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## Precision or Personalized Medicine

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### What is precision medicine?

Precision medicine is a way health care providers can offer and plan specific care for their patients, based on the particular genes, proteins, and other substances in a person's body. This approach is also sometimes called *personalized medicine* or *personalized care*.

With regard to cancer, precision medicine most often means looking at how changes in certain genes or proteins in a person's cancer cells might affect their care, such as their treatment options. But it can have other uses as well.

In precision medicine, doctors use information from certain lab tests to put together a plan of care that usually includes specific recommendations. In some cases, it can help make a more accurate diagnosis and improve treatment. In other cases, it can help people make decisions about healthy habits, earlier [screening tests](#), and other steps they can take that might help lower their risk for a particular cancer.

Your health care providers might not use the exact words "precision medicine" or "personalized medicine." Instead, they might talk to you about genetic, genomic, DNA, or molecular testing. Or they might talk about checking for biomarkers or getting a genetic profile. These are ways doctors and other health care providers might use a precision medicine approach when they are planning your care.

## Gene changes and precision medicine

Precision medicine is largely based on knowing the effects of changes in certain genes (and proteins) inside cells.

**Genes** are pieces of DNA inside each cell. They tell the cell how to make the **proteins** it needs to function. Each gene contains the code (instructions) to make a certain protein, and each protein has a specific job in the cell.

When a cell divides to make new cells, the genes inside the cell are copied. A gene change (also known as a *variant* or *mutation*) happens when there's a mistake in the copying process. Sometimes these changes can come from our parents, who are the source of our DNA. (These are known as *inherited* gene changes.) But gene changes can also happen sometime later in life. (These are known as *acquired* gene changes.) Some gene changes can be harmful, while others may not cause any problems.

## Gene changes and cancer

All cancers are caused by gene changes of some kind. Cancer cells are abnormal versions of normal cells, meaning something changed in the genes of a normal cell to make it turn into a cancer cell. For example, genes that normally help keep cells from growing out of control might get turned off, or genes that normally help cells grow and divide might get turned on all the time.

It takes more than one gene change in a cell for cancer to happen. While we don't yet know all the genes and mutations that could be involved in the development of cancer, there are some we know about and can test for.

To learn more about how changes in genes can lead to cancer, see [Genes and Cancer](#).

In some cases, having an abnormal gene might make a person more likely to develop a certain cancer. Or, if they already have cancer, a specific gene change might mean that their prognosis (outlook) is better or worse than someone with the same cancer who doesn't have that gene change.

In some people with cancer, having a certain gene change in the cancer cells might mean that the cancer most likely will (or will not) respond well to a certain treatment. For example, when a person is diagnosed with cancer, they often get the same treatment as other people with the same type of cancer. But the gene changes in the cancer cells might be different from one person to another, even if they have the same type of cancer. Because of this, the cancers in these people might respond differently to the

same drug. For certain cancer types, doctors can now test for gene changes that can tell them if a person's cancer might respond better to one drug than another.

## Precision medicine in cancer care

Precision medicine is being used for certain cancers to help know what tests and treatment are best. Doctors might use precision medicine to help them:

- Identify people who might be at high risk for cancer, and help these people lower their risk
- Find certain cancers early
- Diagnose a specific type of cancer correctly
- Choose which cancer treatment options are best
- Evaluate how well a treatment is working

## Cancer risk and prevention

Sometimes precision medicine is used for people who are at higher risk for developing certain cancers. For example, a person might realize cancer runs in their family, or their doctor might notice a pattern of cancer in their family. In these cases, the person might meet with a certified genetic counselor and consider having [genetic testing](#). The testing can show if they have an inherited gene change that puts them at a higher risk for certain types of cancer. If so, the doctor might recommend screening and other tests (often at a younger age than usual) to help find cancer early, or they might prescribe medicines or suggest healthy habits that might help lower the person's cancer risk.

## Cancer diagnosis

For people with some types of cancer, their cancer cells might be tested for changes in certain genes (or for proteins made because of these gene changes). This testing can provide information about their cancer, including how it grows and spreads. These tests can go by many names, including:

- **Biomarker testing**
- **Tumor testing, tumor genetic testing, tumor marker testing, or tumor subtyping**
- **Genomic testing, genomic profiling, or genome sequencing**
- **Molecular testing or molecular profiling**

- **Somatic testing**
- **Next generation sequencing**

Testing is often done on a sample of the tumor (from a biopsy or surgery) if possible, but it might also be done using a sample of blood, saliva, or other body fluids.

## **Cancer treatment**

For some types of cancer, testing the cancer cells for certain gene or protein changes (known as **biomarkers**) can affect treatment options. In fact, this is often what people mean when they refer to “precision medicine.”

Gene or protein changes can affect how a cancer responds to certain treatments. Some people's cancers have gene changes that are different from those in other people, even if they have the same type of cancer. For example, not every melanoma skin cancer has the exact same gene mutations, so these cancers don't always respond to a treatment the same way.

One of the ways doctors can find out if certain gene or protein changes might affect how a cancer responds to treatment is with pharmacogenomic testing. This type of testing can tell how the person's body breaks down, absorbs, and uses treatment drugs.

Before starting treatment, doctors can test the cancer cells for certain gene and protein changes to help determine which treatments are likely to work best. The goal is to give treatments that are most likely to work, while avoiding giving treatments that might not work.

The two types of treatment most often used in precision medicine are [targeted drug therapy](#) (drugs designed to attack a specific target on cancer cells) and [immunotherapy](#) (medicines used to help the body's immune system attack the cancer).

To learn more about this type of testing to help guide treatment, see [Biomarker Tests and Cancer Treatment](#).

## **Types of cancer where precision medicine is used**

It's important to understand that precision medicine is not yet used for every type of cancer. However, the hope is that one day, treatments will be customized to the specific gene and protein changes in each person's cancer. A great deal of research is being done in this area.

Some of the more common cancers where precision medicine is being used to help with treatment decisions include:

- Colorectal cancer
- Breast cancer
- Lung cancer
- Certain types of leukemia
- Certain types of lymphoma
- Melanoma
- Esophageal cancer
- Stomach cancer
- Ovarian cancer
- Thyroid cancer

If you have a type of cancer for which treatment options might depend on if the cells have certain gene or protein changes, your cancer will likely be tested for them.

You might need to ask your doctor some questions to know if this type of testing was done. (See below.) People with the types of cancer listed above are usually tested for certain gene or protein changes when they are diagnosed, or shortly after. Some cancers might also be tested for changes if they keep growing during treatment, or if they come back.

### **Limitations to precision medicine in cancer**

Access to the latest precision medicine approaches might be limited in some places. A lot still needs to be learned about how precision medicine can be used in cancer care. Researchers are trying to fill those gaps, both in lab studies and in [clinical trials](#).

Many clinical trials are done with people who have specific types of cancer. But to be part of a precision medicine clinical trial, a person's cancer cells must have certain gene or protein changes that can be targeted by the medicine that's being tested. And precision medicine clinical trials are often available only at larger cancer centers. This means sometimes the chances for taking part in a clinical trial might be limited for some people.

Even when precision medicine is already available outside of a clinical trial, it might not always be used as well as it could be. For example:

- When it comes to learning about **cancer risk**, a person's family history of cancer

might not be well known or evaluated. Or, genetic testing might not have been done, its results might not be adequate, or the results might not be used to make the best decisions about health.

- Regarding **cancer treatment**, even if a person is diagnosed with a type of cancer where tests are available to look for gene or protein changes that might affect treatment options, the cancer might not be tested for these changes.
- The **costs** of biomarker testing and the medicines that might be recommended as a result of testing can sometimes be a concern. (See below.)

It's important to ask questions and know all options that are available to you.

### **Costs of precision medicine in cancer care**

Experts believe precision medicine might help lower health care costs in some ways. For example, it can help guide doctors in choosing treatments that are likely to work best. This means a patient might avoid getting (and having to pay for) treatments that aren't likely to work well, along with unnecessary side effects (and the possible costs that might go along with them).

But precision medicine might also increase some costs. For example:

- Tests for gene and protein changes can be expensive, especially if many changes are being tested for, and insurance might not cover all testing costs.
- For people who are found to be at higher risk for cancer because testing shows they have an inherited gene mutation, there might be increased costs from getting recommended screening tests and other preventive care. (On the other hand, they are doing what's needed to help lower their risk of cancer or to find it early, when it's often easier to treat, which might help them avoid higher costs in the future.)
- For people whose best treatment option is a specific targeted therapy or immunotherapy treatment, based on a cancer's gene or protein changes, the treatment itself might be expensive.

### **Questions to ask your doctor about precision medicine**

#### **If you're concerned about your cancer risk**

If you don't have cancer, but are concerned about your cancer risk because of your family history or some other reason, here are some questions you might want to ask your doctor:

- Do you think my family history puts me at high risk for cancer?
- Should I get genetic counseling and testing?
- How do I find a certified genetic counselor?
- What will happen if I decide to get genetic testing? What will happen if I decide not to?
- How much does genetic counseling and testing cost? Will my insurance cover it?
- What might you learn about my cancer risk from genetic testing?
- How will the results affect my care?
- Will the results lead to more medical tests?
- How might the results affect my family? Would my family members need to get genetic testing?

You can learn more in [Understanding Genetic Testing for Cancer](#).

### **If you have cancer**

If you have cancer, especially one of the cancers listed above in "Types of cancer where precision medicine is used," here are some questions you might want to ask your doctor:

- Do I have a type of cancer that should have biomarker or molecular testing done on it?
- What information will the tests being done on my cancer help you find out?
- Will the results of this testing mean I will need more tests?
- How much will the tests cost? Will my insurance cover them?
- Will these tests help us choose which treatments are best for me?
- If testing shows a specific treatment might be best for me, how much would this treatment cost? Will my insurance cover it?

You can learn more about what questions to ask in [Understanding Your Options and Making Treatment Decisions](#).

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