



Understanding
Your Genetic Test Result

**Positive for a
Deleterious Mutation or
Suspected Deleterious**

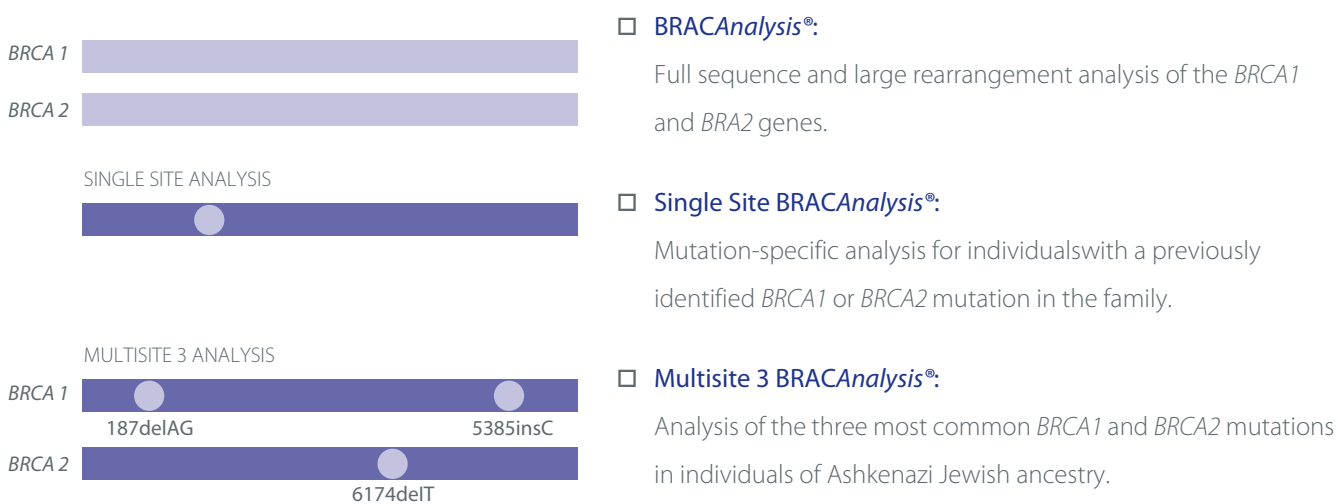


POSITIVE DELETERIOUS MUTATION

This workbook is designed to help you understand the results of your genetic test and is best reviewed with your healthcare provider. Please verify that your test result matches the following information by looking at the patient copy of your test result or contacting the healthcare provider who ordered your test. If your test result does not match, please disregard this brochure, and contact your healthcare provider.

Your Genetic Test Result (check the appropriate boxes below)

THE GENETIC TEST YOU RECEIVED



YOUR TEST RESULT

- ☐ Positive for a Deleterious Mutation on
- ☐ Genetic Variant, Suspected Deleterious

Overview of Your Test Result

Mutations in *BRCA1* and *BRCA2* cause most cases of hereditary breast and ovarian cancer (HBOC), a syndrome that involves various cancers, primarily breast and ovarian.

- You have a mutation or alteration in either the *BRCA1* or *BRCA2* gene.
- You have HBOC syndrome.
- HBOC syndrome increases the risk of various cancers, primarily breast and ovarian.
- Not everyone with HBOC syndrome will develop cancer.

Your Cancer Risks

Having a *BRCA1* or *BRCA2* mutation increases the risk of certain cancers. If you have already had a cancer diagnosis, you have an increased risk for developing another cancer.

If you have NOT had breast or ovarian cancer	Mutation Carrier	General Population
Breast cancer by age 50	33-50%	2%
Breast cancer by age 70	56-87%	7%
Ovarian cancer by age 70	27-44%	<2%
Male breast cancer by age 70	7-8%	.05%

If you HAVE had breast cancer	Mutation Carrier	General Population
Ovarian cancer	15%	not available
Breast cancer after 5 years	27%	3.5%
Breast cancer by age 70	64%	11%

Other cancer risks*	Mutation Carrier	General Population
Prostate cancer by age 80	20%	15%
Pancreatic cancer by age 80	Up to 7%	<1%

* Less information is available about the risks of these and other cancers than about breast and ovarian.

Notes/Questions

Managing Your Risks

Options for reducing cancer risk are available whether or not you have already had a cancer diagnosis. The following are medical management guidelines for *BRCA1* and *BRCA2* mutation positive individuals. Discuss these options with the appropriate medical professionals to determine how you will manage your cancer risks.

INCREASED SURVEILLANCE

- Monthly breast self-exams starting at the age of 18 and clinical breast exams two times a year beginning at age 25.
- Yearly screening with both mammography and Mri beginning at the age of 25.
- Pelvic exam twice a year beginning at age 35 in patients not electing surgery to reduce ovarian cancer risk.
- Transvaginal ultrasound (an imaging technique used to create a picture of the genital tract in women by using a hand held device inserted directly into the vagina) and testing for CA-125 levels in the blood two times a year if surgery to reduce ovarian cancer risk is not done.

RISK REDUCING DRUG THERAPY

- Drugs such as tamoxifen have been shown to reduce the risk of breast cancer in high risk women.
- Birth control pills may reduce the risk of ovarian cancer in women with *BRCA1* or *BRCA2* gene mutations.

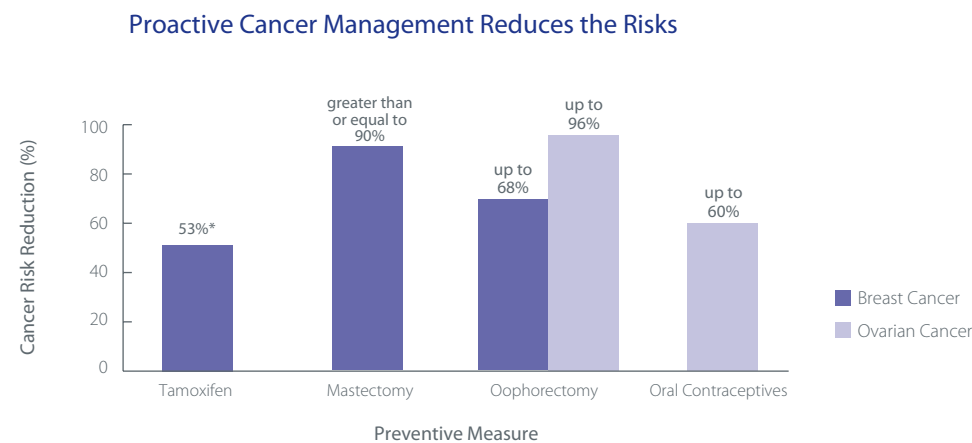
PREVENTIVE SURGERY

- Preventive mastectomy (removal of the breasts) significantly reduces the risk of breast cancer in women with *BRCA1* or *BRCA2* gene mutations.
- Preventive removal of the ovaries and fallopian tubes (Bilateral Salpingo Oophorectomy) significantly reduces the risk of ovarian cancer, and also breast cancer, in women with *BRCA1* or *BRCA2* gene mutations.

FOR MEN

- Monthly breast self-exams and clinical breast exams two times a year beginning at age 35.
- Consider baseline mammogram at age 40.
- Adhere to population screening guidelines for prostate cancer.

Note: Some families also have an increased incidence of pancreatic tumors and melanoma. Consider full body skin exam for melanoma and investigational protocols for pancreatic cancer.



It's a Family Affair

BRCA1 and *BRCA2* mutations are passed on in a family. Now that a mutation has been identified in you:

- Your close blood relatives (parents, children, brothers, and sisters) have a 50% chance of having the same mutation.
- More distant relatives (cousins, uncles, and aunts) also have a chance of having the mutation that runs in your family.
- Generally, the mutation is only going to be found on the side of your family (father's or mother's) that has the history of breast and ovarian cancer.

Your relatives can be offered Single Site BRCA*Analysis*^{®*} to determine whether or not they have the same mutation.

- Relatives interested in genetic testing will need to know your specific mutation. It is best to provide your relatives with a copy of your test result which you can obtain from your healthcare provider.
- Single Site BRACAnalysis® costs much less than a comprehensive test.
- If your relative is:
 - **Positive** for the mutation, he/she has the increased cancer risks associated with *BRCA1* and *BRCA2* mutations and can benefit from appropriate medical management.
 - **Negative** for the mutation, he/she has an average risk of cancer and can follow general population screening guidelines.

Your healthcare provider can assist in determining which of your relatives should consider genetic testing.

* If you had a Multisite3 testing, your relatives may need Multisite3 testing instead of Single Site analysis; talk to your doctor about this possibility.

Myriad has resources available to help you with your genetic test result.

- Contact Myriad's Medical Services Department at Tel. 0041 (0)44 939 91 30 or e-mail helpmed-international@myriad.com for:
 - Answers to questions about your test result.
 - Information about genetic testing for your relatives.
 - A sample letter that can be sent to relatives who may need genetic testing.

If you need a copy of your genetic test result, please contact the healthcare provider who ordered your test.

Notes/Questions

Next Steps

Please work with your healthcare provider to determine the most appropriate next steps for you.

- ☐ Obtain a copy of your test result.
- ☐ Schedule consultations with appropriate healthcare providers (list below).

- ☐ Create a plan for medical management
 - Increased surveillance: -----
 - Chemoprevention: -----
 - Preventive surgery: -----
- ☐ Share your genetic test result with your relatives (identify your specific mutation so relatives can have Single Site testing).
- ☐ Re-contact your healthcare provider on a regular basis for new information about HBOC.

Notes/Questions

Additional Resources

[Myriad Genetics GmbH](#)

E-mail Medical Services with questions about testing:
helpmed-international@myriad.com
www.myriad.com
Tel. 0041 (0) 44 939 91 30

[Europa Donna – The European Breast Cancer Coalition](#)

Is an independent non-profit organization whose members are affiliated groups from countries throughout Europe.
Piazza Amendola 3
20149 Milan – Italy
E-mail: info@europadonna.org
www.europadonna.org
Tel. 0039 02 3659 22 80

[Young Survival Coalition](#)

Serves as a point of contact for women 40 and under with breast cancer. This organization seeks to educate the key communities (medical, research, and legislative) about breast cancerin young women.
www.youngsurvival.org

BRACAnalysis®

A test for Hereditary Breast
and Ovarian Cancer (HBOC) syndrome

A predictive medicine product for hereditary breast and ovarian cancer.

BRACAnalysis® testing assesses a woman's risk of developing hereditary breast or ovarian cancer based on detection of mutations in the *BRCA1* and *BRCA2* genes. This test has become the standard of care in identification of individuals with hereditary breast and ovarian cancer.

This information is provided to help answer some of your questions with respect to cancer risks, hereditary cancer risks and hereditary cancer testing. It is general in nature and is not intended to provide a definitive analysis of your specific risk factors for cancer or your hereditary cancer risks. You should not rely on the information provided herein; but rather, you should consult with your doctor or a qualified healthcare professional to review this information along with your individual health conditions and risk factors.



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