

Genetic Testing



When you or a close family member has cancer, it is natural to ask “why” or “how” this happened. You may wonder if cancer runs in your family and ask how you can determine if it does or not. A test, called genetic testing can help answer this question.

Genetic testing is voluntary. Because testing has benefits as well as limitations, the decision about whether to be tested is a personal one.

This booklet is designed to explain what genetic testing is and when it might be useful. We hope that it will help you learn more about your options and, if needed, talk with your doctor about possible next steps.

Changes in Your Genes

GENES AND GENE MUTATIONS

Genes play an important role in who we are. They affect the way our cells and bodies work. Genes are found inside of cells and made of DNA. Some genes are passed down through families.

Genes can:

- Carry traits – like eye color or artistic ability – from parent to biological child.
- Control how our body makes substances called proteins.
- Tell cells what to do including when to grow and divide, or when to stop growing and dividing.

Genes also hold clues to our health. In cancer, the genes and proteins that make up one person's tumor can be very different from the genes and protein of another person's tumor.

Genes can change. A change in a gene is a mutation. Some mutations can lead to cancer.

There are two types of mutations:

1. Ones that happen in our lifetime (acquired mutations).
2. Ones that are passed down through families (inherited mutations).

This booklet focuses on genetic testing for mutations that are passed down through families. They are inherited mutations. When you hear people talk about “cancer genes,” they are most likely talking about inherited

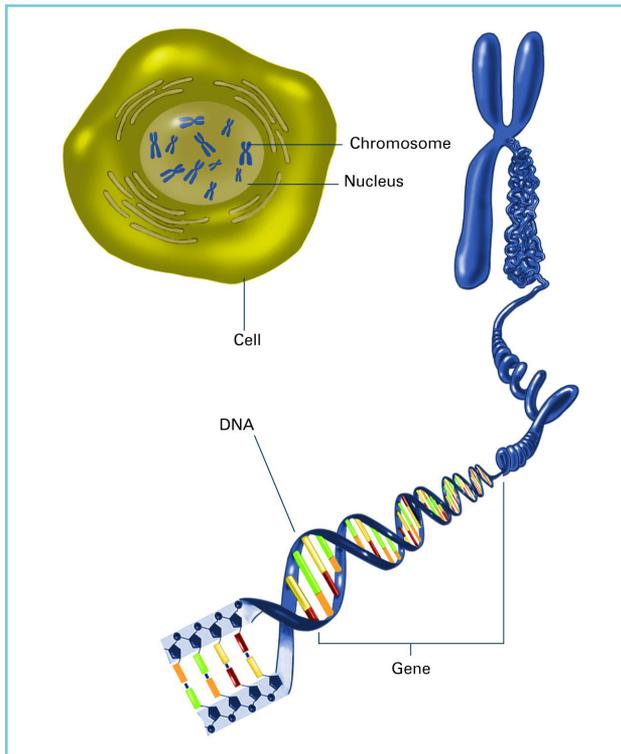
mutations. Read on to learn more about the role of inherited gene mutations in causing cancer and how and when to get tested.

INHERITED MUTATIONS

A genetic mutation is a change in a gene. Mutations in your genes can be inherited from your parents and passed on to your biological children. Some inherited mutations increase the risk of getting cancer. Your genes can be tested for these mutations.

Before we talk about testing, let's review a little about genes and how they are passed from parent to child.

- Our body is made up of cells. Every cell has a nucleus. The nucleus is the center of the cell.
- The nucleus contains 23 pairs of chromosomes. One chromosome in each pair comes from your biological father. The other comes from your biological mother.
- Each chromosome is made up of thousands of genes. Genes are pieces of DNA. They contain codes that make us who we are.
- The DNA inside of a gene is in a certain order or sequence. A change to the sequence can cause the gene to stop functioning normally. These changes can be inherited. They are called genetic mutations.



If your parent has a genetic mutation, it does not mean you will definitely have it too. Likewise, if you have a genetic mutation, it doesn't mean your child will definitely inherit it from you. The odds in each case are 50/50. Here's why:

- Each person has two copies of every gene: one from dad (who has two copies of the gene), and one from mom (who has two copies of the gene). Each parent passes on one of the two they have. A parent who has a genetic mutation may pass on either the mutated gene or the normal gene. The child's chance of getting either the mutated version or the normal one is 50/50—like the flip of a coin.
- If the parents have a second biological child, that child will also have a 50/50 chance of inheriting the genetic mutation.

- The parent is passing on a mutation, not cancer itself. The mutation increases the risk of getting cancer. This means that: If you inherit a mutation that increases cancer risk, it does not mean you are destined to get cancer. It means your risk is higher than average. If you know that you have a higher risk due to a mutation, you can talk with your doctor about your options. Screenings that are likely to find cancer earlier may be recommended. This is why genetic testing is important. Genetic testing reviewed with a health care professional can tell you if you have inherited a mutation.

KEY FACTS ABOUT "CANCER GENES"

- Most cancers are not inherited. Only 5% to 10% of cancers are linked to genetic mutations passed down from a biological parent.
- Some people who have mutations that increase risk of cancer never get cancer.
- A parent can pass on the genetic mutation (and increased risk) even if they never develop cancer themselves.
- A parent with cancer who has a mutation that increases risk of cancer may not pass it on to their biological children (50/50 chance).
- A child who does not inherit the mutation cannot pass it on.

Genetic Testing

WHAT IS GENETIC TESTING?

Genetic testing can help you learn more about your cancer risk and look for changes (mutations) in your genes. The results might tell you more about your chance of getting certain cancers in your lifetime.

Genetic testing may be offered or recommended either because:

- **You have cancer.** If you have cancer, your doctor may test to see if you inherited a mutation that increased your risk of developing this cancer. This may be called “genetic testing for an inherited mutation.”
- **Your family history suggests that you may be at high risk of developing cancer.** If you do not have cancer, a doctor or genetic health care professional may recommend testing to see if you inherited a mutation that increases your risk of getting cancer. This may be called “genetic testing for inherited cancer risk.”

If you have cancer, genetic testing may be useful in two ways:

- It can help determine the best way to treat the cancer. Some treatments work better for people with or without certain genetic mutations. In this way, genetic testing can be a critical part of treating cancer with precision medicine.
- It can let you know if family members may be at risk of inheriting the mutation and developing cancer.

If you do not have cancer, genetic testing may be used to:

- Learn more about your risk.
- Used to help make decisions on disease prevention.
- Inform cancer screening recommendations.
- Help you to make more informed decisions on your lifestyle choices. The results can affect the health care you receive and maybe even the lifestyle choices you make.

Genetic testing is also called germline testing or testing for hereditary cancer. It is different than biomarker testing of the cancer genome.

CANCER TYPES

Genetic tests are available for a number of cancer types. The list below outlines the more common cancer types tested:

- Breast cancer
- Colon cancer
- Endometrial or uterine cancer
- Kidney cancer
- Melanoma
- Ovarian cancer
- Thyroid cancer
- Pancreatic cancer
- Prostate cancer
- Sarcoma
- Stomach cancer

TESTING FOR INHERITED CANCERS

Genetic testing for inherited cancer risk is a useful tool to help you learn more about your own risk of cancer or the risk to your biological children or other family members. If you have cancer, the results may affect your treatment options. It's helpful to know when and how to get tested.

When to Get Tested

Learning that you have cancer, or that a close relative does, may make you want to rush to order a genetic test online. You may hear these tests advertised or discussed. Genetic testing is not always recommended. When it is, it is best done by a professional. There are a few reasons for this:

- The genetic tests you order online are not individualized to your personal and family history and may not include the right genes or mutations. You will not get complete information and may miss out on learning what you need to know.
- Online tests do not come with personal attention from a genetic counselor who can talk with you about the results and what they may mean for you and your family.
- At-home genetic tests are unlikely to be covered by insurance.

If you have already used an at-home genetic test you should take your results to a genetics professional that can help interpret the results and determine next steps or if additional testing is needed.

SHOULD I CONSIDER GENETIC TESTING?

To see if testing might be a good next step for you, answer these questions.

- Do more than three biological relatives on the same side of your family have the same or related cancers?
- Do you have cancer AND have two or more relatives who have had cancer at an early age?
- Do you have a relative with two or more types of cancer?
- Do you or a close biological relative have a rare cancer that is known to run in families?
- Do you have ovarian cancer? (Every person with ovarian cancer should consider genetic testing.)

If you answered yes to any of these questions, ask your health care provider about genetic testing.

How to Get Tested

The best way to get tested is working with a genetics professional. This person is trained in genetics and testing. They also know how to explain genetics to people who are new to the topic. If you have already seen a genetics professional, you can be confident that the right tests are ordered and that the results are read correctly.

You can make an appointment to speak with a genetics professional directly or ask your provider to refer you to one. A genetics professional will talk with you about your family's history. They can help you decide whether testing is right for you. If it is, they will order the recommended tests.

What to Expect

Genetic testing is done on a sample of blood or saliva (spit). The sample is sent to a special lab. The lab can detect DNA changes that suggest a mutation.

Testing provides a positive/negative result. You can find out if you carry a genetic mutation that increases the risk for cancer. It also provides information that you can use to decrease your risk of getting cancer. In most cases, the person who has cancer is the person who gets tested. Depending on the results of the test, other family members may be tested. Genetic tests do not always give a simple yes or no, which is another reason why it is important to see a genetics professional.

Keep in mind:

- If you have cancer, testing may provide information that will help you and help other family members.
- Testing is usually not recommended for children until they are old enough for screenings or procedures to prevent cancer. This is true even when their parent tests positive for an inherited mutation. If you think your children may have a higher risk of cancer due to family history, talk with a doctor. You can also talk with a genetics professional. Share your concerns and ask if or when it makes sense for them to get tested.

The results of genetic testing may help you:

- Know the age when to start cancer screening.
- Know how often to get screened.
- Get screenings that are available only for those at increased risk.
- Understand and learn how to monitor your body for signs and symptoms.
- Learn more about preventing cancer.
- Understand what testing tells you and what it doesn't tell you.
- Find out what your test results mean for other family members.

Inherited Cancer Syndromes

Doctors describe mutations that are passed down through families as syndromes. Genetic testing looks for these common syndromes. Other, much rarer, syndromes may be tested for as well.

SYNDROME NAME	GENE INVOLVED	INCREASES RISK OF
Cowden Syndrome	PTEN	Breast, Colon, Endometrial (Uterine), Kidney, and Thyroid cancers and Melanoma
Familial Adenomatous Polyposis	APC	Colorectal, Soft Tissue, and Brain cancers
Hereditary nonpolyposis colorectal cancer (HNPCC) <i>Also called: Lynch Syndrome</i>	DNA mismatch repair genes MLH1, MSH2, MSH6 or PMS2	Colorectal, Bile Duct, Brain, Breast, Endometrial (Uterine), Kidney, Liver, Ovarian, Pancreas, Prostate, Small Intestine, Skin, and Stomach, and Urinary Tract cancers
Hereditary Breast and Ovarian Cancer Syndrome <i>Also called: BRCA</i>	BRCA1, BRCA2	Breast, Ovarian (including Fallopian Tubes), Pancreas and Prostate cancers
Li-Fraumeni Syndrome	TP53	Soft Tissue Sarcomas (tumors in fat, muscles, nerves, joints, blood vessels, bones, or deep skin), Leukemia, Adrenal Gland, Brain, Breast, and Lung cancer
Multiple Endocrine Neoplasia (MEN)	Two types: <ul style="list-style-type: none"> ■ MEN1 is caused by a mutation in the MEN1 gene ■ MEN2A and MEN2B are caused by mutations in the RET gene. 	Cancers of the endocrine system
Von Hippel-Lindau Disease	VHL gene	Kidney, Pancreas, and less often Adrenal Gland cancers

What to Expect When Going Through Genetic Testing

Genetic testing can be an emotional experience. It may bring fear, stress, sadness, or relief. It can be scary to learn that your risk of developing cancer is higher than average. But when you know you have a mutation, you may be able to take steps to reduce your risk. Through testing, you may learn information that helps your family. The first step is to meet with a genetic counselor.

MEETING WITH A GENETICS PROFESSIONAL FOR GENETIC COUNSELING

A genetics professional is trained in both genetics and counseling. Most counselors have a master's degree in genetic counseling. They are certified through the American Board of Genetic Counseling. They can also be advanced practice providers with specialized training. They often work at large health systems, university hospitals, or cancer centers. They are trained to:

- Assess your risk based on family history and determine if genetic testing is appropriate.
- Explain the testing process.
- Interpret test results.
- Provide support as you cope with the emotional, psychological, and social implications of the results.

- Help you figure out how to talk with family members about the results of tests and what they might mean for them.
- Understand and help find resources to cover the costs of testing.
- Provide an individualized cancer risk management plan for you.

When you meet with a genetics professional, they will:

- Review your medical history and cancer screening history.
- Review of your family's cancer history, including the relatives who had cancer or related conditions, their biological relationship to you, and the ages at which they were diagnosed. You may be asked to provide the history of 3 generations of your family (example – your parents, grandparents and great-grandparents).
- Discuss the possibility of inherited risk.
- Explain the benefits and limitations of testing.
- Tell you about current privacy laws around genetic testing.
- Discuss financial considerations.
- Review the test results (if testing is recommended and after results are in).

Before you can meet with a genetics professional, you will need to find one. Your oncologist's office or cancer center may have a counselor on staff. If they do not, they may be able to refer you to one. If that is not an option, try searching online through the National Society of Genetic Counselors or the American College of Genetics and Genomics. Refer to the resources section at the end of this booklet for more information.

EMOTIONAL JOURNEY

Genetic testing can affect everyone involved in different ways. It can bring a wide range of emotions. The process itself is emotional. The waiting alone may cause anxiety. It can be stressful or hard to ask for testing when it is not offered.

People respond in many ways to the results of genetic tests. It is not unusual to feel:

- **Guilt** – Guilt is a common feeling among people who get tested. Those who test positive may feel guilty about possibly passing on the risk. People who test negative sometimes feel guilt too. Feelings of “why them and not me” can come up if other family members have a mutation.
- **Worry** – A positive test result may lead to worry or fear. You may feel this for yourself or other family members.
- **False hope** – A negative result may make you feel safe. People sometimes feel false hope that they will never get cancer. In reality, cancer can affect anyone, and many factors affect our risk.

- **Fear of discrimination or loss of privacy** – People sometimes worry about how the results of genetic tests will be used. They fear discrimination in health insurance, work, or other places. Fortunately, there are laws to protect us. The Genetic Information Nondiscrimination Act (GINA) was passed in 2008. It provides protection from employment and health insurance discrimination due to genetics. It is illegal to make hiring or insurance decisions based on genetic information. It is also illegal to use the results of your family members' genetic tests for this purpose. If you feel that you are being discriminated against based on genetics, seek help from a lawyer or advocacy organization.
- **Tension within a family** – Families do not always agree on whether to get tested or to share information about testing within the family. Seek support as you have these conversations.

These feelings are real and may not go away on their own. As you look into testing, seek support. Your genetics professional can help you navigate this emotional journey.

CSC's Emotional & Financial Resources

CANCER SUPPORT HELPLINE

The **Cancer Support Helpline** is here for you. Calling the Cancer Support Helpline can offer guidance on not only emotional support, but also guidance on the cost of care and useful resources. When you call 1-888-793-9355, you reach trained staff who can help you find:

- Local and national resources, including support groups, transportation services, financial assistance, and other programs.
- Short-term cancer coping assistance.
- Short-term housing resources.
- Treatment decision planning.
- Information on the cost of cancer and its treatment.
- Information on clinical trials.
- Information on genetic testing and biomarker testing.
- Access to an online distress screening program, CancerSupportSource®.
- General information about the Cancer Support Community.
- The names of other programs and services to help answer your questions.

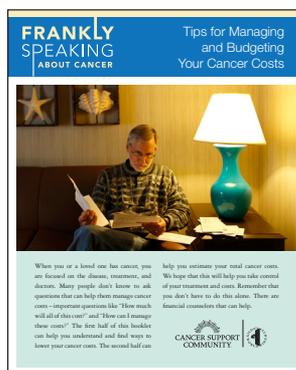
Remember, you are not alone. There are people and groups that can help.

OPEN TO OPTIONS



If you are facing a cancer treatment decision, Open to Options® is a research-proven program that can help you prepare a list of questions to share with your doctor. In less than an hour, our Open to Options specialists can help you create a written list of specific questions about your concerns for your doctor. Call 888-793-9355 to schedule an appointment.

MANAGING COSTS



The cost of genetic testing varies. In some cases, the cost may be covered by insurance. Whether it is covered will depend on the reason for testing, the test being done,

and the insurance policy. When costs are reasonable, people sometime pay out of pocket for genetic testing. If you cannot afford the cost of the test, assistance may be available. Talk with the genetic counselor about the cost and how to pay for it. Refer to CSC's *Frankly Speaking About Cancer: Tips for Managing and Budgeting Your Cancer Costs* for more information.

Genetic Testing Information & Support

Cancer Support Community

888-793-9355

www.CancerSupportCommunity.org

Abramson Cancer Center

Telegenetics Program

800-789-7366

www.pennmedicine.org/cancer/navigating-cancer-care/programs-and-centers/telegenetics-program

American Cancer Society

800-227-2345

www.cancer.org

American College of Genetics and Genomics - Find a Genetic Clinic

clinics.acmg.net

Breastcancer.org

610-642-6550

www.breastcancer.org/risk/factors/genetics

CancerCare

800-813-4673

www.cancercare.org

Cancer.net

888-651-3038

www.cancer.net

Centers for Disease Control Genetic Testing Information

www.cdc.gov/genomics/gtesting

Fight Colorectal Cancer

877.427.2111

www.FightColorectalCancer.org/biomarked

FORCE (Facing Our Risk of Cancer Empowered)

866-288-7475

<https://www.facingourrisk.org>

GO2 Foundation for Lung Cancer

800-298-2436

www.go2foundation.org

LUNGevity Foundation

321-407-6100

www.LUNGevity.org

National Cancer Institute (NCI)

800-422-6237

www.cancer.gov

NCI Genetic Testing Information

800-422-6237

www.cancer.gov/about-cancer/causes-prevention/genetics

Patient Advocate Foundation

800-532-5274

www.patientadvocate.org

Cancer Support Community Resources

Cancer Support Helpline® — Have questions, concerns or looking for resources? Call CSC's toll-free Cancer Support Helpline (888-793-9355), available in 200 languages Mon - Fri 9am - 9pm ET and Sat-Sun 9am – 5pm ET

Open to Options® — Need help making a cancer treatment decision? Our trained specialists can help you create a list of questions to share with your doctor. Make an appointment by calling 888-793-9355 or by contacting your local CSC or Gilda's Club

Frankly Speaking About Cancer® — Trusted information for cancer patients and their loved ones is available through publications, online, and in-person programs

Services at Local CSCs and Gilda's Clubs — With the help of 170 locations, CSC and Gilda's Club affiliates provide services free of charge to people touched by cancer. Attend support groups, educational sessions, wellness programs, and more at a location near you. www.CancerSupportCommunity.org/FindLocation

Cancer Experience Registry® — Help others by sharing your cancer patient or cancer caregiver experience via survey at www.CancerExperienceRegistry.org

MyLifeLine — CSC's private, online community allows patients and caregivers to easily connect with friends and family to receive social, emotional, and practical support throughout the cancer journey and beyond. Sign up at www.MyLifeLine.org

Grassroots Network — Make sure your voice is heard by federal and state policy makers on issues affecting cancer patients and survivors by joining our Network at www.CancerSupportCommunity.org/become-advocate

FRANKLY SPEAKING ABOUT CANCER: GENETIC TESTING PROGRAM PARTNERS

Association of Community Cancer Centers, CancerCare, Colorectal Cancer Alliance, LUNgevity, Melanoma Research Alliance, Oncology Nurses Society, Ovarian Cancer Research Alliance

THIS PROGRAM WAS MADE POSSIBLE WITH GENEROUS SUPPORT FROM:



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Lilly Oncology

NeoGenomics

For more information and resources, please visit the website at www.CancerSupportCommunity.org. For print copies of this booklet or other information, visit Orders.CancerSupportCommunity.org or call 888-793-9355.

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