HEALTH BASICS

Breast cancer and your genes

This explains how inherited and tumor gene changes can affect breast cancer risk, testing, and care.

WHAT IT IS
How gene changes relate to breast cancer risk and care

WHERE IT COMES UP
Family history, breast cancer care, or genetic test results

WHY IT MATTERS
Helps guide risk assessment, treatment, and family testing

WHAT IT IS NOT
Not a disease or a breast cancer diagnosis

WHO TO ASK
Genetic counselor, oncology team, or primary care clinician

HOW IT IS CHECKED
Blood or saliva testing, and sometimes tumor testing

Overview

Breast cancer and your genes is a way of understanding how genetic changes may affect cancer risk or care. It is not a disease or a diagnosis.

Genes are sections of DNA that give your cells instructions. A genetic change is often called a variant . Most variants do not cause disease. Some harmful variants can raise the chance of developing certain cancers.

The best-known breast cancer genes are BRCA1 and BRCA2 . Other genes linked with breast cancer include PALB2, TP53, PTEN, CDH1, ATM and CHEK2. These genes do not all carry the same level or type of risk.

Most breast cancers are not caused by a harmful variant inherited from a parent. Cancer can also develop through genetic changes that build up in breast cells during a person's life.

Having an inherited harmful variant does not mean that you have breast cancer or will certainly develop it. A negative genetic test also does not guarantee that breast cancer will never occur.

How it works

You inherit one copy of most genes from each biological parent. An inherited genetic change is called a germline variant . It is usually present throughout the body and can sometimes be passed to biological children.

Some germline variants interfere with how cells repair damaged DNA or control growth. This can make it easier for further cell changes to build up. Over time, those changes may contribute to cancer.

People of any sex can inherit and pass on breast cancer-related variants. A family history on your father's side is just as relevant as one on your mother's side. Some variants are also linked with ovarian, pancreatic, prostate, stomach or other cancers.

Genetic changes found only in cancer cells are called somatic variants . They are acquired during life and are not usually inherited. Testing a breast cancer sample can find somatic changes that may help guide treatment.

Tumor testing and inherited-risk testing answer different questions:

A laboratory may report a variant as pathogenic, likely pathogenic, uncertain, likely benign or benign. Pathogenic means there is strong evidence that the variant can contribute to disease. A variant of uncertain significance , or VUS, means there is not enough evidence to know whether it matters.

- Germline testing usually uses blood or saliva to look for inherited variants
- Tumor testing examines cancer tissue for changes that may affect treatment
- A tumor result may suggest an inherited variant, but blood or saliva testing is usually needed to confirm it

Why it matters for your health

An inherited pathogenic variant may change how your healthcare team assesses your future cancer risk. Depending on the gene and your circumstances, it may affect discussions about screening, risk-reducing medicines or surgery. These choices are personal and should be based on the specific result, your medical history and your preferences.

If you already have breast cancer, some inherited or tumor variants may affect treatment choices. For example, certain cancers with BRCA-related changes may respond to medicines that target weaknesses in DNA repair. A result may also influence discussions about surgery or monitoring for another cancer.

A confirmed inherited variant can be relevant to biological relatives. Testing relatives for a

known family variant is called cascade testing . It can show who carries that specific variant and who does not.

Genetic results can also bring uncertainty or worry. They may raise questions about children, siblings or parents. A genetic counselor can explain what the result does and does not mean without assuming that one choice is right for everyone.

What can go wrong

Harmful variants in certain genes are associated with named hereditary cancer syndromes. Examples include hereditary breast and ovarian cancer associated with BRCA1 or BRCA2, Li-Fraumeni syndrome associated with TP53, PTEN hamartoma tumor syndrome and hereditary diffuse gastric cancer associated with CDH1.

These conditions have different cancer associations. A gene name alone is not enough to interpret risk. The exact variant, personal history and family history all matter.

Testing can also produce unclear or misleading situations:

Direct-to-consumer tests may cover fewer genes or variants than clinical testing. Results that could affect medical care may need confirmation in a clinical laboratory.

A genetic result is not an emergency and does not cause sudden symptoms. Arrange routine follow-up to review a positive, uncertain or confusing result. Contact a healthcare professional promptly for a new breast lump, bloody nipple discharge, skin dimpling or another new breast change, but these changes are not caused by receiving a genetic report. Emergency care is not usually needed for a genetic finding itself.

- A VUS may be mistaken for a harmful result, even though it should not be treated as proof of increased risk
- A negative result may be falsely reassuring if no affected relative was tested first
- Some tests examine only a few variants and may miss other relevant changes
- Tumor results may be confused with inherited results
- A variant may be reclassified as scientific evidence develops
- Unexpected family relationships or information may sometimes be revealed

What you can do

If you are concerned about inherited risk, record cancer diagnoses on both sides of your

biological family. Include the type of cancer and, if known, the age at diagnosis. Breast, ovarian, pancreatic, prostate and certain other cancers may be relevant.

Questions you can ask include:

If you have breast cancer, ask whether you are being offered germline testing, tumor testing or both. Ask what clinical decision the test is intended to inform.

Keep a copy of your full laboratory report. Record the laboratory name, exact gene and variant, classification and test date. A brief patient portal message may leave out important details.

Review a VUS with the ordering service rather than using it to make major medical decisions. Laboratories may update classifications, so ask how you would be told about a change.

If you already know that a pathogenic variant runs in your family, bring a copy of that relative's report. Testing for the exact family variant is usually more informative than relying on memory of the gene name.

There may be nothing you need to do if testing is not indicated and your routine breast care is appropriate for your age and history. A genetics professional can help separate useful testing from testing that is unlikely to provide a clear answer.

- Does my personal or family history suggest genetic counseling?
- Would testing an affected relative first give a clearer answer?
- Which genes would the proposed test examine?
- Could the result affect my treatment, screening or relatives?
- What would a negative result or VUS mean in my situation?
- Are there privacy, insurance or employment protections where I live?

Common questions

Does having a BRCA variant mean I have breast cancer?

No. A pathogenic BRCA1 or BRCA2 variant raises the chance of certain cancers, but it is not a cancer diagnosis. Some people with these variants never develop breast cancer.

Can breast cancer skip a generation?

A harmful variant can be passed through someone who never develops the associated cancer. Small families, limited health information and differences in risk can make a hereditary pattern difficult to see.

Can I inherit a breast cancer-related variant from my father?

Yes. People of any sex can carry and pass on these variants. Cancer history on both sides of the biological family matters.

Is a VUS a positive result?

No. A VUS is an uncertain result, not proof of increased cancer risk. Medical decisions are generally based on personal and family history until the variant is better understood.

If my genetic test is negative, am I at average risk?

Not necessarily. A negative result may be reassuring when a known family variant was tested for and not found. In other situations, family history or other factors may still affect risk.

Should children be tested?

Many breast cancer-related genes mainly affect adult health, so testing children may not provide an immediate medical benefit. Some genes can affect care during childhood. A genetics service can advise based on the exact gene and family situation.

Sources

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