

On the results section

You will find a summary of the findings from analysing the genes and details about the variants found during the analysis.

Result:.....
THE CLINICALLY MUTATION..... WAS IDENTIFIED IN THEGENE.
Note: "CLINICALLY SIGNIFICANT," as defined in this report, is a genetic change that is associated with the potential to alter medical intervention.
ADDITIONAL FINDINGS: No Variant(s) of Uncertain Significance (VUS) identified
VARIANT DETAILS

herediGENE 1 of 4
Genetika Medical S.A.
52, Spilaton Ave., 15344 Gerakas, Athens, Greece
www.genetika.com
Laboratory Director: George Nasiloukas PhD

SAMPLE INFORMATION

Name:	Date Received:
Father's Name:	Date of Report:
Date of Birth:	Req. Physician:
Material:	Report No.:

HerediGENE: Hereditary Cancer Panel by Next Generation Sequencing

Result:

THE CLINICALLY MUTATION WAS IDENTIFIED IN THE GENE.

Note: "CLINICALLY SIGNIFICANT," as defined in this report, is a genetic change that is associated with the potential to alter medical intervention.

ADDITIONAL FINDINGS: No Variant(s) of Uncertain Significance (VUS) identified

VARIANT DETAILS

INTERPRETATION

The BRCA2 gene has been associated with increased risk of breast and / or ovarian cancer. Studies in high risk families indicate that deleterious mutations may confer as much as 60% risk of breast cancer and 44% risk of ovarian cancer by age 70 in women (J Natl Cancer Inst;2013;105:812-822). Mutations in BRCA1 and BRCA2 have been reported to confer 83% and 62%, respectively, risk of a second breast cancer by the age of 70 (J Natl Cancer Inst;2013;105:812-822). Furthermore, mutations may also confer an increased (albeit low) risk of mal breast cancer (AJHG 62:676-689, 1998), as well as other cancers. Patient must be referred to the genetic counseling unit for adequate interpretation of the study and post-genetic support. First-degree relatives of this individual have a 50% chance of having the same mutation. Predictive testing of this mutation should be offered to all at-risk adult relatives after receiving genetic counseling.

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On the interpretation section

You will find an analysis of the findings along with risk estimations and other clinically significant information produced by the HerediGENE test.

INTERPRETATION
<p>The BRCA2 gene has been associated with increased risk of breast and / or ovarian cancer. Studies in high risk families indicate that deleterious mutations may confer as much as 60% risk of breast cancer and 44% risk of ovarian cancer by age 70 in women (J Natl Cancer Inst;2013;105:812-822). Mutations in BRCA1 and BRCA2 have been reported to confer 83% and 62%, respectively, risk of a second breast cancer by the age of 70 (J Natl Cancer Inst;2013;105:812-822). Furthermore, mutations may also confer an increased (albeit low) risk of mal breast cancer (AJHG 62:676-689, 1998), as well as other cancers. Patient must be referred to the genetic counseling unit for adequate interpretation of the study and post-genetic support. First-degree relatives of this individual have a 50% chance of having the same mutation. Predictive testing of this mutation should be offered to all at-risk adult relatives after receiving genetic counseling.</p>

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SAMPLE INFORMATION

Name:	Date Received:
Father's Name:	Date of Report:
Date of Birth:	Req. Physician:
Material:	Report No.:

HerediGENE: Hereditary Cancer Panel by Next Generation Sequencing

Result:

THE CLINICALLY MUTATION WAS IDENTIFIED IN THE GENE.

Note: "CLINICALLY SIGNIFICANT," as defined in this report, is a genetic change that is associated with the potential to alter medical intervention.

ADDITIONAL FINDINGS: No Variant(s) of Uncertain Significance (VUS) identified

VARIANT DETAILS

INTERPRETATION

The BRCA2 gene has been associated with increased risk of breast and / or ovarian cancer. Studies in high risk families indicate that deleterious mutations may confer as much as 60% risk of breast cancer and 44% risk of ovarian cancer by age 70 in women (J Natl Cancer Inst;2013;105:812-822). Mutations in BRCA1 and BRCA2 have been reported to confer 83% and 62%, respectively, risk of a second breast cancer by the age of 70 (J Natl Cancer Inst;2013;105:812-822). Furthermore, mutations may also confer an increased (albeit low) risk of mal breast cancer (AJHG 62:676-689, 1998), as well as other cancers. Patient must be referred to the genetic counseling unit for adequate interpretation of the study and post-genetic support. First-degree relatives of this individual have a 50% chance of having the same mutation. Predictive testing of this mutation should be offered to all at-risk adult relatives after receiving genetic counseling.

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SAMPLE INFORMATION

Name: _____ Date Received: _____
 Father's Name: _____ Date of Report: _____
 Date of Birth: _____ Ref. Physician: _____
 Mother's: _____ Report No.: _____

herediGENE: Hereditary Cancer Panel by Next Generation Sequencing

Result: NEGATIVE

NO CLINICALLY SIGNIFICANT MUTATIONS IDENTIFIED

Note: "CLINICALLY SIGNIFICANT" as defined in this report, is a genetic change that is associated with the detection of a clinical condition of the individual.

ADDITIONAL FINDINGS: No Variant(s) of Uncertain Significance (VUS) identified

INTERPRETATION

This result reduces the likelihood of a genetic predisposition to cancer. However, some mutations in genes of this panel may not be detected by the used methodology. Genes not included in this panel, may also contribute to increased cancer risk. Molecular genetic testing results be correlated with the clinical symptoms and family history of the patient.

DETAILS ABOUT NON-CLINICALLY SIGNIFICANT VARIANTS

All individuals carry DNA changes (i.e., variants), and most variants do not increase an individual's risk of cancer or other diseases. When identified, variants of uncertain significance (VUS) are reported. benign variants (Pharmacogenetics) are not reported and available data indicate that these variants most likely do not cause increased cancer risk. Present evidence does not suggest that non-clinically significant variant findings be used to modify patient medical management beyond what is indicated by the personal and family history and any other already significant findings.

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SAMPLE INFORMATION

Name: _____ Date Received: _____
 Father's Name: _____ Date of Report: _____
 Date of Birth: _____ Ref. Physician: _____
 Mother's: _____ Report No.: _____

herediGENE: Hereditary Cancer Panel by Next Generation Sequencing

Result: POSITIVE

ONE CLINICALLY SIGNIFICANT MUTATION WAS IDENTIFIED IN THE _____ GENE

Note: "CLINICALLY SIGNIFICANT" as defined in this report, is a genetic change that is associated with the potential to alter medical intervention.

ADDITIONAL FINDINGS: No Variant(s) of Uncertain Significance (VUS) identified

VARIANT DETAILS

INTERPRETATION

The BRCA2 gene has been associated with increased risk of breast and / or ovarian cancer. Studies in high risk families indicate that deleterious mutations may confer as much as 85% risk of breast cancer and 65% risk of ovarian cancer by age 70 in women (J Natl Cancer Inst 2013;32(8):2322). Mutations in BRCA1 and BRCA2 have been reported to confer 65% and 62%, respectively, risk of a second breast cancer by the age of 70 in High Cancer Risk (J Natl Cancer Inst 2013;32(8):2322). Furthermore, mutations may also confer an increased (other than 65% risk of most breast cancer (AJHG 2016;48: 1995), as well as other cancers.

Patients must be referred to the genetic counseling unit for adequate interpretation of the study and post-genetic support.

First degree relatives of this individual have a 50% chance of having the same mutation. Predictive testing of this mutation should be offered to all at risk individuals after receiving genetic counseling.

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SAMPLE INFORMATION

Name: _____ Date Received: _____
 Father's Name: _____ Date of Report: _____
 Date of Birth: _____ Ref. Physician: _____
 Mother's: _____ Report No.: _____

herediGENE: Hereditary Cancer Panel by Next Generation Sequencing

Result: VARIANT OF UNCERTAIN CLINICAL SIGNIFICANCE IDENTIFIED

The Variant of Uncertain Clinical Significance was identified in the _____ GENE

Note: "CLINICALLY SIGNIFICANT" as defined in this report, is a genetic change that is associated with the potential to alter medical intervention.

ADDITIONAL FINDINGS:

VARIANT DETAILS

INTERPRETATION

The BRCA2 gene has been associated with increased risk of breast and / or ovarian cancer. The clinical significance of this sequence change is uncertain at this time, but this sequence change is not expected to explain the individual's reported condition. The result does not eliminate the possibility that this individual's condition has a genetic component.

Some mutations in genes of this panel may not be detected by the used methodology. Genes not included in this panel, may also contribute to increased cancer risk.

Analysis of 1st degree relatives of this patient will help delineate the pathogenicity of this finding. Molecular genetic testing results should always be correlated with the clinical symptoms and family history of the patient.

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A **negative** report indicates that no clinically significant mutation has been identified. In this report you will find an interpretation of the findings along with details about non clinically significant variants.

A **positive** report indicates that there are pathogenic mutations identified. In this report you will find details about the variants detected and extensive interpretation of the risk of the individual, along with analysis of the findings.

A **VUS (Variant of Unknown Significance)** report indicates that there are mutations found for this individual that have not been yet characterized as pathogenic or as benign. An even more extensive *in silico* bioinformatic analysis is performed in these cases.

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Name: _____ Report No.: _____

PATIENT'S FAMILY TREE

Note: The information shown on the family tree has been provided by the patient and not by medical records.

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Assuming that family history is provided, a comprehensive family tree is included in the HerediGENE report.