Understanding the Report
HerediGENE

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 THE .....GENE.

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

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

VARIANT DETAILS


1 of 4

Genekor Medical S.A.
52, Spaton Ave., 15344, Gerakas, Athens, Greece
www.genekor.com

Laboratory Director: George Nasioulas 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

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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 (albeir low) risk of mail breast cancer (AJHG 62:676-689, 1998), as well as other cancers.
Patient must be referred to the genetic counceling 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 counceling.

GENEKOR | 52 Spaton Ave, Gerakas, 15344 Athens | info@genekor.com www.genekor.com | Tel. (+30) 210 6032138 Fax. (+30) 210 6032148
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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.

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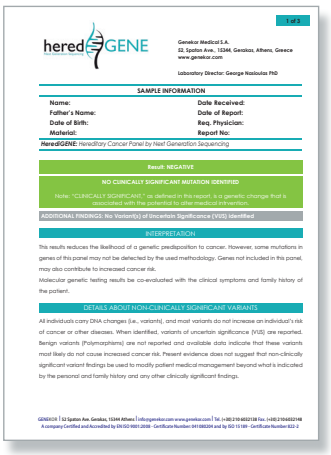
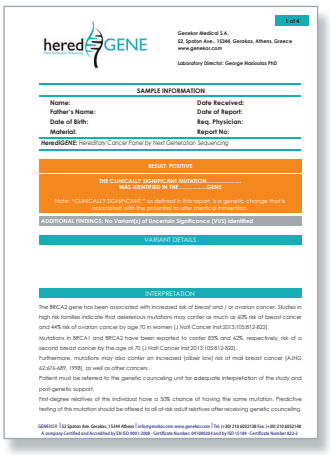
HerediGENE

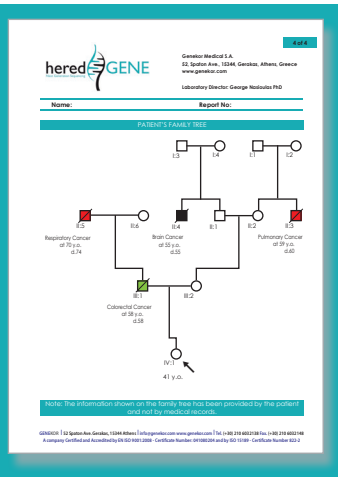




A **negative** report indicates that no clinically significant mutation has been identified. In this report you will find an interpretation of the findings along with details about non clinically significant variants.

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.



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