



BRCA Mutations

Learning that you or a loved one carries a mutation in the BRCA1 or BRCA2 gene can be upsetting and even shocking. You may feel overwhelmed by the amount of information you now need to consider and how it may affect your life. But knowing about this mutation can help you make better medical decisions. This resource will explain what BRCA gene mutations are and how they might affect you and your family.

GENES AND MUTATIONS

Genes carry the information that pass on traits that can be inherited from parents to children. Chemically, genes are made of DNA. They affect the way our cells and bodies work.

Sometimes we experience changes in our genes. These changes are called mutations. Some mutations develop over time and are acquired during your lifetime. Other mutations can be inherited from your parents and passed on to children. Gene mutations can make people more prone to develop certain diseases, like cancer.

WHAT DOES IT MEAN TO HAVE A MUTATION IN THE BRCA GENES?

The most well-known genetic mutation that can lead to cancer occurs in the BRCA genes. Everyone is born with BRCA genes (BRCA1/ BRCA2). BRCA genes make proteins that help repair damaged DNA. If you test positive in a BRCA genetic test (are BRCA+), that means there is a mutation in your gene. A mutation in one of the BRCA genes may cause the proteins they make to stop working. Without these proteins, harmful errors in a cell's DNA can build up. This can cause the cell to grow and divide without control, leading to cancer.

BRCA gene mutations can increase your risk of developing certain cancers, like breast or ovarian cancer. In the United States, people assigned female at birth typically have a 13% chance of developing breast cancer at some point in their life. With a BRCA1 gene mutation the chances of developing this cancer increase to about 55 to 72 percent. With a BRCA2 gene mutation the chances increase to roughly 45 to 69 percent. The risk of ovarian cancer in the United States,

is about 2%. With a mutation in the BRCA1 gene, a person's risk of ovarian cancer increases to about 39 to 46 percent. For people with a BRCA2 mutation the risk of ovarian cancer increases to about 10 to 27 percent. Research shows that mutations in the BRCA2 gene may also increase a person's risk for developing other cancers. These can include melanoma, prostate, colon, and some pancreatic cancers.

Not every person who has a BRCA1 or BRCA2 mutation will develop cancer. Knowing you have the mutation does not tell you everything about your risk either. It cannot tell you about your exact risk percentage, at what age you may develop cancer, or even which cancer you may develop. Because the mutation can cause your risk to increase drastically, it is important to monitor your health with your doctor during routine appointments.

WAYS TO REDUCE CANCER RISK

If you or a loved one carries a mutation in a BRCA gene, there are a number of options to help manage your cancer risk. These include:

- Getting routine screenings, like your annual mammogram
- Asking your doctor about additional screenings, like breast MRIs or regular endoscopies of the upper abdominal tract
- Asking your doctor about preventative medications you can take
- Talking with your doctor about preventive breast surgery options



KNOW YOUR HISTORY AND GET TESTED: GENETIC TESTING AND BIOMARKER TESTING

If you or a loved one has been diagnosed with cancer, talk with your family. Sharing your cancer diagnosis with your family can help everyone prepare. Everyone can take action to better their health. Ask your doctor about testing if you haven't done so already. Having this information not only impacts your own cancer risk, but that of your family members too.

Two kinds of tests can provide you with more information:

Genetic testing looks for mutations or changes in your genes. Testing to see if you inherited a mutation that increases your risk of getting cancer when you have no diagnosis of cancer can be called “genetic testing for inherited cancer risk.” If you have been diagnosed with cancer, doctors may recommend you see a genetic counselor to test your genes for an inherited mutation. The genetic counselor will ask detailed questions about who in your family has had cancer and at what age.





The test typically involves a blood or saliva sample. It usually takes a couple of weeks to get the results.

Breast and ovarian cancer can run in families through the BRCA genes. If you are diagnosed with any form of cancer, be sure to ask for genetic testing. This is especially important if you have a family history of breast or ovarian cancer. If you test positive for BRCA or even another cancer gene, talk with the genetic counselor about the next steps – for you and for your family members.

Biomarker testing looks for biomarkers in your cancer sample. A biomarker is a sign of disease or abnormal function that can be measured in your blood, tissue, or bodily fluid. In cancer, biomarkers are often used to help choose the best treatment for you. These biomarkers can be proteins, genes, or gene mutations. Biomarkers can tell your doctor about the subtype of the cancer as well as if there is a chance that the cancer may return (cancer recurrence).

Even if your genetic tests show you did not inherit a BRCA1 or BRCA2 mutation, your

cancer cells may contain or develop one over time. This is called a somatic mutation. Your doctor will want to test the cancer for these mutations through comprehensive biomarker testing. Knowing if you inherited a BRCA mutation or if your cancer tumor developed one helps your doctor decide which treatments are best for you.

COPING WITH A BRCA GENE MUTATION

Living with inherited cancer risk can create challenges that you have never faced before. You may now have to figure out the best path forward to stay on top of your future cancer risk. Try not to feel pressured to make your decisions quickly. Talk with your health care team about how much time you can reasonably take to fully process the information before making any decisions.

Bring up any questions or concerns to your health care team. During your initial appointment, you are likely getting a crash course in cancer genetics while also facing a great deal of uncertainty. Knowledge is power, and you have options to stay on top of your future cancer risk. Talk with your doctor about the different ways to lower your cancer risk.



QUESTIONS TO ASK YOUR HEALTH CARE TEAM

Have you performed biomarker or genetic testing?

How will the results of these tests affect my treatment options and long-term outlook (prognosis)?

Should I continue taking my hormone replacement therapies and/or birth control?

When and how should I tell my family about my results?

Should my family go through genetic testing?

Can I still have children?

What options do I have for fertility preservation? At what point during my treatment can I do that?

What can I do to reduce my risk or find cancer at an earlier stage?

When is the right time to consider medication or surgery?

Should I see a specialist?

How often will I need to see my regular doctor, gynecologist, or oncologist?

Am I eligible for any clinical trials?

What resources do you have for practical, financial, and emotional support?





What are the pros and cons of each option? For future appointments, write down your questions ahead of time and bring someone with you to take notes if possible. Keep in mind that recommendations and what you are comfortable with now may also change over time.

Take care of your overall health. Try to eat well and get enough sleep. Try to stay physically active, even if that means taking a 15-minute walk. Limit any use of substances, including smoking and alcohol. Try not to judge yourself harshly if these changes don't happen right away. Doing things that make you happy can also help reduce stress and keep you going no matter where you are in your experience.

Stay connected to others and find support around you. It is important to share your feelings with people you are comfortable with and who are important to you. Give your partner or spouse permission to feel too. Let them know what they can do to help support you. Ask for help if you feel overwhelmed. Talking with a counselor or therapist can help you cope with distress, anxiety, and depression. Share your concerns with a health care professional you trust. Your treatment team may be able to connect you with a support group with others going through similar experiences.

GENETIC TESTING RESOURCES

National Cancer Institute: Cancer Genetics Services Registry · www.Cancer.gov/CancerTopics/Genetics/Directory

The National Society of Genetic Counselors · www.NSGC.org

BREAST CANCER RESOURCES

Cancer Support Community · 888-793-9355 · www.CancerSupportCommunity.org/Breast-Cancer

Living Beyond Breast Cancer · 855- 807- 6386 · www.LBBC.org

National Breast Cancer Foundation · 972-248-9200 · www.NationalBreastCancer.org

National Cancer Institute (NCI) · 800-422-6237 · www.Cancer.gov

NCI Clinical Trials Registry · 888-422-6237 · www.Cancer.gov/ClinicalTrials

SHARE Cancer Support · 844-275-7427 · www.ShareCancerSupport.org



CANCER SUPPORT COMMUNITY RESOURCES

Cancer Support Helpline® — Have questions, concerns or looking for resources? Call CSC's toll-free Cancer Support Helpline (888-793-9355), available in 200 languages Mon-Thurs 11am-8pm ET and Fri 11am-6pm ET.

Open to Options® — Preparing for your next appointment? Our trained specialists can help you create a list of questions to share with your doctor. Make an appointment by calling 888-793-9355 or by contacting your local CSC or Gilda's Club.

Frankly Speaking About Cancer® — Trusted information for cancer patients and their loved ones is available through publications, online, and in-person programs.

Services at Local CSCs and Gilda's Clubs — With the help of 190 locations, CSC and Gilda's Club network partners provide services free of charge to people touched by cancer. Attend support groups, educational sessions, wellness programs, and more at a location near you. www.CancerSupportCommunity.org/FindLocation.

Cancer Experience Registry® — Help others by sharing your cancer patient or cancer caregiver experience via survey at www.CancerExperienceRegistry.org.

MyLifeLine® — CSC's secure, online community welcomes anyone impacted by cancer to easily connect with community to reduce stress, anxiety, and isolation. Create a personal network site and invite friends & family to follow your journey. And participate in our discussion forums any time of day to meet others like you who understand what you are experiencing. Join now at www.MyLifeLine.org.

Grassroots Network — Make sure your voice is heard by federal and state policy makers on issues affecting cancer patients and survivors by joining our Network at www.CancerSupportCommunity.org/Become-Advocate.

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