Genetic Testing for Cancer: What You Need to Know

Here is some basic information about genetic testing and how it's used to learn about inherited cancer risk. It will help you understand what genetic testing is, but it won't give you specific information or advice on genetic testing for any one disease. A doctor or a genetic counselor who knows about the diseases in your family is better able to talk with you about your risks and what you might learn from genetic testing.

Genes, mutations, and cancer risk

Genetics is the field of science that looks at how traits (such as eye color) are passed down from parents to their children through genes.

Genes are pieces of DNA (deoxyribonucleic acid) inside each of our cells that instruct them how to make the proteins the body needs to function. DNA is the genetic "blueprint" found in each cell. Genes affect inherited traits passed on from a parent to a child, such as hair color, eye color, and height. They also affect whether a person is likely to develop certain diseases, such as cancer.

In humans, genes are located on 23 pairs of long strands of DNA called *chromosomes*. One of each chromosome pair comes from the mother, and the other from the father. Each chromosome can contain hundreds or thousands of genes that are passed from the parents to the child. The genes you were born with are in every cell in your body.

Changes to these genes, called *mutations*, play an important role in the development of cancer. Mutations can cause a cell to make (or not make) proteins that affect how it grows and divides into new cells. Certain mutations can cause cells to grow out of control, which can lead to cancer. Usually several gene changes are needed before a cell becomes cancer. To learn more about different types of cancer-related genes, please see our document called *Genes and Cancer*.

Some gene changes that lead to cancer may be inherited from a parent, but most are not. Only about 5% to 10% of all cancers are thought to be related to an inherited gene change that strongly affects a person's risk for a certain type of cancer.

Most cancers start because of gene mutations that happen sometime during a person's lifetime. Sometimes these gene changes have an outside cause, such as exposure to sunlight or tobacco. But gene changes can also just be random events that sometimes happen inside a cell, without an obvious cause.

These types of *acquired* (as opposed to inherited) mutations only affect the cells that grow from the mutated cell. They do not affect all the cells in the person's body. This means all the cancer cells will have the mutations, but the normal, non-cancerous cells of the body will not have them. This is very different from inherited mutations, which are in every cell in the body – even the cells without cancer.

What is genetic testing?

Genetic testing is the process of using medical tests to look for changes (mutations) in a person's genes or chromosomes. Hundreds of different genetic tests are used today, and more are being developed.

Genetic testing for diseases that can be inherited

Genetic testing can be used in different situations. The type of testing most often used to check for cancer risk is called **predictive gene testing**. It's used to look for gene mutations that might put a person at risk of getting a disease. It's usually done in families with a history that suggests there's a disease that may be inherited. An example is testing for changes in the BRCA1 and BRCA2 genes (known breast cancer genes) in a woman whose mother and sister had breast cancer.

Genetic testing is also used for other reasons:

- **Carrier testing** can be used to help couples learn if they carry a gene mutation for a disorder they might pass on to a child, such as cystic fibrosis, sickle-cell anemia, or Tay-Sachs disease.
- **Prenatal screening** can be used to diagnose some conditions in babies before they are even born, such as Down syndrome.
- **Newborn screening** is the most widespread form of genetic testing. Newborns are screened for a number of inherited conditions such as phenylketonuria (PKU), cystic fibrosis, sickle cell disease, and others. The tests required vary from state to state.

All of these forms of genetic testing, including predictive gene testing, look for gene changes that are passed from one generation to the next and are found in every cell in the body. Except for the newborn screening tests, they are used mainly for people with certain types of disease that seem to run in their families. They are not needed by most people.

Genetic testing for cancer risk

Cancer-related genetic tests are most commonly done as predictive genetic tests. They may be used:

- To see if a person has a certain gene mutation known to increase the risk for a certain cancer (or cancers)
- To confirm a suspected gene mutation in a person or family

Testing cancer cells for genetic changes

Sometimes after a person has been diagnosed with cancer, the doctor will order tests to look for gene changes in a sample of the cancer cells. These tests can give information on a person's outlook (prognosis) and can sometimes help tell whether certain types of treatment might be useful.

These types of tests look for gene changes **only** in the cancer cells that are taken from the patient. These tests are not the same as the tests used to find out about inherited cancer risk.

This document does not cover gene testing done on cancer cells. For more about this kind of testing and its use in cancer treatment, see our information on specific types of cancer.

The rest of this document focuses on predictive genetic testing for inherited mutations as they relate to cancer.

Who should have genetic testing?

Genetic testing to learn about your cancer risk is done only if you decide to have it. If you are concerned about your family's pattern of disease, cancer you've had in the past, or other cancer risk factors, you may want to talk to your doctor about whether to have genetic testing.

Doctors will sometimes suggest patients have genetic testing if others in their family have had a certain disease or certain patterns of disease. If you have any of the following, you might consider genetic testing for yourself:

- Several first-degree relatives (mother, father, sisters, brothers, children) with cancer, especially if they've had the same type of cancer
- Cancers in your family that are sometimes linked to a single gene mutation (for instance, breast, ovarian, and pancreatic cancer).
- Family members who had cancer at a younger age than normal for that type of cancer
- Close relatives with rare cancers that are linked to inherited cancer syndromes
- A physical finding that is linked to an inherited cancer (such as having many colon polyps)
- A known genetic mutation in one or more family members who have already had genetic testing

You need to know what tests may be available. For some types of cancer, no known mutations have been linked to an increased risk. Other cancer types may have known mutations, but genetic tests are not yet available for them. For more information on the types of cancers for which inherited genes may be important, please see our document *Family Cancer Syndromes*.

It's important to find out how useful testing may be for you **before** you do it. Meeting with a genetic counselor before the actual test can help you know what to expect. The counselor can tell you about the pros and cons of the test, what the results might mean, and what your options would be at that point. You will want to think about how the results might affect you and your relatives, and discuss these issues with the counselor before your test.

After the test is done, your genetic counselor will share the results with you. Some people choose to bring a family member to help share results with other family members. Although testing does not always give you clear answers, genetic counselors are trained to interpret and explain the test results and what they might mean to you and your family. If a mutation is found, the counselor will talk to you about which of your family members might also be affected. It will be important for those family members to know exactly which mutation was found and in which gene. They can then discuss this information with their doctors and consider being tested, too.

What are the benefits of genetic testing?

The obvious benefit of genetic testing is the chance for a better understanding of your risk for a certain disease. Testing is not perfect, but it can often help you make decisions about your health.

A negative result on a genetic test in families at risk for a specific cancer may help relieve anxiety or uncertainty. In the same way, a positive result can help you make important decisions about your future, perhaps including things you can do to help lower your risk. A positive result may also lead to finding disease earlier, when treatment is more likely to be helpful.

For people already diagnosed with cancer, genetic testing of the tumor can often help determine the prognosis (outlook), and can sometimes even help in deciding which treatments will most likely work.

As scientists learn more about the genes that contribute to cancer, genetic testing will probably become more helpful in learning about a person’s cancer risk.

What are the drawbacks of genetic testing?

Genetic tests have several potential problems you should think about before being tested:

Limited answers

Genetic tests do not give precise answers about inherited diseases, especially about breast and colon cancer. A positive test result does not always mean you will get the disease. The test can tell what *might* happen, but it cannot tell what *will* happen. On the other hand, a negative result does not mean you have no risk of getting the disease.

As with many medical tests, genetic tests may be flawed, or test results may be read wrong. Though many steps are taken to prevent this, genetic testing is not tightly regulated, and different labs may have different ways of looking for a certain mutation.

Sometimes a genetic test may be done even though the result has little chance of helping the person. When this happens, the test may create anxiety when the original intent was to relieve it. This is why counseling before you get the test is so important – you’ll want to know exactly how the result could help you, and if it would help enough to make up for the stress it can cause.

Psychological or emotional impact

Many people are anxious even before they get their test results. They may think about how the result might affect them and their families, and how they can talk about and manage the information.

Learning that you have or might develop a serious disease can be frightening. The person being tested may find it even more upsetting if family members have already died of the disease in question.

A positive genetic test result can also affect other family members. More family members may need to be tested. Sometimes family secrets are revealed as a result – paternity, adoptions, or other difficult issues may come up.

Having a gene or passing a gene on to children can bring out feelings of guilt or anger. The test results might also affect future relationships with a spouse or other family members, which can be stressful, too.

In some cases, more medical tests or procedures may have to be done as a result of genetic testing. For example, if a gene mutation for colorectal cancer is found, more tests like colonoscopy may be recommended. These extra tests can also be sources of stress and anxiety.

Privacy issues

Most people who ask about the privacy of genetic information are concerned about how the information may be used in ways that can harm them. Most Americans are afraid that employers and insurance companies might get and use their genetic information. They fear an employer could find out and discriminate by not hiring or promoting them, but the Genetic Information Nondiscrimination Act (GINA) law forbids most employers from doing this (see below). Some people are even concerned that adoption efforts could be stopped based on this type of sensitive information.

On the other side, some companies and researchers are concerned about there being too many restrictions on this information, which could be useful to scientists.

The privacy issue becomes even more complicated when many family members may be affected by a single positive genetic test result.

For these reasons, it’s important to think about who might learn about your results and with whom you will want to share your results before you decide on testing.

National law prohibits discrimination by employers and health insurers

The Genetic Information Nondiscrimination Act (GINA) bars discrimination based on genetic information by employers with more than 15 employees and health insurers. GINA defines genetic information as:

- A person’s own genetic test results
- The genetic test results of family members
- One or more family members known to have a genetic disease or disorder

Employers: GINA bars employers from discriminating on the basis of genetic information in hiring, termination (firing or layoffs), pay, or other personnel actions such as promotions, classifications, or assignments. The law applies no matter how they got the information.

Employers are not allowed to require genetic testing and can't collect genetic information except for very limited exceptions. For instance, it may be allowed when information is needed to meet the requirements of family and medical leave laws or to watch for harmful effects from hazardous workplace exposures.

Employers must keep genetic information confidential. They can't release or share genetic information except when:

- The employee asks them to
- Fulfilling a request from a health researcher
- Complying with medical leave law
- Disclosing or reporting to a public health agency

Health insurers: GINA bars health insurers (including group health plans, individual plans, and Medicare supplemental plans) from turning down people or charging higher premiums for health insurance based on genetic information or the use of genetic services. This includes genetic counseling and testing. The law also bars these insurers from asking for or requiring genetic tests. GINA applies to all health insurance plans (including federally regulated ERISA plans, state-regulated plans, and private individual plans).

A few states have stronger laws than GINA. GINA does not replace state laws against genetic discrimination that are broader in scope. Rather, GINA establishes a national "floor" of protection while allowing states to impose stronger protection for patients.

GINA's protections do not extend to life insurance, disability insurance and long-term care insurance. It also doesn't require health insurance to cover genetic testing. GINA does not apply to very small employers (with fewer than 15 employees), nor does it apply to military health plans, the Veterans Administration, or the Indian Health Service. GINA does not apply to federal employees who get health coverage through the Federal Employees Health Benefits Plans.

Genetic testing ads: promises that don't always deliver

The social, legal, and ethical challenges posed by genetic information are further complicated by the way genetic tests are sometimes marketed. Manufacturers now advertise and promote genetic testing to doctors and to the public. Sometimes they make the test sound much more helpful and decisive than it has actually proven to be. This can be harmful because decisions about testing may be made based on incomplete information, or even on the basis of misleading or wrong information. A lot of tests don't give the answers they seem to promise. Reputable genetics counselors (see the section below, "Genetic counseling") can help you know what to expect from your test results.

Who pays for genetic testing?

Genetic testing is complicated, and it can cost a lot. Some tests cost more than others, but the final bill can be thousands of dollars. Be sure you have an idea of how much it will cost you before you have testing done.

If you have health insurance, you must decide if you're going to ask your insurance company to help pay for testing. For the most part, state and federal laws do not require insurance companies to pay for predictive testing. Some companies may cover the testing, while others may not.

The new federal health care law says that genetic testing should be covered for some women at higher risk for getting breast and/or ovarian cancer. The amount of coverage depends on your health plan, so you have to find out what's included in your plan.

What happens during genetic testing?

Genetic testing is typically done only if you and your health care team feel sure that it's the best thing for you and your family. Usually there are several steps before the actual testing is completed. (Some genetic tests are sold as home-based kits that you can buy online. The process for these tests will vary depending on the company that offers them. For more information on these types of tests, see the section "What's the future of genetic testing?")

Risk assessment

The first step in genetic testing is to find out how likely you are to develop a certain disease. This risk is based on the pattern of disease in your family (called a *family history*) and other factors. Your risk may be looked at by a doctor, nurse, or trained genetic counselor.

You will be asked about your reasons for wanting genetic testing. Your family history (as far back as possible and up to the present day) will be reviewed in depth. The patterns of disease in your family will be identified. It's important to confirm the illnesses in your family by medical records and/or death certificates when possible. If the genetic testing is for a certain type of cancer, you may also be asked about your own medical history. This may include early detection practices, gynecologic history (for women), lifestyle factors, and exposure to things that can cause cancer (*carcinogens*). You may also have a physical exam that focuses on the cancer of concern in your family.

It's often helpful to look at your family's attitudes about cancer and the possibility of a family member being "blamed" for the cancer. Cultural beliefs, support systems, and finances may also play a role in how your family views cancer. These topics may be discussed, too.

Genetic counseling

Genetic counselors have special training and graduate degrees in their field. Some doctors and advanced practice oncology nurses with special training may also do genetic counseling.

The purpose of genetic counseling is to give you information in an unbiased or neutral way so that you or you and your family can make your own decisions about whether to get tested. Sometimes the need for testing is clear and a doctor will recommend testing and follow-up care. But even if

that's the case, you have the right to refuse testing. You may feel more comfortable making a decision after you talk with a genetic counselor. In fact, some health insurers require you to get genetic counseling before genetic testing will be covered.

The counselor will discuss how families inherit cancers and how genes are passed on to children, as well as the types of cancer seen in the family and estimates of the person's cancer risk. The pros and cons, cost, and limits of testing are discussed too, as well as who in the family should consider being tested. More than one family member may be offered testing.

For example, if testing shows that the person has a high cancer risk, the counselor may also talk about the best ways to manage it. These may include lifestyle changes, early detection, watching for signs and symptoms of cancer, medicines to reduce cancer risk, or even preventive surgery.

Many of these issues require an expert counselor. An increased risk of cancer, especially for children, and the potential for discrimination can be frightening. The counselor will explore ways to cope, as well as your specific fears and concerns. How to discuss the test results and what they mean with other family members is another key topic.

Your doctor can probably refer you to a genetic counselor in your area. If not, you can find a list of certified genetic counselors on the websites of the National Society of Genetic Counselors or the National Cancer Institute. (See the "To learn more" section at the end of this document for more information.)

Informed consent

After risk assessment and genetic counseling, if you decide to be tested you'll be asked to give your informed consent in writing. The process of informing you and your family about testing should cover:

- The purpose of the genetic test
- The reason for offering the test to you and/or other family members
- The type and nature of the genetic condition being tested for
- Test accuracy
- Pros and cons of testing (including the limits of what the results might tell you)
- Other testing options you could use
- Treatment options that might be available depending on the test results (if this applies to you)
- Further decisions that may need to be made once the results are back
- The possible consent to use the results for research purposes after the test
- Availability of counseling and support services
- Your right to refuse testing

For more information on this process, please see our document *Informed Consent*.

Specimen collection and lab testing

Once you have signed the consent form, lab tests are done on cells taken from your body. Genetic tests may be done on a sample of blood, cheek cells, urine, amniotic fluid (the fluid that surrounds a fetus during pregnancy), or other body tissues.

Genetic tests for cancer usually mean you will give several tubes of blood. A thin needle is put into a vein (usually in your arm) to get the blood.

Getting test results

Once the testing is complete, the lab reports the results in writing to the doctor or genetic counselor. You will then be given the results during another counseling session. This may not happen until several weeks after the samples are taken. The accuracy of the test and the meaning of the results will be discussed with you in detail.

If the result is positive

If the results of the test are *positive*, you have a mutated gene (or genes) that may place you at risk. Your risk of developing the cancer will be discussed in light of the fact that you have the mutation.

Many people are concerned or anxious after learning they are at increased risk for cancer. This is normal. The results may lead to strains in family relationships as well. Concern about being treated differently may become more real.

Even after the testing is complete there's often a great deal of uncertainty. In most cases, there's no way to know if the disease will even develop. And even if you were to know you will get the disease, you still wouldn't know when it might develop. Your doctor or counselor can help you sort through your options at this point, including what you might be able to do to help lower your risk.

If the result is negative

If the test result is *negative*, it means the gene mutation that was tested for is not present. You will probably feel relieved that the test did not show you are at increased risk.

But it's important to understand that genetic test results can't always guarantee you're not at increased risk. For example, there might still be a chance that you have a mutation that you were not tested for. (Many genes can have more than one kind of mutation that can result in higher cancer risk.) And rarely, the test result may be a "*false negative*." This means the test reads negative but the mutation is actually there. But even a result that's truly negative does not mean your risk is zero – it just means you have the same average risk as most other people.

Family members with negative test results may feel guilty if other family members test positive. Your doctor or genetic counselor can help you understand what the test results mean for you and your family and how to deal with them.

If the result is inconclusive

If there's not enough information to know whether the genetic changes are present, the test result is said to be *inconclusive*. The person (and family) may still be at a higher risk of developing cancer. Taking steps to lower your risk and have some control over your health can be helpful for some people, but not having a sure result can still cause anxiety and frustration. Your doctor or genetic counselor can help you understand what the results might mean for you and your family and help you cope with them.

If the result is variance of unknown or uncertain significance

This result can happen when genetic tests show that a person has an unusual form of a gene, but its meaning is unclear. It can be a normal variant, simply a different version of gene that isn't seen often enough to be sure, or there may be some other explanation. For some, a *variance of unknown or uncertain significance* (VUS) result can cause anxiety, frustration, and even anger because this result gives no information to guide future decisions. Your doctor or genetic counselor can help you understand what the results might mean for you and your family and help you cope with them.

What if genetic testing shows an increased cancer risk?

If your genetic test result is positive or inconclusive for a gene mutation that could increase your risk of cancer, managing the risk should become a priority.

Some of the ways you might lower your risk include taking medicines (called *chemoprevention*), having preventive (or *prophylactic*) surgery, and making lifestyle changes. Doing what you can to find cancer early (through testing and awareness of early signs and symptoms), when treatment is most likely to be successful, is also important for people at increased risk.

Your doctor may recommend one or more of these approaches, but it's important to understand how much they could affect your risk before you decide on a course of action. You will also want to be sure you understand their risks and downsides before deciding on a plan.

Chemoprevention

Chemoprevention is the use of medicines to prevent cells from developing into certain types of cancer. Several medicines are being studied and used to help lower the risk of certain cancers in people known to be at high risk. For example, the drugs tamoxifen and raloxifene can be used to reduce breast cancer risk. Each person's risk and medical situation must be considered carefully so that any harmful effects of the drugs do not outweigh the benefits. As we learn more about genetic diseases, we should find out more about using medicines to prevent cancer.

Prophylactic (preventive) surgery

Prophylactic (preventive) surgery is another option in some cases. For example, some women at high risk for ovarian cancer may decide to have their ovaries removed once they've had their children.

Changes in lifestyle factors

Ask your doctor if there are lifestyle changes you can make that could affect your risk, such as limiting alcohol intake and exercising to lower your risk of colon or breast cancer. In some cases the effect of these changes on risk might be small compared to the increased risk from the mutation, but you may still want to ask your doctor what you can do.

Cancer detection tests and awareness

Early detection tests may be started at an earlier age or be done more often, or special tests may be needed if you have a positive genetic test result. For instance, a person known to have an increased risk of colon cancer might need to start screening earlier than the usual age of 50. Likewise, a woman with a genetic mutation that raises her risk of breast cancer might need breast MRI scans along with her mammograms to look for early signs of the cancer.

It's also important to be aware of the possible signs and symptoms of cancers you are at higher risk for, and to see a doctor right away if anything concerns you. Finding and treating cancer early offers the best chance of having a good outcome.

Sharing results with family members

If you have a gene mutation that raises your risk, there may be other factors to think about, too, such as whether to tell other family members who might also be at increased risk. Telling them might help them decide if they should get tested or adopt some of the approaches to try to lower their risk.

On the other hand, some test results may cause more anxiety than anything else, and some family members may not want to know their own risk. This is especially true if there's not much they can do with the results. You may want to speak with family members before you get tested to find out if they want to know your results.

How else might genetic information be used?

Patients, families, and health care professionals are not the only ones interested in genetic information. Here are some of the other groups who might use this information:

Medical and pharmaceutical researchers

Medical and pharmaceutical researchers are interested in low-cost access to genetic information and materials. This is why the pharmaceutical industry (companies that make medicines) has opposed donor ownership of genetic material. If a person owns their genetic information, the pharmaceutical companies might have to pay the donor for access to the information. Members of the pharmaceutical lobby have argued against people owning their own genetic information, stating it would drive up drug costs, which would be passed on to the consumer.

Today, medical researchers must get the individual's informed consent before any studies of tissue samples and DNA can be done.

Insurers

Insurers also could benefit from access to genetic information.

In the past, health insurers could use this type of information when deciding who to cover and how much to charge for insurance. But this has changed. Federal law, based on the Genetic Information Nondiscrimination Act (GINA), no longer allows health insurers to base these decisions on the results of genetic information..

Note that GINA does not restrict use of genetic information for life insurance, disability insurance, or long-term care insurance. It also does not say that health insurers must pay for any particular genetic test or treatment.

Employers

Employers are allowed to ask for genetic testing when it is used to monitor exposure to potentially toxic chemicals and substances in the workplace. But discrimination and employment decisions based on genetic information are barred at the national level for most employers. GINA prohibits the use of genetic information in workplace employment decisions for non-governmental organizations. For more on GINA, see the section called “What are the benefits of genetic testing?”

What’s the future of genetic testing?

Without question, genetic testing will play a larger role in cancer risk assessment, detection, and treatment in the future.

The Human Genome Project and The Cancer Genome Atlas

The Human Genome Project was a huge undertaking in which government and private researchers mapped the entire sequence of human DNA (known as the *human genome*). The project took more than 10 years and was completed in 2003. Scientists now estimate that the 3 billion or so “letters” that make up the human genome contain about 25,000 genes. The next step is to identify these genes and learn what each one does. This basic genome map will allow researchers to identify cancer-related genes more quickly.

An even larger undertaking, The Cancer Genome Atlas (TCGA), is now under way. Researchers are working to map the genes of cancer cells to try to learn how those genes are changed to produce the cancer. The TCGA uses an integrated network of hundreds of researchers across the United States and Canada to collect tissue samples from people with certain types of cancer. This project will further our understanding of the genetic basis of cancer, making genetic testing more useful in the future.

Personalized treatment

There are some genetic tests – some available, and some still being developed – that may help a doctor choose a person’s medicines.

For example, some tests can show whether a person's cancer is likely to respond to a certain treatment based on its gene changes. Many new cancer medicines are being developed based on the known gene changes often found in cancer cells. In some cases, doctors can now test cancer cells for these gene changes to find out if the treatment is likely to be helpful.

Other tests can show whether a person's body can process certain medicines normally. Tamoxifen, for instance, is a drug that can be used to reduce the risk of breast cancer coming back. But the body must make a certain enzyme to activate the drug. Some studies suggest that tamoxifen may work better in women who inherited the genes that make more of the activating enzyme. Overall, though, studies have not yet clearly proven this link. They also have not shown that people who are tested have better treatment outcomes. Right now, most doctors give tamoxifen without genetic testing, but further studies may lead to different options in the future.

More is being learned all the time about the gene changes in cancer cells and about how people process drugs based on their genes. This is expected to give doctors useful information about choosing medicines in the future.

Better technology

At this time, genetic testing can cost a lot, and it can take several weeks to get the results. But as better technologies are developed, tests are becoming more accurate and are able to look at more than one gene at a time.

DNA chip technology is one example of this. These chips allow researchers to look for changes in many different genes at the same time and analyze which genes are active. In the future, this type of DNA testing may help doctors learn a person's risk of developing different diseases. This could make it easier to customize treatment for that person.

Other areas of research may impact how important genetic testing will become. One is the field known as *epigenetics*, which looks at ways genes are turned on and off other than just through mutations. Another is the field of *proteomics*, which looks at patterns of proteins in the body. (Genes are the blueprints for making proteins.) Researchers in these fields are developing tests that may someday be used along with or instead of standard genetic tests.

Home-based genetic test kits

Over the past few years, some genetic tests, marketed for a number of purposes, have become available as at-home kits that can be bought on the Internet. The kits include a small container to collect a saliva sample or a swab to rub inside the cheek. A few require blood, so the consumer must visit a medical facility to have it taken. Users mail the sample to a lab, where it's tested for genes related to cancer or other conditions.

These tests may give some people access to genetic testing who might not otherwise have it. But while many of the companies marketing these tests make some type of genetic counseling available, there are concerns about whether people will use it (especially before buying a test). The decision to have genetic testing is complicated, and without proper background information and counseling, people might not understand the full implications of getting tested. They also may not be able to interpret their test results correctly.

There are other concerns, too. According to the US Food and Drug Administration (FDA), which regulates the makers of genetic tests, some of these tests are not scientifically validated. Others

give results that are only useful if the person has a full medical evaluation. The FDA notes that genetic tests are very complex and they should be done in a specialized laboratory. They further recommend that the results be interpreted by a doctor or genetic counselor who understands the value of the test and how to best interpret and use the results.

If you are thinking about using an at-home testing kit, talk to your doctor or a genetic counselor first. This will help you decide if such testing is right for you and help prepare you for what you might learn. You should know the pros, cons, and limits of genetic testing before you have it done. And most people will need a trained professional to help work out exactly what the results mean for them and their families.

Dealing with genetic information

Managing the information we get from genetic testing continues to be an important issue. Many questions about patient rights and privacy still need to be resolved, and may become even more complex in the future. Other gray areas include genetic testing of children up for adoption, testing children for diseases they might develop as adults, and prenatal testing. Both national and international legislation are needed to address these concerns.

To learn more

More information from your American Cancer Society

We have some related information that may also be helpful to you. Free copies of these materials may be ordered from our toll-free number, 1-800-227-2345, or you can read them online at www.cancer.org.

Informed Consent (also in Spanish)

Family Cancer Syndromes

Genes and Cancer

National organizations and websites*

Along with the American Cancer Society, other sources of information and support include:

National Society of Genetic Counselors
Website: www.nsgc.org

> The “Consumer Information” link on the website offers detailed information on genetic counseling, questions to ask before genetic testing, a guide to collecting family history, info on genetic testing and genetic counselors, and a directory of genetic counselors.

National Cancer Institute
Toll-free number: 1-800-422-6237 (1-800-4-CANCER)
TTY: 1-800-332-8615
Website: www.cancer.gov

A listing of professionals who offer services related to cancer genetics (cancer risk assessment, genetic counseling, genetic susceptibility testing, and others) can be found at: www.cancer.gov/cancertopics/genetics/directory.

Hereditary Cancer Center (HCC)
Toll-free number: 1-800-648-8133
Website: http://medicine.creighton.edu/hcc

Maintains the Early Detection Registry Network (EDRN), a nationwide registry where any carrier of a cancer genetic mutation may register to be invited to participate in appropriate research studies for their specific genetic mutation.

** Inclusion on this list does not imply endorsement by the American Cancer Society.*

No matter who you are, we can help. Contact us anytime, day or night, for information and support. Call us at **1-800-227-2345** or visit www.cancer.org.

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