FRANKLY SPEAKING ABOUT CANCER

BRCA1/BRCA2 Mutations

What's In Your Genes: How changes in the BRCA1/BRCA2 genes might affect you and your family

Our genes contain the codes for producing the proteins that are the building blocks of our bodies and the foundation of our health. But sometimes we inherit changes in our genes. Scientists are finding that these changes (also called mutations) make some people more prone to develop certain diseases.

For example, everyone has the BReast CAncer (BRCA) genes – BRCA1 and BRCA2. Our bodies typically read these genes and produce proteins that help repair damaged DNA. But if there is a change in one of these genes – much like a typo – these protective proteins may not work as well. Without this "autocorrect," harmful errors in a cell's DNA can build. As these mistakes accumulate, they can allow the cell to grow and divide uncontrollably. If this happens, breast, ovarian or, in some cases, a melanoma or pancreatic cancer can develop.

> *"When I learned I was BRCA positive, I was told 'your body is the same as it has been for the past 30 years, you are just learning more about it.' This has helped me so much."*
>
> *— Jessica*

If you or other people in your family have had breast or ovarian cancers, especially at an early age, your doctor may recommend genetic counseling and testing.

Learning that you or a loved one carries a mutation in either the BRCA1 or BRCA2 genes, can be upsetting – even shocking. You may feel overwhelmed by the amount of information you now need to consider and how that affects your life. But knowing that you have a mutation can help you make better medical decisions – even if you are being treated for cancer or have a history of cancer. Together, you and your health care team can develop a plan to try to prevent or find cancer early.

CANCER SUPPORT COMMUNITY

GILDA'S CLUB WORLDWIDE

Genetics and Cancer Support & Resources

Bright Pink	312-787-4412	brightpink.org
Facing Our Risk of Cancer Empowered	866-288-7475	facingourrisk.org
National Cancer Institute	800-422-6237	cancer.gov
National Society of Genetic Counselors	312-321-6834	nsgc.org

What does having a mutation in the BRCA1 or BRCA2 gene mean?

Overall, it means that you are much more likely to develop breast and/or ovarian cancer. How much more likely? There is a 50 to 85 percent chance that you will develop breast cancer during your lifetime. By comparison, women in the general population only have a 12 percent lifetime risk of the disease.

Your risk of ovarian cancer also jumps from 1-2 percent (for the average woman) to between 15 to 60 percent lifetime risk. Research also points to slightly higher rates of melanoma, prostate and pancreatic cancers, especially among BRCA2 carriers. This information not only impacts your own cancer risk, but that of your family members too.

But there are still many unknowns. For example, being a carrier does not mean that cancer is a sure thing. Current testing also can't tell you:

- Your exact risk
- At what age you may develop cancer
- Which cancer you may develop (breast, ovarian, pancreatic or skin)

> *"BRCA1/2 can be especially challenging because the threat of cancer extends beyond your own risk to other family members, who may also be your closest supports. Balancing everyone's needs and coping styles can make health decisions and family communication more complicated."*
>
> *— Karen Hurley, PhD, Psychologist*

Finding out if you carry the (mutated) gene

Trying to understand the science behind cancer and genetics and thinking in terms of your future risk can be daunting. A genetic counselor – someone with specific training in how certain cancers run in families – can help.

Genetic counselors ask detailed questions about who in your family (on both your mom and dad's side) has had cancer and at what ages. This information helps them determine 1) if you or other family members need testing and 2) which genetic test is most appropriate (there are other genes linked to breast and ovarian cancer aside from BRCA1 and BRCA2). These days, many insurance companies require genetic counseling by a certified provider before genetic testing is ordered. Most hospitals have a genetic counselor on staff; however, if there isn't one in your area, a company called Informed DNA provides genetic counseling over the phone.

Among other things, a genetic counselor can help:

- Assess the likelihood that the cancers in your family are hereditary
- Determine which genetic testing is best for you/family members
- Guide you through the "what ifs" and pros and cons of testing
- Explain what the results might mean for you and your family
- Craft a tailored medical management plan based on your results and family history
- Refer you to other resources

Genetic tests typically involve a blood or saliva sample. It usually takes a couple of weeks to get the results.

During this process, many people find it helpful to seek additional support from other people who have a mutation, support groups, or a psychologist to help them sort through their feelings and medical choices.

Questions about Testing

- What is the right genetic test for me?
- How will this information affect my cancer risk? Other people in my family?
- Can my insurance company drop me if I have a mutation?
- How do I protect my privacy? Will I lose my job?
- Will I be able to get life or disability insurance?

I already have cancer, so how is this going to help me?

Genetic tests do not always give a simple yes or no answer. Testing a family member who already has cancer generally provides the most information, and should be done first, if possible.

Knowing whether or not you have a mutation in one of the BRCA genes can:

- Tell you and your doctor about your risk for other cancers in the future. This can help shape future treatment, as well as how intensely you should be followed. For example, if you have had cancer in one breast, it may change how the risk of cancer in the other breast should be managed.
- Help other people in your family learn about their risks.

BRCA Facts

- It's not just a woman's concern – men can carry and pass on a BRCA mutation; and they can also develop breast, prostate, pancreatic or skin cancers if they carry a mutation
- Children of carriers have a 50/50 chance of inheriting the mutation
- 1 in 10 breast cancers can be tied to a hereditary predisposition
- BRCA-related breast cancers often present at younger ages and tend to be more aggressive

Now what?

You found out that you or a loved one carries a mutation in a BRCA gene. So now what?

Fortunately, there are a number of options that can help manage your risk of breast and ovarian cancers. It's often a very personal choice.

Options for Monitoring or Reducing Your Risk of **Breast Cancer**

	RECOMMENDATIONS	COMMON CONSIDERATIONS
SCREENING	Beginning at age 25 OR 10 years younger than the earliest breast cancer in your family (whichever is first): • Annual mammogram and breast MRI, each spaced 6 months apart • Clinical breast exams every 6 months	• Frequency of screenings can cause anxiety, renew fears • Early detection improves survival • Option for someone who doesn't want to consider medications or surgery
MEDICATION (CHEMOPREVENTION)	Tamoxifen® or raloxifene (Evista®)	• Side effects • Can't be taken when pregnant or breastfeeding • May not be a good fit for certain tumor types • Provides up to 50% risk reduction for breast cancer • May already be part of a breast cancer treatment plan and can also reduce risk of second cancer
PREVENTIVE (PROPHYLACTIC) BREAST SURGERY	Removing both breasts before cancer strikes, or deciding to remove the second breast if you've already had cancer in the other	• Feels radical for some women • Concerns about what "they" will look like • Potential problems like pain, loss of sensation, etc. • Risks of surgery • Logistics during recovery (not being able to lift for period of time, etc.) • Can reduce likelihood of breast cancer by more than 90%

Options for Monitoring or Reducing Your Risk of **Ovarian Cancer**

	RECOMMENDATIONS	COMMON CONSIDERATIONS
SCREENING	Beginning by age 30 OR 5-10 years younger than the youngest ovarian cancer diagnosis in the family (whichever is youngest): • Transvaginal ultrasound and blood test to check for levels of CA-125 every 6 months	• Anxiety about screening or that an early stage cancer might be missed • Option for young women and those who don't want to consider surgery
MEDICATION (CHEMOPREVENTION)	• Birth control pills	• Other risks (blood clots) • May not be appropriate for women who have had breast cancer • Can reduce the risk of ovarian cancer (by up to 60%) if taken for several years
PREVENTIVE (PROPHYLACTIC) SURGERY	• Removing the ovaries and fallopian tubes either by age 40, after childbearing or 10 years before the earliest ovarian cancer in the family (whichever is youngest)	• Early menopause (hot flashes, sexual changes, problems sleeping, moodiness, etc.) • Usual risks of surgery • Concerns about hormone replacement therapy • Worries about exchanging a cancer risk for other risks (heart disease, osteoporosis) • Cancer risk can be significantly reduced (by >90-95%) • If performed before menopause, also reduces breast cancer risks

An annual skin and eye exam may be recommended for people with mutations in the BRCA2 gene.

Wading through the options

Figuring out the best path forward to stay on top of your future cancer risk may take some soul searching.

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 decisions. Being thorough now can help reduce the chance of regrets later on.

- **Don't be shy about asking questions.** You are likely getting a crash course in cancer genetics while also facing a great deal of uncertainty.
- **Think about how medical choices to lower your cancer risk fit your life.** What are the pros and cons of each option? How might each affect your life and the activities that give it meaning?
- **Take deep breaths.** You may get some (sometimes strong) opinions from other people about what you should do. Stay true to yourself.
- **Talk to other women who have had preventive surgery or take tamoxifen.**

Keep in mind that recommendations and what you are comfortable with now may also change over time. Your age and life priorities will likely play a role.

Common Concerns

Like other people who carry a BRCA1 or BRCA2 mutation, you may experience:

- **Ongoing uncertainty** – about what this means, what you and your family members might do with the information and if you even want to know.
- **Fears** that you are damaged, or that cancer is already lurking in your body.
- **Body issues** – conflicting feelings about your breasts or how your body and sex life might change with certain preventive treatments.
- **Pressure** to check things off your life's list faster (having children, finding a partner, etc.).
- **Concerns about when and how to tell your family** – remember, while this information may be difficult to take in, it is something previous generations didn't have access to and may help save lives.
- **Feelings of guilt** that you might pass the harmful gene onto your children.
- **Concerns that you won't be able to get affordable life or disability insurance** — the laws that protect against genetic discrimination do not extend to life and disability insurance.
- **Costs of care.** In the vast majority of cases, insurance will cover the cost of BRCA testing for people who are eligible. There are resources to help if you don't have insurance.

Having a BRCA1 or BRCA2 mutation can present special challenges and worries for many young women. For example, many feel added pressure to find a life partner or start having children. There are resources and counselors available to help work through all of these concerns. Visit *www.brightpink.org.*

Coping & Next Steps

Living with inherited cancer risk can create challenges that you have never faced before. Think about other times when you had to do something difficult or faced serious news – what helped you get through it? Some tips for coping:

- **Be empowered.** Knowledge is power, and you have options to stay on top of your future cancer risk.
- **Pace yourself.** Think about the one thing you need to do next, and ask for help if you feel overwhelmed.
- **Use your support system well.** Surround yourself with people who will really listen without judging you and who remind you of life outside of the BRCA1/BRCA2 gene.
- **Be prepared for appointments.** Write down your questions ahead of time and bring someone with you to take notes.
- **Keep all of your health care providers up-to-date on any changes in your family history.** Recommendations for managing your risk may change as a result. Try to keep information about your family's cancer history, medications and results of screening tests in one place.
- **Use distraction in small doses.** Bring a favorite book, magazine, or music with you when waiting for your mammogram or related medical visit.
- **Use positive self-talk.** "One day at a time" or "Other families have heart disease or diabetes, mine has a high cancer risk."
- **Keep up healthy routines.** Exercising, eating well and doing things that make you happy can help reduce stress and keep you going no matter where you are on the spectrum from testing to treatment.
- **Give your partner or spouse permission to feel too.** Let them know what they can do to help support you.
- **Seek professional support.**

Questions About Carrying the Gene

- What can I do to reduce my risk or find cancer at an early, non life-threatening stage?
- Should I see a breast specialist?
- When and how should I tell my children?
- When is the right time to consider medication or surgery?
- Should I have my ovaries and fallopian tubes out or a total hysterectomy? If so, when?
- Can I still have children?
- How often will I need to see my regular doctor, gynecologist or oncologist?
- Are there other doctors I should see or tell (for example, my ophthalmologist or dermatologist?)

Valuable Resources

Help is available for you and your loved ones.

CSC Resources for Support

Cancer Support Helpline® 1-888-793-9355
Our free helpline is open Monday – Friday 9:00 am – 8:00 pm ET. Anyone impacted by cancer can call to talk to a call center counselor. We will connect you with local and national resources, and help you find the right support.

Affiliate Network
Over 50 locations plus more than 100 satellites around the country offer on-site support groups, educational workshops, and healthy lifestyle programs specifically designed for people affected by cancer at no cost.

For more information, visit www.cancersupportcommunity.org or call 1-888-793-9355.

Other Resources and Support

Bright Pink	brightpink.org
Cancer Legal Resource Center	disabilityrightslegalcenter.org
Facing Our Risk of Cancer Empowered	facingourrisk.org
Informed DNA	informeddna.com
MedlinePlus [search: BRCA]	medlineplus.org
National Cancer Institute: Cancer Genetics Services Registry	cancer.gov/cancertopics/genetics/directory
The National Society of Genetic Counselors	nsgc.org

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The Cancer Support Community provides this information as a service. This publication is not intended to take the place of medical care or the advice of your doctor. We strongly suggest consulting your doctor or other health care professionals to answer questions and learn more.

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