Test It

BRCA1 & BRCA2

Hereditary Breast & Ovarian Cancer Syndrome

* Comprehensive genetic testing includes:

- ☑ Full Sequencing of BRCA1/BRCA2
- ☑ Detection of large genomic rearrangements

*NCCN: National Comprehensive Cancer Network (http://www.nccn.org)

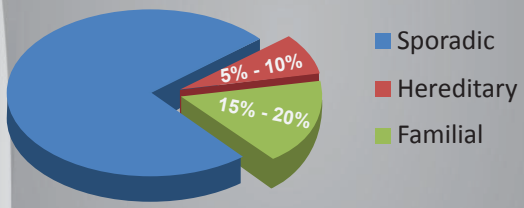
Hereditary Cancer Testing: Is it Right for You?

This manual is designed to assist you to decide whether testing for Hereditary Breast and Ovarian Cancer Syndrome is right for you. Your decision should be discussed and reviewed with a healthcare provider.

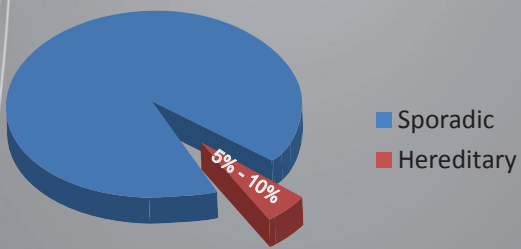
Introduction

Most cancer in general and breast cancer specifically is sporadic. This means that these cancers are not transmitted from generation to generation.

Breast Cancer



Ovarian Cancer



Where does the Hereditary Breast and Ovarian Cancer Syndrome originate from?

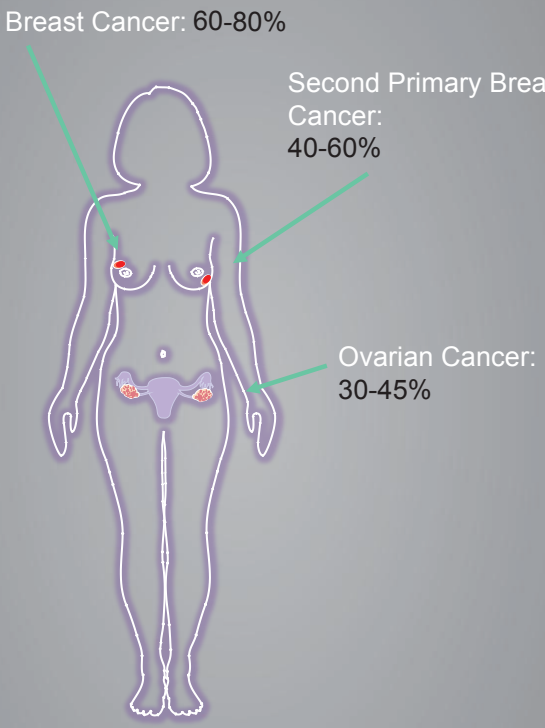
Recent studies have shown that alterations in the *BRCA1* and *BRCA2* genes are responsible for the majority of hereditary breast and ovarian cancer. Such alterations increase the predisposition for breast and / or ovarian cancer and other cancers as well. A smaller percentage of hereditary breast and ovarian cancer cases is due to alterations in other genes such as *PALB2* and *CHECK2*

It is a Family Issue…

- ✔ Mutations on BRCA1 & BRCA2 genes can be passed down from generation to generation.
  - ✔ If you are a mutation carrier then your parents, children, or siblings have a 50% chance of carrying that same mutation.
  - ✔ Other relatives such as cousins, aunts, or uncles can have that same mutation as well.
- ✔ It is important to share your results with your family members.

What is the Risk for BRCA Mutation Carriers?

BRCA1-Related Cancers: Life-time Risk



Increased risk for other types of cancer:

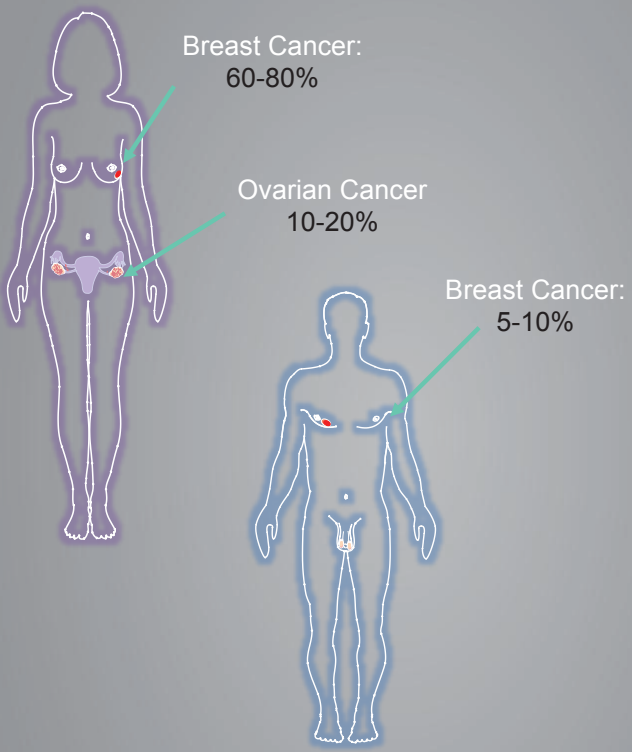
Colon: 10-15%

Male Breast Cancer: 1-5%

Pancreas: 2-3%

What is the Risk for BRCA Mutation Carriers?

BRCA2-Related Cancers:
Life-time Risk



Increased risk of developing cancer:

Prostate: 15-25%

Pancreas: 3-5%

Melanoma: 3-5%

Do you have a Personal or Family History?

- ❑ Breast cancer before age 50. *(Ref. ASBS, NCCN)*
- ❑ "Triple negative" (ER-, PR-, Her2 normal) breast cancer diagnosed prior to age 60. *(Ref. ASBS, ESMO, NCCN)*
- ❑ Two Primary breast cancers, either bilateral or ipsilateral. *(Ref. ASBS, ESMO, NCCN)*
- ❑ Breast and history of ovarian cancer in the same patient at any age *(Ref. NCI, ESMO)*
- ❑ A first degree relative with bilateral breast cancer or breast and ovarian cancer. *(Ref. NCI)*
- ❑ Two first-degree relatives who've had breast cancer, one of whom diagnosed < 50. *(Ref. NCI)*
- ❑ A combination of first and second degree relatives diagnosed with breast and ovarian cancer (one cancer type per patient). *(Ref. NCI)*
- ❑ Pancreatic cancer associated with a family history of breast and ovarian cancer. *(Ref. ASBS, NCCN)*
- ❑ A male relative who's had breast cancer. *(Ref. ASBS, NCCN)*
- ❑ Identified BRCA mutation. *(Ref. NCI)*
- ❑ Other:_____

ASBS: American Society of Breast Surgeons (Ref. Position Statement on BRCA Genetic Testing for Patients With and Without Breast Cancer)
NCI: National Cancer Institute (Ref. http://www.cancer.gov/cancertopics/factsheet/Risk/BRCA)
ESMO: European Society of Medical Oncology. (Ref. Annals of Oncology 22 (Supplement 6): vi12–vi24, 2011)
NCCN: National Comprehensive Cancer Network (http://www.nccn.org)

Full analysis of BRCA1 & BRCA2 genes: Full sequencing by NGS and detection of large genomic rearrangements by MLPA Possible results:

Positive Result

- **Increased Risk for developing breast and/or ovarian cancer.** Knowing your individual risk will assist you and your physician to plan accordingly for your personal risk reduction strategy.

Negative Result - From having a Full Sequence Analysis

- **Reduced chance of Hereditary Breast and Ovarian Cancer:** This result shows that you do not have an elevated risk of breast and/or ovarian cancer from the BRCA1 & BRCA2 genes but does not exclude the small likelihood of having hereditary breast cancer due to other genes.

Negative Result - When a mutation has been detected in another family member

- **Risk same as general population:** This result shows that you do not have an elevated risk for ovarian and/or breast cancer.

Unclassified Variant

- **The risk of cancer has not been fully defined based on international bibliography:** It is uncertain in its significance of developing breast and ovarian cancer.

Benefits and Limitations.

Benefits	Limitations
• Individualized assessment of hereditary cancer risk • Information to be used in the medical management of the patient and family planning • Important information for family members • Decrease in stress and anxiety	• The analysis does not detect all causes for hereditary cancer

NGS: Next Generation Sequencing
MLPA: Multiplex Ligation-dependent Probe Amplification

GeneKor S.A. operates molecular biology laboratories with state of the art equipment suitable to provide the highest quality of Molecular Services. We provide a diversification of services focused on improving patient care utilizing the latest technologies of prognostic, predictive, and personalized medicine.

The Laboratories of GeneKor are certified by the European Molecular Quality Network (EMQN) and the College of American Pathologists (CAP) for the BRCA1 and BRCA2 analysis.

Sample requirements: Peripheral Blood in EDTA.

Methodology: BRCA1 & BRCA2 full sequencing by NGS and detection of large genomic rearrangements by MLPA.

Results delivery: 10-15 working days from the time the sample arrives to GeneKor.

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