

steps

Who's at risk?

Genetic testing for breast cancer is a hot topic, but it can be confusing.

Here's a guide to help you understand what it's all about

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Women with breast cancer often wonder what caused their cancer and are concerned that other female family members might get it too, worrying that it might be inherited. There's been a lot of media coverage about testing for genes that may indicate a greater risk, but not much has been written about the process itself — and the possible consequences. This leaves a lot of unanswered questions. We'll provide you with some answers.

What are genes anyway?

Genes are the blueprints for all living things — they determine how the body will function and grow. They're made up of DNA and are carried on string-like structures called chromosomes. Genes come in pairs: one

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towards awareness

member of each pair is inherited from the mother and the other comes from the father. They determine physical characteristics as well as body processes, such as growth rate and metabolism. We each have a genetic blueprint that's uniquely ours.

We all have copies of BRCA1 and BRCA2 genes — named for **B**reast **C**ancer because they're associated with a greater breast cancer risk if they have mutations (errors) in them. They normally control cell growth in organs such as the breast and ovary.

Mutations and risk

If you have a genetic susceptibility to cancer, this can arise from a change in a gene that makes it act differently. An alteration or “mistake” in the BRCA genes could impair their normal function, allowing cells to multiply excessively, which can lead to cancer.

BRCA1 and BRCA2 genes show what's called a dominant pattern of inheritance. This means that a single inherited mutation or alteration in one of them is enough to cause a susceptibility to breast and ovarian cancer. It can come from either your father or mother, but we all have two copies of every gene. For cancer caused by BRCA1 or BRCA2 changes, a second sporadic — that is, chance — alteration in the other copy of the same gene must be there. In other words, both copies must be affected before pre-cancerous cells can appear.

Mutations in these genes carry an increased chance of developing breast and ovarian cancer. It's very important to remember, however, that it's only a risk. Just because a woman has this inherited factor, it doesn't mean she'll necessarily get cancer. We can't predict when — or if — this will occur in a particular person.

The family history factor

When researchers discovered the BRCA1 and BRCA2 relationship to breast and ovarian cancer, it opened the door to developing tests for their mutations. Testing is generally offered to women who have a significant family history of breast and/or ovarian cancer.

You're considered to have a strong history if two or more close family members have had breast and/or ovarian cancer, and if breast cancer was diagnosed before age 50. Close family members include your mother, sister, aunt or grandmother on either side, and even male members — brother, father or uncle.

How is testing done?

A blood test has been designed to look specifically at the BRCA1 and BRCA2 genes for any mutation or alteration. The clinic performing the test will:

- determine your eligibility for testing by looking at the family history of cancer as well as your personal background (for example, ethnicity)

- provide genetic counselling before taking the blood sample (see box below)
- obtain your informed consent to do the test
- schedule a session to discuss your results
- arrange for any necessary follow-up visits
- provide referrals to other health professionals as appropriate

After you're given the blood sample, a two-step process begins. The first involves looking for BRCA gene mutations in your sample and comparing it to that of a family member who's had breast or ovarian cancer. This helps identify an inherited predisposition.

Genetic counselling

It's not as intimidating as it looks. Here's what to expect:

- a review of your family medical history
- confirmation of pathology records of relatives who developed cancer
- an overview of basic genetic information
- an individual risk assessment
- information on the advantages, disadvantages and limitations of testing
- a recommendation about testing and the meaning of test results
- screening recommendations for those who are at risk of developing breast cancer

If the test turns out **negative** — there's no BRCA change or mutation — but there's a strong family history, then it's called an **uninformative result**. This means that there may be mutations in BRCA1 and BRCA2 that we can't detect with the current testing, or there may be other mutations in genes we haven't yet discovered.

A **positive** test result, which reveals a mutation, indicates that there's an inherited, greater chance of getting breast and ovarian cancer (see box below). Other blood relatives may then choose to participate in genetic testing (the second step). If their tests are negative, they're not at increased risk and can set their minds at ease.

Emotional concerns

Participating in genetic testing is a complicated process. If you're con-

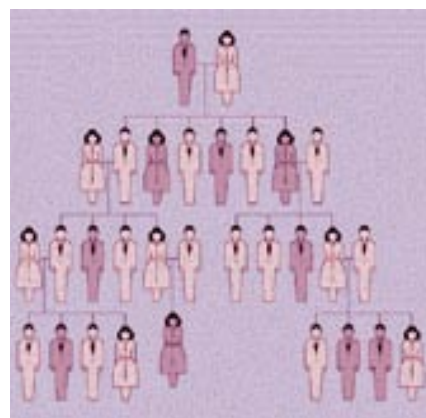
sidering it, discuss the pros and cons with a professional who's specially trained in genetics. Keep in mind that the test has some limitations — for example, it can't determine who will or won't get cancer. The psychosocial impact on you and your family members should also be weighed.

If the result is positive, you may feel one of several emotions. These of course vary, depending on the person. Some of the more common ones include: anxiety, depression, fear, anger, guilt and decreased body image and self-esteem. You might also have sleep problems, intrusive thoughts about cancer and constant worry, and your family and sexual relationships may change. A health-care professional can help you sort through these troubling issues.

Pros and cons

Genetic testing can resolve a woman's fears and guide decisions with regards to screening and prevention. If the test is positive, it can prompt her to follow screening recommendations. She may want to let loved ones know she's at increased risk and might decide to make some lifestyle changes — such as eating a healthy diet and exercising — to minimize her chance of developing cancer. Close family relatives might want to get tested as well. She might also choose to be part of a research project that follows women with this genetic mutation. For women whose test turns out fine, particularly if there's a known mutation in the family, it can bring peace of mind.

But there are some disadvantages, too. If the test is positive, it may bring on emotions such as depression, anger or anxiety, or there may be difficulty coping with cancer risk. Other areas of life may be involved as well — for instance, it might be harder to qualify for health or life insurance.




Even a negative test can have consequences. For example, it might cause a false sense of security — it doesn't mean you won't get breast cancer. The chance is still there, but it may be as low as that of the general population, depending on the family history. Regardless of

“Just because a woman has this inherited difference, it doesn't mean she'll necessarily develop cancer”

the results, it's still important to maintain a healthy lifestyle to minimize the risk.

Down the road

Whether to get tested depends on several factors, with family history being the most influential. It's not a decision to be taken lightly. Some women might not want to know whether they have the mutation — a personal decision that must be respected. If your doctor offers you the test, talk it over with your family, your doctor and a genetic counsellor before taking this important step. Understanding all the pros and cons will help you make an informed decision. 

The facts on risk

What difference does a BRCA1 or BRCA2 mutation make?

- Women with a BRCA1 gene mutation have a 38–86% lifetime risk of developing breast cancer and a 15–40% chance of ovarian cancer up to age 70 or 80, but this appears to be lower for BRCA2 mutation carriers.
- The general population risk of breast cancer is 7% by age 70 and 10% by age 80.
- Those in families with known genetic alterations who don't carry it themselves have the same chance as the average person of developing breast or ovarian cancer.