



BRCA Genetic Testing: What You Need to Know

Finding out if you have any genetic mutations can tell you about your risk of developing breast and ovarian cancers

By Cynthia Ramnarace

When Joanna Rudnick's mother tested positive for BRCA, the so-called breast cancer gene, Joanna's life changed in ways she never could have prepared for.

At 27 years old, Joanna immediately also got tested to see if she, too, was a carrier of the BRCA gene mutation. A simple blood draw confirmed that she had a higher-than-average chance of someday developing breast and ovarian cancers. "It's difficult to prepare yourself for how much that information really can change your life," says Rudnick, who directed *In the Family*, a documentary film about her experience.

After testing positive, Rudnick initially felt like "a ticking time bomb." But later, she became grateful for the information. "I hopefully will not suffer the same fate as many of my family members who were shocked by these diseases," she says.

What Is BRCA?

Between 5 and 10 percent of women who develop breast cancer have a hereditary form of the disease. In many cases this is due to a mutation, or error, in one of two genes: BRCA1 and BRCA2. When these genes function properly, they help prevent cells from growing abnormally. Mutations disable this disease-preventing function, thus increasing the odds that cancerous cells may form.

Should You Be Tested for BRCA?

There are many factors affecting whether or not you should get tested for BRCA. In general, testing may be appropriate if you have:

- Multiple close blood relatives affected by breast and/or ovarian cancer
- At least one close relative with both breast cancer and ovarian cancer
- At least one close relative who developed breast and/or ovarian cancer before age 50
- At least one close blood relative who developed more than one breast tumor
- Male blood relatives who developed breast cancer
- Ashkenazi Jewish heritage

"The more breast and particularly ovarian cancers there are among your blood relatives, the higher the chance you will have a genetic predisposition to disease," says Martee L. Hensley, MD, Everyday Health cancer expert and associate attending physician in gynecologic medical oncology at Memorial Sloan-Kettering Cancer Center in New York. Breast and ovarian cancer survivors should also consider genetic testing. Because BRCA increases your risk of cancer at both sites, you may want to know if you should consider prophylactic treatment such as ovary removal or mastectomy to reduce your risk of a second cancer.

What BRCA Testing Tells You

If you test negative for a BRCA gene, your risk of breast cancer is the same as that of the general population: 13 percent. If you test positive for either BRCA1 or BRCA2, your lifetime risk is between 36 and 85 percent for developing breast cancer and between 16 and 60 percent for ovarian cancer. The range is wide, which is why genetic counseling in tandem with testing is so important, according to Angela Trepanier, president of the National Society of Genetic Counselors. "If I take a family history and it's loaded with breast and ovarian cancers, the guess is that the risk is probably at the higher end," she says.

What Should I Do if My Test Is Positive?

If you test positive for either gene, doctors recommend one of the following:

- Increased monitoring for both cancers, including annual mammograms and magnetic resonance imaging (MRI) for breast cancer, and routine transvaginal sonograms and blood screenings for CA125, a marker for ovarian cancer. However, while breast cancer can be detected in its earliest stages using these screening methods, early ovarian

cancer screenings are not as accurate.

- Risk-reducing surgery, including a double, or bilateral, mastectomy and the removal of the ovaries (medically known as an oophorectomy).

Ovary removal is recommended for all BRCA carriers once they have passed the childbearing stage of their lives — generally between ages 35 and 40 for BRCA1 carriers and between ages 35 to 45 for BRCA2 carriers.

In the Los Angeles oncology practice of Kristi Funk, MD, 70 percent of the women who test BRCA-positive eventually opt for mastectomy and 30 percent choose routine surveillance. "It comes down to how a woman feels about her breasts and her sexuality, and how much of her femininity is tied up in her breasts," says Dr. Funk, who is director of patient education at the Saul and Joyce Brandman Breast Center, at LA's Cedars-Sinai Medical Center.

Other Considerations

Genetic testing is expensive and may not be covered by insurance. You should also consider the implications that your decision will have on family members (who might not want to know that they are at increased risk if your test comes back positive) and spouses or partners. Having a strong, informed support network will help you make the crucial decisions that might lie ahead.

Ultimately, the decision to be tested for BRCA should be made only after speaking with a health-care professional trained in genetics. For more information or for help finding someone with this training, contact the National Society of Genetic Counselors or the National Cancer Institute.



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