Genetics & Biomarker Testing

Gifts of Knowledge
For You & Your Family
“Genetic and Biomarker Testing: Gifts of Knowledge for You and Your Family” is a story told in two parts featuring characters from SHARE’s previous novels who undergo genetic and biomarker testing. As you read about sisters Yvette and Silvia, we hope you learn the purpose and importance of genetic testing. As you read about Ana, who was diagnosed with metastatic breast cancer, we ask that you be proactive about discussing testing with your doctor. We hope these stories will inform, empower, encourage, and clarify some genetic and biomarker testing basics.

Questions this novela will address:

• What is genetic testing? Why is it important?
• Can siblings have different genetic testing results?
• According to NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) • • who should get tested?
• What medical professionals can give genetic testing information or test results?
• How important is it to share your cancer history with your family?
• What does a genetic counselor do? How do I find a genetic counselor?
• How does it change my risk for other cancers if I have an inherited mutation?
• Is it true men can develop breast cancer?
• What is biomarker testing?

RESOURCES
American Board of Genetic Counseling (ABGC) developed a directory to assist health care professionals, patients, the public, and other genetic counselors in locating genetic counseling services. https://www.abgc.net/about-genetic-counseling/find-a-certified-counselor.aspx/1-913-222-8661

American Cancer Society (ACS) researches cancer and treatments, promotes healthy lifestyles to help you prevent cancer, and fights for lifesaving policy changes. They provide everything from emotional support to the latest cancer information to anyone affected by cancer. https://www.cancer.org/1-800-227-2345

American College of Medical Genetics and Genomics (ACMG) has a searchable medical genetics clinic services database. https://clinics.acmg.net/1-301-718-9603

FORCE (Facing Hereditary Cancer EMPOWERED) improves the lives of individuals and families facing hereditary cancer by providing expert-reviewed information to help people make informed medical decisions. https://www.facingourrisk.org/1-866-288-RISK (7475)

NCCN Guidelines for Patients® aims to help cancer patients talk with their doctors about the best treatment options for their disease. https://www.nccn.org/patients/guidelines/cancers.aspx1-215-690-0300


SHARE is a national nonprofit that supports, educates, and empowers women affected by breast, ovarian, uterine, or metastatic breast cancer, with a special focus on medically underserved communities. All of SHARE’s services are free of charge. SHARE’s services include support groups, expert-led educational programs, national breast cancer helpline, ovarian cancer helpline, uterine cancer helpline, metastatic breast cancer helpline, online communities, educational tools, clinical trial assistance, outreach to medically underserved communities, survivor-patient navigation, and more.
Ten years ago, Jacqueline, the Survivor Patient Navigator, interprets what the doctor says from English to Spanish for Silvia, the patient.

I KNEW IT, I’M GOING TO DIE!

«The doctor says the biopsy result was cancer. The tumor is malignant.»

A WEEK LATER.

We’ll remove the tumor and some of the tissue around it that may have cancer cells. This procedure is called a lumpectomy.

A WEEK LATER.

He believes they were able to remove all the cancerous tumors. We have to wait for the pathology report to be sure.

Is she going to need chemotherapy?

The pathology report will let us know if she needs this type of treatment.
How are you feeling?

“I was so tired when I got home after the lumpectomy. I felt some numbness and pain around the surgery area, but after a week, I was back to my routine.”

Today, I’d like to recommend genetic testing for you, Silvia. Genetic testing looks for specific inherited changes or mutations in genes, which are made of DNA. As a breast cancer patient, it can inform your treatment plan.

Genetic tests are done using a blood or saliva sample, and results are usually ready in a few weeks.

“Genetic testing plays a part in determining the risk of developing certain diseases and in choosing appropriate medical treatment. For breast cancer, identifying people with inherited cancer-associated mutations, like in the BRCA1 and BRCA2 genes, is important because some mutations increase the risk of other cancers, like ovarian cancer or can help inform your treatment plan.”

“Why is genetic testing so important?”
«IF I GET TESTED, DOES THAT MEAN MY SISTER IVETTE DOESN'T NEED TO? CAN SIBLINGS HAVE DIFFERENT RESULTS?»

BECAUSE WE SHARE GENES WITH OUR FAMILY, IF YOU HAVE A MUTATION, IVETTE MAY HAVE THE SAME ONE, BUT SIBLINGS CAN HAVE DIFFERENT RESULTS--

--EVEN IF THEY SHARE THE SAME PARENTS, EACH CHILD DOES NOT ALWAYS GET AN EXACT COPY OF THE SAME GENES. BUT BECAUSE THESE MUTATIONS ARE INHERITED, KNOWING WHETHER YOU CARRY ONE OF THESE MUTATIONS CAN BE IMPORTANT INFORMATION YOU MAY WANT TO SHARE WITH YOUR FAMILY, SO THEY CAN POTENTIALLY DISCUSS THEIR RISK FACTORS WITH A DOCTOR.

«SILVIA WANTS TO KNOW SOME PRACTICAL INFORMATION ABOUT GENETIC TESTING.»

SOME BENEFITS INCLUDE:

• LEARNING WHETHER YOU HAVE A GENETIC VARIANT CAN PROVIDE RELIEF FROM NOT KNOWING.
• BETTER KNOWLEDGE OF YOUR HEALTH AND CANCER RISKS.
• INFORMATION TO HELP MAKE INFORMED MEDICAL AND LIFESTYLE DECISIONS TO LOWER RISKS.
• CHANCE TO HELP OTHER FAMILY MEMBERS KNOW ABOUT THEIR POTENTIAL RISKS AND HAVE A FULLER PICTURE OF THEIR HEALTH.
• UNDERSTANDING WHETHER YOU HAVE A GENETIC VARIANT COULD INFORM YOUR TREATMENT IF YOU'VE ALREADY BEEN DIAGNOSED WITH A DISEASE.

SOME CONSIDERATIONS ARE:

• TEST RESULTS CANNOT DEFINITIVELY PREDICT THAT YOU WILL OR WILL NOT GET A PARTICULAR DISEASE. A NEGATIVE TEST ONLY MEANS YOU DON'T HAVE THE GENETIC VARIANT YOU WERE TESTED FOR. DISEASES HAVE CAUSES BEIDES GENETICS, LIKE ENVIRONMENTAL (AIR, WATER) AND LIFESTYLE (UNHEALTHY DIET OR SMOKING).
• TESTING MIGHT INCREASE STRESS AND ANXIETY. IF A GENETIC TEST COMES BACK POSITIVE, IT DOESN'T MEAN YOU WILL GET THE DISEASE. YOU COULD GET A POSITIVE TEST NOW, AND YOU MIGHT NEVER GET THIS DISEASE.
• RESULTS IN SOME CASES MAY RETURN INCONCLUSIVE OR UNCERTAIN.
• POSSIBLE IMPACT ON FAMILY RELATIONSHIPS. FOR EXAMPLE, ONE RELATIVE WANTS TO KNOW IF THEY HAVE A PARTICULAR VARIANT, BUT ANOTHER DOESN'T. OR ONE PERSON DISCOVERS THEY HAVE A GENETIC VARIANT, BUT ANOTHER FAMILY MEMBER FINDS THEY DON'T.

THANK YOU BOTH. LET ME THINK ABOUT IT A BIT AND TALK TO MY SISTER TOO.

AND IF YOU HAVE MORE QUESTIONS AT HOME, YOU HAVE MY NUMBER. DON'T HESITATE TO CALL.
Doctor, I brought my sister Ivette with me. My sister told me about genetic testing, and I have some questions. I'm concerned about the rest of our family too. Who should get tested?

According to the current NCCN Guidelines® (Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic Version 2.2022) that address the risk of developing cancer, anyone who has a personal history of breast cancer and one or more of these criteria:

- **Diagnosed at or before age 45.**
- **Diagnosed between ages 46-50 with:**
  - Additional primary breast cancer
  - 1 or more close relatives (parents, siblings, children) with breast, ovarian, pancreatic, or prostate cancer.
  - Unknown or limited family history.
- **Diagnosed at or before age 60 with triple-negative breast cancer.**

- **Diagnosed at any age with:**
  - 1 or more close relatives with breast cancer diagnosed at age 50 or before.
  - 2+ close relatives with breast cancer at any age.
  - 1 or more close relatives with invasive ovarian cancer, pancreatic cancer, and/or prostate cancer that is metastatic, intraductal/cribriform, or high- or very high risk (eg, Gleason score 8 or higher, PSA >20 ng/mL).
  - Male relative diagnosed with breast cancer.
  - Ashkenazi Jewish ancestry.
  - Diagnosis of male breast cancer.
Who can give me genetic testing information or test results?

A medical geneticist, primary care doctor, specialist, or nurse practitioner can order the test. The lab your sample is sent to then reports the results in writing to your doctor or genetic counselor, who can discuss the findings with you. At our practice, I would be the person to discuss testing with, order the test, and review the results with you.

How important is it to share my results with my family? My daughter would want to know, but I’m not sure about other family members.

Your test results are private, and sharing is a personal decision. But keep in mind how important the information could be to close relatives who might also carry the mutations you do. Sharing that information could help family members identify their risks sooner if you’re positive.

I’m not saying it will be easy to talk to your family. People react with anxiety, guilt, fear, and denial regarding cancer risk. Your daughter might feel grateful to have information she can take to her doctor. Other relatives might not want to know your results or be tested. All you’d be doing is making information available to them and allowing them to make their own health decisions.

Am I prepared for whatever the results will be? What do I say to Belkis or Roberto? What will I do differently if the results are positive or negative? What if Silvia is positive and I’m negative? Or what if I turn out to be positive?
Genetic testing can have three possible results: positive, negative, or positive for “variant of uncertain significance” (VUS). A VUS result means that a mutation was found that may or may not increase the risk for cancer because not all gene mutations are harmful. A VUS result indicates that results were unclear at the time of testing, so it could not be determined whether the gene change is harmful and increases cancer risk or if it’s harmless and doesn’t increase risk.

Your test results were negative, meaning that a breast cancer gene mutation was not identified.
Silvia, your test results were positive, meaning you have a breast cancer gene abnormality. We should discuss how your BRCA status will affect your treatment options.

“WHAT DOES THIS MEAN FOR ME NOW?”

Women with breast cancer and a BRCA1 or BRCA2 abnormality have a much greater risk of developing breast cancer in the other breast or ovarian cancer. Receiving radiation therapy, chemotherapy, or hormonal therapy after a lumpectomy may help lower the risk of recurrence in those with hormone sensitive breast cancer. Let’s discuss your options.
I flew over here as soon as I finished with my last client.

I got my genetic testing results today. The doctor said I’m positive for a BRCA1 mutation.

I need to tell Gerardo and contact his doctor to see if he should get testing done too. I should tell Mami and Papi too.

There’s something else. We went through my options, and together with the pathology report and my testing results, we decided I’ll start chemotherapy this week.

I can help you talk to Gerardo’s doctor, and we can talk to Mami and Papi together.

I was afraid of this. Hasn’t Silvia been through enough?

I was hoping you’d have the same negative results I did. What happens now?
Mi'jo, I have to talk to you about a few things.

I was tested to see if I have any known cancer-causing mutations in my genes. I have one in a gene called BRCA1.

We could speak to your doctor if you'd like to see what he recommends in regards to genetic testing to see if you may have inherited my genetic mutation. You're almost an adult, and soon you'll be taking care of your own appointments.

Remember, always tell your doctor that your mother was diagnosed with breast cancer and a BRCA1 mutation.
Terrible. Her genetic test results were positive, meaning that a mutation is present in one of her genes, BRCA1.

She feels guilty and wonders if Gerardo may have inherited the same mutation. She’ll talk with him about the possibility of getting tested since she knows it’s important.

These tests can’t say one way or the other 100% that a person will develop a disease. I wouldn’t want it done. I’d spend my whole life waiting for something to get me.

At first, I thought the same, but what if I had a mutation, and Belkis might be at a higher risk for cancer? I would want her to know as early as possible so her doctor could monitor her and order the right tests at her check-ups.

I’ll call Gerardo after Tia Silvia speaks with him to see how he’s doing.

Great idea. He’ll appreciate you checking on him.
ANA, I asked you to come in for this follow-up with a family member because the back and hip pain you've been having isn't muscular. We found breast cancer that has metastasized.

Consider joining a metastatic telephone support group; you'd have the support of other women with a metastatic diagnosis in addition to our support group here.

I finally stopped feeling hopeless about this disease, and then my treatment stopped working. How can I live on this rollercoaster?

You're not alone, Ana. This support group exists so that you don't have to go through the ups and downs alone.

Weeks later, it makes me happy to see you feeling better these days, amor.

A year later, Ana's son accompanies her as Ana begins her second line of treatment due to her cancer progression.

Three years ago.

Hi, I'm Ana. I registered to train to become a support group facilitator.

Yes! We spoke on the phone. Come in. We'll get started in a moment.

SHARE Support Group Facilitator Training
- Welcome
- Introductions
- About

Hi, I'm Ana. I registered to train to become a support group facilitator.
What a coincidence we talked about genetic counseling today in the group. My doctor recommended it, but I just wasn’t sure. Do you think you might be able to accompany me to the appointment?

Absolutely, Aiko. Let’s coordinate our times tomorrow.

I’m up to my nose in appointments. Do you think this genetic counseling is necessary?

I do. According to the National Comprehensive Cancer Network, all of us with metastatic breast cancer should get genetic and biomarker testing.

Months Later

A Week Later

What’s the difference?

We inherit our genes at birth and pass them on to our children. Genetics explain why you have black hair, and I have brown hair. With cancer, genetic testing looks for specific changes or mutations in genes like BRCA1 and BRCA2 that may increase the chances of getting cancer. For example, suppose a person has a family history of breast cancer or already has cancer. In that case, they may get genetic testing for specific mutations that may increase the chance of developing breast or other cancers. However, not all cancers have a hereditary link.
Now, biomarker testing is done for people who already have cancer. It can tell us more about the cancer itself by looking at proteins, genes, and some other matter. Each person’s cancer is different, and some biomarkers can impact how certain treatments work. This can be beneficial information for those of us with metastatic breast cancer. For example, suppose your doctor sees that a mutation matches a known cancer cell defect? In that case, they might recommend a treatment designed to target that specific defect, a targeted therapy. In some instances, some biomarkers can be used to track how your cancer is responding to treatment.

The cost can depend on the complexity of your tests, but don’t let that scare you. Many insurance plans cover the cost of genetic testing, especially when your doctor recommends it. Add that question about cost to the list we made, so you can remember to ask the genetic counselor.

What about the cost of genetic testing? It sounds expensive.

Ana, the cost can depend on the complexity of your tests, but don’t let that scare you. Many insurance plans cover the cost of genetic testing, especially when your doctor recommends it. Add that question about cost to the list we made, so you can remember to ask the genetic counselor.
As a genetic counselor, I provide information, offer support, and address your questions and concerns. We'll discuss your medical history, and I'll take a detailed family history, including at least three generations. Your family tree will include which family members have had cancer, the type, and their age at diagnosis. We'll form a strategy for genetic testing that best meets your needs. I'll explain current laws regarding the privacy of genetic information, and I'll share the benefits and limitations of genetic testing for you and your family.

I have some questions. How likely is it that I have a cancer-related mutation? Which genetic test do you recommend? I have metastatic breast cancer. Will genetic testing help predict if it will progress? Does health insurance pay for testing?

I tested positive when I had genetic testing done three years ago. I never told anyone. I was so stressed with treatments and the newness of it all. I said I'd deal with it later, and it slipped under more pressing things.

Hi, I have some genetic testing results from three years ago. Do you think I could schedule an appointment with the counselor to discuss them?

Everyone is going to be upset I didn't say anything sooner. What if I've passed the mutation on to Isabelita? What if my granddaughter gets cancer when she grows up because of my genes?
How do I share this with my family? I’d feel terrible if I passed that genetic mutation to my son or my granddaughter.

How do I share this with my family? I’d feel terrible if I passed that genetic mutation to my son or my granddaughter.

If you share this with David, you give him the chance to monitor and take preventive measures that could reduce his and his daughter’s risk of disease or complications. It is not too late to share the information. A delayed diagnosis could affect treatment options. Yes, he may have strong feelings about not having the opportunity to decide if he wanted to have these results earlier. Still, he’ll now have the same access to testing and related risk-reducing options.

Think about whether you want to tell David one-on-one or with other relatives present like his spouse or father. Consider how he likes to receive information, what he already knows about cancer in the family and genetics. Emphasize that your positive result doesn’t mean he or his daughter will necessarily get cancer in the future. Give him a copy of your genetic test results so he’ll have the name of the specific gene and mutation. If he has questions you can’t answer, remind him of resources like SHARE, the cancer support organization you’re connected to, his doctor, or a genetic counselor.

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It might be an uncomfortable conversation, but I have to tell him if it could help him decide to get tested. I’ll talk to him one-on-one this weekend.
I'm not sure, but we can make an appointment with a genetic counselor to find out.

If I have the BRCA2 inherited mutation, does that change my risk for other cancers?

How do you even find a genetic counselor?

Male breast cancer forms in the breast tissue of men. Though breast cancer is thought of as a women's disease, it does occur in men. Some men inherit mutated genes from their parents that increase the risk of breast cancer. Mutations in one of several genes, especially--

Your doctor may be able to give you a referral. Or your health insurance company may be able to find a medical geneticist or genetic counselor that's in your plan. For example, the National Society of Genetic Counselors (NSGC) and the American Board of Genetic Counseling (ABGC) offer online searchable directories of genetic counselors. The American College of Medical Genetics and Genomics (ACMG) has a searchable database of medical genetics clinic services. We could also make an appointment with the counselor I saw. She even does telehealth.

--BRCA2, would put you at greater risk of developing breast and prostate cancers. Because you have a strong family history of cancer, your doctor did recommend you consider genetic testing.

I would like to get tested. Not only for myself, but I have a daughter I'm concerned about too. I'll talk to my wife tonight.
Aiko, your genetic test results are positive for a BRCA mutation.

How does that affect my treatment options?

Let’s talk about precision medicine. The idea is to treat you in a way that’s tailored to you and your specific situation instead of treating you the same as any other woman with metastatic breast cancer.

There are treatments developed and approved for specific subtypes of cancers, such as those dependent on estrogen or progesterone (ER- or PR-positive) or those that are HER2 positive.

Now let’s talk about biomarkers. A biomarker is any molecule in the human body that we can measure that helps us to either predict whether a certain treatment might work for a cancer (predictive), or it might be prognostic for how likely it is that a cancer may recur, or it may be able to measure how well a therapy is working. Biomarker tests look at blood, tumor, or other tissue samples for changes or abnormalities caused by cancer. Testing helps your team understand the disease, what treatment would be appropriate, what you are at risk for, and your response to treatment.
Genetic testing can help you understand your risk for cancer, help you make medical decisions, and take action to lower your cancer risk or detect cancer early.
Glossary

**Biomarker Testing** looks for chemicals within the body that can help to diagnose and even track some types of cancers. The term “Biomarker Testing” can include “Genomic Testing.”

**BRCA1** is a gene that normally acts to prevent the growth of cells in the breast but when mutated, it predisposes a person to breast cancer.

**BRCA2** is a gene that gives directions for creating a protein that acts as a tumor suppressor, but when mutated, it predisposes a person to breast cancer.

**DNA** is a long molecule that contains our distinctive genetic code. DNA carries the instructions for creating all the proteins in our bodies.

A **gene** carries the information that determines your features or characteristics inherited from your parents.

**Genetic Counseling** provides information about genetic conditions and how they might affect you or your family.

**Genetic Counselors** are healthcare professionals who specialize in medical genetics and counseling. Genetic counselors can be part of your healthcare team, providing risk assessment, information, and supportive counseling to people and their families at risk for, or diagnosed with, an inherited condition.

**Genetic Testing** looks at DNA found in blood or saliva to find mutations that can cause diseases such as cancer. Genetic testing can help you understand your risk for cancer, help you make medical decisions, and take action to lower your cancer risk or detect cancer early. If you’ve already been diagnosed with cancer, genetic testing can help you make medical decisions about treatment.

**Genomic Testing** examines cancerous tissue to help provide information on how the tumor is likely to behave.

**Germline Mutation** is another name for an inherited mutation linked to a disease like cancer. Examples of germline mutations include BRCA1, BRCA2, PALB2, ATM, and others.

A **molecule** is the smallest particle of a substance that keeps all the properties of the substance and is composed of one or more atoms.

A **mutation** is a change in a DNA sequence.

A **pathology report** is a document that contains the diagnosis determined by observing cells and tissues under a microscope.
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Call our toll free helpline numbers: 844-ASK-SHARE (844-275-7427) in English 800-314-6948 en Espanol

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