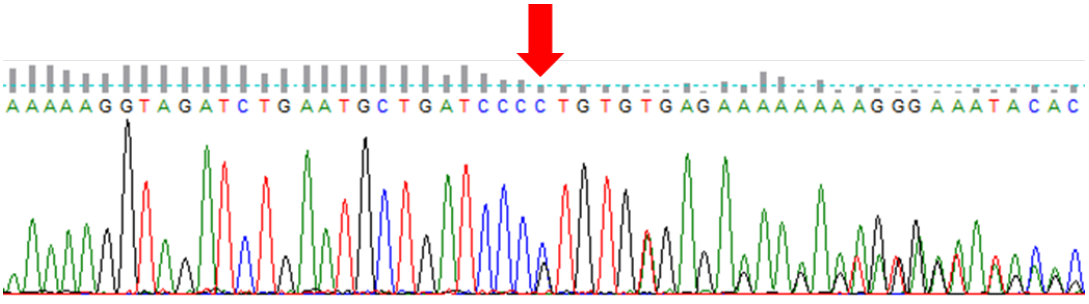
Molecular Genetic Test Report (BRCA1 / BRCA2)

BRCA1 exon10
c.1039_1040delCT (p.Leu347fs) Heterozygote → Detected

Control(-)



Patient_name



www.macrogen.com | ngsclinic@macrogen.com | Tel. +82-2-2180-7000 | Fax. +82-2-2180-7100

Target region

Gene	Transcript ID	Exon
BRCA1	NM_007294.3	Exon1-Exon24
BRCA2	NM_000059.3	Exon1-Exon27

Data analysis

Type	Name
Reference genome	GRCh37/hg19
Panel	Axen™ BRCA1/2 panel
Quality check & trimming	FASTQC, fastp
Alignment tools	BWA mem
Variant calling tools	GATK-v4.0
Annotation tools and databases	BRCA_exchange version 48 Clinvar Jun,2021

QC

RESULT

Mean Depth(X)	50X coverage(%)
3813	100.0

PASS : Mean Depth ≥ 100X, 50X coverage ≥ 90%

FAIL : Mean Depth < 100X, 50X coverage < 90%