Genetic Test Results & Clinical Report

GeneHealth UK

Patient information				Reporting information	
C4C number	54321P			Requester	Genetic Counsellor
Surname	Surname			Fax report to	01223 281 316
Forename	Forename			Sample date	16/11/16
Date of birth	03/04/88	Sex (F / M)	F	Report date	01/12/16

Clinical information	
Test requested	BreastGene panel (see footnote for genes tested)
Reason for request	Unaffected. Mother had breast cancer at age 47. Other cancers in family.

Results	
DNA sequence variants	No mutations detected in any of the BreastGene panel genes (see footnote for genes tested)
DNA copy number variants	No exonic copy number variants detected in BRCA1, BRCA2 or STK11

Interpretation and comments

NO MUTATIONS DETECTED IN THE BREASTGENE PANEL

This result reduces the likelihood of this individual developing breast or any other cancer caused by any of the predisposition genes in the BreastGene panel. This result does not exclude the small risk from other rarer heritable causes of breast cancer.

Genotype and report by	Report authorised by
Reporter Clinical Scientist, HCPC registration CSxxxxx	Authoriser Consultant Clinical Scientist, HCPC registration CSxxxxx

Test information

Genes tested: BRCA1, BRCA2, ATM, CHEK2, PALB2, TP53, CDH1, STK11, PTEN, NBN

Sequencing: Data files (FASTQ) are generated by Health in Code SL (http://www.healthincode.com/). The panel is sequenced using a custom Agilent SureSelect probe capture assay on Illumina HiSeq 1500 system. All coding sequence plus splice sites (+/- 20 bp of intronic sequence) sequenced to minimum depth of 20X.

Sequence analysis, interpretation and reporting: Provided by GeneHealth UK Clinical Scientists. FASTQ files analysed using a pipeline based on Illumina BaseSpace and validated custom applications. Annotation and interpretation using Illumina BaseSpace and Alamut Visual applications. Known or suspected benign variants are not reported. Known or suspected pathogenic variants are reported. Variants of uncertain clinical significance are reported as such and are subject to expert annual review by GeneHealth UK Clinical Scientists.

Variants reported above use nomenclature recommended by the Human Genome Variation Society (http://www.hgvs.org/mutnomen/) and include the appropriate reference sequence. Nucleotide numbering is based on the A of the ATG Met start codon being designated as nucleotide 1.

Genetic Test Results & Clinical Report

GeneHealth UK

Patient information				Reporting information	
C4C number	12345P			Requester	Genetic Counsellor
Surname	Surname			Fax report to	01223 281 316
Forename	Forename			Sample date	17/11/16
Date of birth	11/04/64	Sex (F / M)	F	Report date	01/12/16

Clinical information	
Test requested	BreastGene panel (see footnote for genes tested)
Reason for request	Triple negative breast cancer at age 50, maternal family history of breast cancer

Results	
DNA sequence variants	**Heterozygous for NM_007294.3(BRCA1):c.547G>T, p.(Gly183Ter)** No pathogenic variants detected in the other genes in the BreastGene panel
DNA copy number variants	No exonic copy number variants detected in BRCA1, BRCA2 or STK11

Interpretation and comments

PATHOGENIC BRCA1 MUTATION

This individual has been found to carry a pathogenic mutation in the BRCA1 gene. This mutation is likely to be the cause of her recently diagnosed breast cancer and may also be responsible for the breast cancers seen in her maternal relatives.

This individual is at risk of further breast or ovarian cancers. Appropriate surveillance and/or risk-reducing surgery should be considered.

Confirmatory testing for this mutation can be offered for this individual's affected relatives and predictive testing can be offered for any unaffected close relatives following appropriate genetic counselling

Genotype and report by	Report authorised by
Reporter Clinical Scientist, HCPC registration CSxxxxx	Authoriser Consultant Clinical Scientist, HCPC registration CSxxxxx

Test information

Genes tested: BRCA1, BRCA2, ATM, CHEK2, PALB2, TP53, CDH1, STK11, PTEN, NBN

Sequencing: Data files (FASTQ) are generated by Health in Code SL (http://www.healthincode.com/). The panel is sequenced using a custom Agilent SureSelect probe capture assay on Illumina HiSeq 1500 system. All coding sequence plus splice sites (+/- 20 bp of intronic sequence) sequenced to minimum depth of 20X.

Sequence analysis, interpretation and reporting: Provided by GeneHealth UK Clinical Scientists. FASTQ files analysed using a pipeline based on Illumina BaseSpace and validated custom applications. Annotation and interpretation using Illumina BaseSpace and Alamut Visual applications. Known or suspected benign variants are not reported. Known or suspected pathogenic variants are reported. Variants of uncertain clinical significance are reported as such and are subject to expert annual review by GeneHealth UK Clinical Scientists.

Variants reported above use nomenclature recommended by the Human Genome Variation Society (http://www.hgvs.org/mutnomen/) and include the appropriate reference sequence. Nucleotide numbering is based on the A of the ATG Met start codon being designated as nucleotide 1.