CellMax

PINK Breast Cancer Genetic Risk Test

Test Report

CellMax Life

Test Report

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CONTENTS

Test Report

CellMax PINK Breast Cancer Genetic Risk Test

1-1. Patient Information

Requisition Number	
Patient Name	
ID Number	
Date of Birth	
Gender	☐ M ☐ F
Patient Phone Number	
Patient E-mail	
Name of Physician	
Physician Phone Number	
Date of Collection	
Date of Report	

Test Report

CellMax PINK Breast Cancer Genetic Risk Test

1-2. Test Report Summary

Summary Result: Negative

No Clinically Significant Genetic Mutations Detected

Gene	Mutation	Interpretation

The classification and interpretation of all variants identified in the test reflects the current state of scientific and medical understanding at the time the report is generated. The five variant classification categories include pathogenic, likely pathogenic, variant of uncertain significance (VUS), likely benign, and benign. CellMax Life only reports pathogenic variants, which have strong lines of evidence associated with increased cancer risk. A summary of all variants found can be provided for each patient upon request by their physician for an additional charge.

A positive test result for a pathogenic mutation in a gene means that your lifetime risk(s) of developing the associated cancer(s) is significantly higher than an individual who does not have a mutation. **It does not mean that you have cancer or that you will eventually develop cancer in your lifetime.** Likewise, a negative result **does not mean that you do not have cancer, or that you will not develop cancer at some point in your lifetime.**

Comments

Electronic Signature

Lab Supervisor
Amita Gupta, CGMBS MTP01001934

Date

CellMax Life

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Test Report

CellMax PINK Breast Cancer Genetic Risk Test

1-4. List of Genes Tested

Gene Symbol	Gene Name	Pathogenic Gene Mutation Detected
ATM	*ataxia telangiectasia mutated*	No
BARD1	*BRCA1 Associated RING Domain 1*	No
BRCA1	*breast cancer 1, early onset*	No
BRCA2	*breast cancer 2, early onset*	No
BRIP1	*BRCA1 interacting protein C-terminal helicase 1*	No
CDH1	*cadherin 1, type 1, E-cadherin (epithelial)*	No
CHEK2	*checkpoint kinase 2*	No
NBN	*nibrin*	No
NF1	*neurofibromin 1*	No
PALB2	*partner and localizer of BRCA2*	No
PPM1D	*protein phosphatase, Mg2+/Mn2+dependent 1D*	No
PTEN	*phosphatase and tensin homolog*	No
RAD51C	*RAD51 homolog C (S. cerevisiae)*	No
STK11	*serine/threonine kinase 11*	No
TP53	*tumor protein p53*	No

Test Report

CellMax PINK Breast Cancer Genetic Risk Test

2-1. About the Test

CellMax Life has developed a next-generation sequencing-based test for identifying hereditary cancer susceptibility mutations. The test uses advanced next-generation sequencing (SMSEQ™) targeting 15 genes with a high-degree of analytical sensitivity and specificity. Validation using industry standard methods yielded an accuracy of >99.99%. The genetic panel was curated by a team of genetic specialists to include 15 genes reported in the literature as being associated with increased risk for breast cancer and possibly other cancer/tumor types.

The assay performed was developed and its performance characteristics determined by CellMax Life, a clinical laboratory certified under Clinical Laboratory Improvement Amendments (CLIA #05D2119032) to perform high-complexity testing. It has not been cleared or approved by the FDA. This test is used for clinical purposes, and should not be regarded as investigational or for research.

Details about Mutations and Variants

Genetic Variants

All persons carry genetic variants inherited from their parents. A variant can be used to describe a change in a DNA sequence that may be pathogenic, likely pathogenic, unknown significance, likely benign, or benign. Most variants do not cause an increase in the risk of cancer or other disease. The classification and interpretation of all variants identified in the test reflects the current state of scientific and medical understanding at the time the report is generated. Variants are classified by pathogenicity by taking into account the reported variant, and the allelic frequencies from population studies and clinical databases (e.g. 1000 Genomes, ClinVar). A positive test result indicates that an individual has inherited a pathogenic mutation in specific genes and, therefore, has an increased risk of developing certain cancers. It is important to understand that a positive test result does not necessarily mean that the individual will actually develop cancer over their lifetime. Some individuals who inherit pathogenic mutations will never develop the associated cancer(s). A negative test result indicates that an individual has not inherited a pathogenic mutation in any of the genes tested, but does not eliminate the lifetime risk of developing certain cancers.

Pathogenic Variants

Certain mutations in certain genes are associated with an increased risk for cancers and/or hereditary syndromes. These mutations are associated with the potential to alter medical intervention. A pathogenic variant directly contributes to the development of cancer. The variant has strong lines of evidence that associates it with significantly increased cancer risk and necessary clinical action.

Likely Pathogenic Variants

A likely pathogenic variant is very likely to contribute to the development of cancer. The variant has fewer strong lines of evidence that associates it with significantly increased cancer risk.

Uncertain Significance Variants (VUS)

A variant of uncertain significance does not have enough information at this time to support a more definitive classification. There is insufficient evidence to determine if the variant is associated with increased cancer risk.

Likely Benign

A likely benign variant is not expected to have a major effect on cancer. However, additional evidence is needed to confirm this assertion.

Benign

A benign variant has strong lines of evidence that does not associate it with an increased cancer risk.

Test Report

CellMax PINK Breast Cancer Genetic Risk Test

Genes Tested

ATM	*BARD1*	*BRCA1*	*BRCA2*	*BRIP1*	*CDH1*	*CHEK2*	*NBN*	*NF1*
PALB2	*PPM1D*	*PTEN*	*RAD51C*	*STK11*	*TP53*			

Test Limitations

Inherited mutations in certain genes are associated with hereditary cancer syndromes or increased risk to various cancer types. The test interrogates and reports single nucleotide variants, insertions, and deletions in genomic DNA. Large scale genomic rearrangements, copy number variants, as well as structural changes are not detected. CellMax Life only reports pathogenic variants, which have strong lines of evidence associated with increased cancer risk.

A negative test result means that the laboratory did not identify a variant that is of pathogenic significance in any of the genes under consideration. This result can indicate that a person is not a carrier of a specific genetic mutation, or does not have an increased risk of developing a certain disease. It is possible, however, that a disease-causing genetic alteration was missed because many tests cannot detect all genetic changes that can cause a particular disorder. Even with genetic sequencing, mutation detection is not 100% sensitive, since sequencing will not detect large genomic rearrangements and large indels. A negative test result, therefore, does not completely rule out the possibility that the patient is a mutation carrier. Further testing may be required to confirm a negative result.

2-2. References

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Test Report

CellMax PINK Breast Cancer Genetic Risk Test

Disclaimer

DNA studies do not constitute a definitive test for any disease conditions in an individual. This test was developed and its performance characteristics determined by CellMax Life. Clinical decisions regarding care and treatment of customers should not be solely based on this test. How this information is used to guide customer care is the responsibility of the physician. You may wish to speak with a trained genetics professional to discuss the results and subsequent options from this test.

The CellMax Life test is designed to assist health care practitioners in providing additional clinical information. The information therein should not be relied upon as being complete or accurate, nor should it be considered as inclusive of all proper treatments or methods of care or as a statement of the standard of care. Medical knowledge develops rapidly and new evidence may emerge between the time information is developed to when it is published or read.

Genetics is about probabilities, not certainties. It is important to realize that many variables impact our genes so the outcome is not certain even if we have a known genetic risk factor. An unfavorable mutation means you are much more likely to be affected by that gene than others who do not carry the same mutation. But the tremendous benefit of genetic testing is that you can influence the gene by the choices you make in your lifestyle, diet, nutrition, supplements, exercise and even your outlook. Genes can be turned on and off by our lifestyle choices and if you have an unfavorable mutation you can still influence a lot of control over its expression. The absence of a pathogenic mutation does not eliminate an individual's risk of developing cancer, as cancer can be caused by, but not limited to, both inherited and acquired genetic mutations. Sources of acquired genetic mutations include various factors such as aging, environment, and lifestyle choices. The test is not recommended for new bone marrow transplant recipients.

The information herein is not continually updated and may not reflect the most recent evidence. The information addresses only the topics specifically identified therein and is not applicable to other interventions, diseases, or stages of diseases. This information does not mandate any particular course of medical care. Further, the information is not intended to substitute for the independent professional judgment of the treating physician, as the information does not account for individual variation among customers. CellMax Life provides this information on an "as is" basis, and makes no warranty, express or implied, regarding the information. CellMax Life specifically disclaims any warranties of merchantability or fitness for a particular use or purpose. CellMax Life assumes no responsibility for any injury or damage to persons or property arising out of or related to any use of this information or for any errors or omissions.

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