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# Precision or Personalized Medicine

## What is precision medicine?

Precision medicine is a way health care providers can offer and plan specific care for their patients, based on the person's genes (or the genes in their cancer cells). It's sometimes called *personalized medicine* or *personalized care*. Precision medicine looks at how a specific [gene change](#)<sup>1</sup> (gene mutation) might affect a person's risk of getting a certain cancer or, if they already have cancer, how their genes (or genes in their cancer cells) might affect treatment.

The approach uses information from genetic tests to help doctors put together a plan for care that usually involves very specific recommendations. In some cases, precision medicine 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 towards prevention if they are at 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 looking 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.

## What are gene mutations?

Precision medicine is based on knowing the effects of certain gene mutations (changes). When thinking about precision medicine, it can be helpful to understand what gene mutations are and how they can affect a person's risk for cancer or treatment for a cancer.

Each cell in a person's body has DNA, which contains our **genes**. Genes are the

instructions your cells use to make proteins needed to keep your body working normally.

When cells divide to make new cells, the genes inside those cells are copied. A gene change happens when there's a mistake in the copying process. Sometimes these changes come from a parent (*inherited* gene changes). But they can also happen sometime later in life (*acquired* gene changes). Some gene changes can be harmful, while others may not cause any problems.

## Gene mutations and cancer

All cancers are caused by a genetic change or mutation of some kind. Cancer cells are mutated versions of normal cells, meaning something changed in a normal cell to make it turn into a cancer cell. Experts agree that it takes more than one gene mutation 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. Depending on the type of mutation, an abnormal gene change might make a person more likely to develop a certain cancer. Or, if they already have cancer, the abnormal gene might mean that the cancer may not respond well to a certain type of treatment or drug. In some people with cancer, a specific gene mutation might mean that their prognosis (outcome) is better or worse than someone with the same cancer who does not have that gene change.

For example, when a person is diagnosed with cancer, they often get the same treatment as other people with that type of cancer. Sometimes a specific drug is used to treat a cancer with a certain abnormal gene. But gene changes might be different from one person to another, even if they have the same type of cancer. Because of this, each of those people might have a different response to the same drug. For certain cancer types, doctors can test for gene changes that can tell them if a person might respond better to one drug than another.

You can learn more in [Genetics and Cancer](#)<sup>2</sup>.

## Precision medicine in cancer

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 who might be at high risk for cancer

- Prevent some types of cancer
- Find certain cancers early
- Diagnose a specific type of cancer correctly
- Choose what treatment options are best
- Evaluate how a treatment is working

## Cancer risk and prevention

Sometimes precision medicine is used for people with certain cancers or 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 patient might meet with a certified genetic counselor and consider having [genetic testing](#)<sup>3</sup>. The testing can show if they have a gene change or mutation 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, and prescribe medicines or suggest healthy habits that might help lower cancer risk.

## Cancer diagnosis

For people with a cancer diagnosis, their tumor might be tested for certain types of gene changes or proteins made from those gene changes. This testing can provide information about how their cancer grows and spreads. These tests might be called *biomarker tests*, *chromosome tests*, *gene tests*, or *biochemical tests*. It might be done using a blood or saliva sample, biopsy tissue, or body fluids. If the test is done using a biopsy sample (from a tumor), it's done in a special lab and might be called by different names, such as *DNA mutational analysis*, *genomic testing*, *proteomics*, *biomarker testing*, *tumor profiling*, *cytogenetics*, *next generation sequencing*, or *molecular testing*.

## Cancer treatment

In some cancers, the gene testing done on a tumor can affect treatment choices. This is because certain gene changes can affect how a tumor responds to certain treatments. And some tumors have gene changes that are different from other tumors of the same type. For example, not every melanoma skin cancer will have the exact same gene mutations. This means these tumors might not respond to a treatment the same way. The goal is to give a treatment that can target a gene mutation, without causing too many side effects, and to avoid giving treatments that might not work. You can read more about 2 types of treatment often used in precision medicine: [targeted therapy](#)<sup>4</sup> and

[immunotherapy](#)<sup>5</sup>.

You can also learn more in [How Genes Can Help in the Diagnosis and Treatment of Cancer](#)<sup>6</sup>.

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

It's important to understand that precision medicine is not used for every cancer. However, the hope is that one day, treatments will be customized to the specific gene changes in each person's cancer. Much 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 a treatment is available to target a gene mutation that's common in your type of cancer, you (or your tumor) will likely be tested for it. You might need to ask your doctor some questions to know if testing was done. People with the types of cancer listed above are usually tested for certain gene changes when they are diagnosed, or shortly after. Some cancers are also tested if they get worse or come back.

### **Drawbacks and limitations to precision medicine in cancer**

Access to the latest precision medicine research might be limited. A lot still needs to be learned about how precision medicine can be used in cancer. Researchers are trying to fill those gaps, both in labs and in [clinical trials](#)<sup>7</sup>.

Many clinical trials are done with patients who have specific types and stages of cancer. But to be part of a precision medicine clinical trial, a person must have a certain genetic change that can be targeted by a medicine that's being tested. And precision medicine

clinical trials are often available only at larger cancer centers. This means sometimes the chances to be part of a clinical trials can be limited.

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

- A patient'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 its results are not used to make decisions about health.
- Even if a person is diagnosed with a type of cancer where gene tests are available, the tumor might not be tested to find out if there is a gene mutation that might change treatment choices.
- The cost of gene testing and the tests and medicines that might be recommended as a result of gene testing can be a concern.

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

### **Cost of precision medicine in cancer**

Experts believe precision medicine could help lower health care costs in some ways. This is because precision medicine can help guide doctors in choosing the right tests, which can then help them choose the treatments that will work best and hopefully have the fewest side effects. This means a patient might avoid getting treatments that are not likely to work well, along with unnecessary side effects.

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

- Tests for gene mutations can be expensive, and insurance might not cover all genetic counseling or testing costs.
- For people who are at high risk for cancer because of a gene mutation, there might be increased costs from getting recommended screenings and other preventive care. (On the other hand, they are doing what's needed to help prevent a cancer diagnosis or to find it early when it's often easier to treat, which can prevent higher costs in the in the future.)
- For people who need a specific targeted therapy or immunotherapy treatment because of a tumor's genetic makeup, the drug might be very 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 a family history or 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 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 will the results affect my family? Do my family members need to get genetic testing?

You can learn more in [Understanding Genetic Testing for Cancer](#)<sup>8</sup>.

## If you have cancer

If you have cancer, especially one of the cancers listed above in "Types of cancer precision medicine is used for," 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?
- Will the tests being done on my tumor help you find out more information about my cancer?
- Will the results of this gene testing mean I will need more tests?
- How much will the biomarker or molecular tests cost? Will my insurance cover them?
- Will these tests help you choose which treatments are best for me?
- How much will my treatment cost? Will my insurance cover it?

You can learn more about what questions to ask in [Understanding Your Options and Making Treatment Decisions](#)<sup>9</sup>.

## Hyperlinks

1. [www.cancer.org/cancer/cancer-causes/genetics/genes-and-cancer/gene-changes.html](http://www.cancer.org/cancer/cancer-causes/genetics/genes-and-cancer/gene-changes.html)
2. [www.cancer.org/cancer/cancer-causes/genetics.html](http://www.cancer.org/cancer/cancer-causes/genetics.html)
3. [www.cancer.org/cancer/cancer-causes/genetics/understanding-genetic-testing-for-cancer.html](http://www.cancer.org/cancer/cancer-causes/genetics/understanding-genetic-testing-for-cancer.html)
4. [www.cancer.org/treatment/treatments-and-side-effects/treatment-types/targeted-therapy.html](http://www.cancer.org/treatment/treatments-and-side-effects/treatment-types/targeted-therapy.html)
5. [www.cancer.org/treatment/treatments-and-side-effects/treatment-types/immunotherapy.html](http://www.cancer.org/treatment/treatments-and-side-effects/treatment-types/immunotherapy.html)
6. [www.cancer.org/cancer/cancer-causes/genetics/genes-and-cancer/genes-in-cancer-diagnosis-and-treatment.html](http://www.cancer.org/cancer/cancer-causes/genetics/genes-and-cancer/genes-in-cancer-diagnosis-and-treatment.html)
7. [www.cancer.org/treatment/treatments-and-side-effects/clinical-trials.html](http://www.cancer.org/treatment/treatments-and-side-effects/clinical-trials.html)
8. [www.cancer.org/cancer/cancer-causes/genetics/understanding-genetic-testing-for-cancer.html](http://www.cancer.org/cancer/cancer-causes/genetics/understanding-genetic-testing-for-cancer.html)
9. [www.cancer.org/treatment/treatments-and-side-effects/planning-managing/making-decisions.html](http://www.cancer.org/treatment/treatments-and-side-effects/planning-managing/making-decisions.html)

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**Written by**



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