Hereditary Breast and Ovarian Cancer Testing and Genetic Counseling Services



BRCAssuresm







Because Knowledge is a Powerful Tool.

What is BRCA?

The *BRCA1* and *BRCA2* (breast cancer 1 and 2) genes are two genes found within everyone's normal genetic material. When mutations, or gene alterations, are found within these genes, the mutations can cause an increased risk for several specific types of cancer, including breast cancer and ovarian cancer.^{1,2} The *BRCA* mutations can be passed down through a family and are associated with hereditary breast cancer and ovarian cancer.

More than 200,000 individuals are diagnosed with breast cancer each year.³ Having a *BRCA* mutation significantly increases your risk of developing breast, ovarian, or other specific types of cancers (see table).⁴

Genetic testing for *BRCA1* and *BRCA2* mutations is available for those individuals at increased

BRCA mutations account for 5 -10% of all breast cancers.⁵

risk and can help provide information about your risk for developing breast, ovarian, or other cancers. This brochure will provide you with general information on *BRCA* testing and inheritance in order to help you understand if *BRCA* testing may be right for you. It is always important to speak to your physician or a genetics professional for additional information or to answer any other questions you may have.

Who is at risk of having a BRCA mutation?

BRCA mutations can occur in any ethnic or racial group and in both men and women. The estimated frequency of a *BRCA* mutation occurring in the general population is 1 in 400. There are specific ethnic groups that are at higher risk for having a *BRCA* mutation. The Ashkenazi Jewish population has a high risk of carrying a *BRCA* mutation with a frequency of 1 in 40.⁴

Risk of Cancer in Individuals with a <i>BRCA1</i> or <i>BRCA2</i> Mutation ⁴			
Cancer Type	General Population (no mutation)	Individuals with Mutation	
		BRCA1	BRCA2
Breast	12%	50 - 80%	40 - 70%
Ovarian	1 - 2%	24 - 40%	11 - 18%
Male Breast	0.1%	1 – 2%	5 - 10%
Prostate	15% (N. European Origin) 18% (African Americans)	up to 30%	up to 39%
Pancreatic	0.5%	1 - 3%	2 - 7%

Who should consider BRCA testing?

Reviewing your family history is important when considering if *BRCA* testing is right for you. The American College of Obstetricians and Gynecologists⁶ and The National Comprehensive Cancer Network⁷ both support testing in individuals with a high risk or strong family history of breast or ovarian cancer. Testing may be more appropriate for those with a personal or family history of:⁴

- Breast cancer at a young age (<50 years)
- Ovarian cancer
- Multiple primary breast cancers either in the same or opposite breast
- Both breast and ovarian cancer
- Triple-negative breast cancer (estrogen receptor negative, progesterone receptor negative, & HER2/neu negative)
- Male breast cancer
- Ashkenazi Jewish heritage
- Pancreatic cancer with breast or ovarian cancer in the same individual or the same side of the family
- Two or more relatives with breast cancer, with one diagnosed at age 50 or younger
- Three or more relatives with breast cancer at any age
- A previously identified *BRCA1* or *BRCA2* mutation in the family

In general, predictive genetic testing for hereditary breast and ovarian cancer is not recommended for at-risk individuals younger than 18 years of age.^{4,7}

How is a BRCA mutation inherited?

BRCA1 and *BRCA2* mutations can be passed down, or inherited, from either your mother or father. *BRCA* mutations are inherited in an autosomal dominant fashion, which means that having just one copy of an altered *BRCA1* or *BRCA2* gene can increase your chance of developing certain cancers; you don't need to inherit a mutation from both your mother and your father.⁸ When either your mother or father carries a *BRCA* mutation, there is a 50% chance you will also have the same mutation.⁵ It is important to remember that not everyone who inherits a *BRCA* mutation will develop cancer; individuals carrying a *BRCA* mutation only have an increased risk of developing cancer.

What is the purpose of BRCA testing?

The purpose of *BRCA* testing is to see if you are carrying a *BRCA* mutation and could therefore be at increased risk for developing certain cancers, including breast or ovarian cancer. Having this information can allow your physician or genetics professional to provide the appropriate additional screening and/or prevention options that could help reduce your risk of developing cancer. The BRCA*ssure* test requires a sample of blood and filling out a family history questionnaire. Results are usually ready within 2-3 weeks.

What do the results mean?

BRCA genetic testing could give you a few possible results: positive, negative or an uncertain result. In all cases, you should discuss the results and any appropriate follow-up with your physician.

Positive result

A positive test result means that you have inherited a *BRCA* mutation and have an increased risk of developing breast or ovarian cancer. This test result does not indicate if you will actually develop cancer.

A positive test result could also have important implications for your family members. You would have inherited this mutation from your mother or your father, and therefore your relatives could also be at risk for carrying a *BRCA* mutation. There will also be a chance that you will pass this mutation on to your children. It can be helpful to discuss this test result with your family members so they too can decide if genetic testing is right for them.

Negative result

A negative test result means that your chance of carrying a *BRCA* mutation has been greatly reduced, but not completely eliminated. This does not mean that you cannot develop breast

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What do the results mean? (cont.)

cancer. Most people diagnosed with breast cancer do not carry a BRCA mutation.⁵ Individuals with negative results will still require routine screening for breast and other cancers.

Uncertain result

An uncertain result means that an alteration in the BRCA1 or BRCA2 gene has been detected but, at this time, it is not clear if the specific gene alteration will lead to cancer or not. Genetic information is evolving and expanding every day and researchers are constantly learning more about the gene alterations seen on the BRCA1 and BRCA2 genes and their associated cancer risks. Individuals with uncertain results should discuss appropriate surveillance and follow up with their physicians.

What options do I have if I carry a BRCA mutation?

If you are positive for a BRCA mutation, there are several options that you and your physician could consider to reduce the risk for developing cancer. Your options may include:⁴

- Increased or enhanced screening for cancer
- Proactive (prophylactic) surgery
- Drug therapy (chemoprevention)
- Evaluation of your family members' risk

It is important to discuss these options with your doctor and/or your genetic counselor to understand which option is best for you.

References

- 1. BRCA1. Genetics Home Reference. Available at: http://ghr.nlm.nih.gov.gene/BRCA1. Accessed October 22, 2013.
- 2. BRCA2. Genetics Home Reference. Available at: http://ghr.nlm.nih.gov.gene/BRCA2. Accessed October 22, 2013.
- 3. Breast Cancer. National Cancer Institute. Available at: http://www.cancer.gov/ cancertopics/types/breast. Accessed October 29, 2013. 4. Petrucelli, N, et al. BRCA1 and BRCA2 Hereditary Breast and Ovarian Cancer. Gene
- Reviews. Available at: http://www.ncbi.nlm.nih.gov/books/NBK1247. Accessed October 22, 2013. 5. Hereditary Breast and Ovarian Cancer. Cancer.net. Available at: http://www.cancer.
- net/cancer-types/hereditary-breast-and-ovarian-cancer. Accessed October 31, 2013. 6. Hereditary Breast and Ovarian Cancer Syndrome. ACOG Practice Bulletin, Number 103, April 2009; reaffirmed 2013.

April 2009; featimmed 2013.
Hereditary Breast and/or Ovarian Cancer Syndrome. NCCN Guidelines Version
4.2013. Available at: http://www.nccn.org/professionals/physician_gls/pdf/genetics_
screening.pdf. Accessed October 30, 2013.
Breast Cancer. Genetics Home Reference. Available at: http://ghr.nlm.nih.gov/

condition/breast-cancer. Accessed October 22, 2013.

Genetic Counseling for BRCAssureSM

Your doctor may refer you to a genetic counselor to discuss your risk for hereditary breast and ovarian cancer and your testing options. A genetic counselor is a healthcare professional with a master's degree in human genetics or genetic counseling. The role of a genetic counselor is to help you better understand your genetic risks and the tests available to you. Their training enables genetic counselors to discuss technical genetic information in practical, useful terms. Integrated Genetics genetic counselors are all board certified by the American Board of Genetic Counseling or are active candidates for certification.

What happens during a breast and ovarian cancer genetic counseling session?

The genetic counselor will lead you through a discussion to evaluate your medical history for relevant information, review your family history, and construct a family tree. Throughout the discussion the genetic counselor will use this information in order to:

- Evaluate your risk for breast and ovarian cancer and your risk to be a carrier for the BRCA1/2 genes.
- Discuss the genetic testing options which may be appropriate for you.
- Explain the potential results of BRCA1/2 testing and what those results may mean in the context of your family history.
- Provide an overview of cancer screening options and risk reduction measures.

The decision to accept or decline any recommended genetic testing is up to you. Additional written educational materials and/or information about community resources are available for you upon request.

How do I schedule an appointment with an Integrated Genetics genetic counselor?

If your physician recommends genetic counseling with Integrated Genetics, you can call 855-GC-CALLS or 855-422-2557 to obtain additional information and to schedule an appointment.

www.mytestingoptions.com

About Integrated Genetics and Integrated Oncology

Our team of experts has been a leader in genetic testing and counseling services for over 25 years.

This brochure is provided by Integrated Genetics and Integrated Oncology as an educational service for physicians and their patients.

For more information on our genetic testing and counseling services, please visit our web sites:

www.mytestingoptions.com www.integratedgenetics.com www.integratedoncology.com







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Integrated Genetics Genetic Counseling 855-GC-CALLS or 855-422-2557