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## [Understanding Pharmacogenomics](#) [1]

This section has been reviewed and approved by the [Cancer.Net Editorial Board](#) [2], 05/2016

Pharmacogenomics studies how medications interact with inherited genes. This includes:

- How inherited genes affect the way bodies **process** medications
- How inherited genes affect the way bodies **respond** to medications

Genetic differences mean that a drug can be safe for one person but harmful for another. One person may experience severe side effects from it. Another may not, even when given a similar dose.

### **How pharmacogenomics differs from genetic testing**

- **Standard genetic testing.** Standard genetic testing searches for specific genes. For example, *BRCA1* and *BRCA2* genes are linked with a higher risk of breast and ovarian cancer. The results of these genetic tests may prompt preventative steps. Such steps may include more frequent cancer screening, lifestyle changes, or preventive treatment.
- **Pharmacogenomics.** Pharmacogenomics is also a kind of genetic testing. It looks for small variations within genes. These variations may affect whether the genes can activate

or deactivate specific drugs. These test results can help the doctor choose the safest and most effective drug and dose.

Pharmacogenomics is constantly changing. Researchers continue identifying gene variations that affect responses to drugs. And as [personalized medicine](#) [3] grows, testing for gene variations may become more common.

## Why drugs work differently in different people

- **Drug activation.** Many drugs that treat cancer are not fully active. They need to be turned on by proteins that speed up chemical reactions in the body. These proteins are called enzymes.

Each person inherits variations in enzymes. The variations affect how fast drugs are changed to their active form. For example, some people break down drugs slowly. This means standard doses of treatment may not work as well.

- **Drug deactivation.** Drugs also need deactivation to limit the drug's exposure to healthy tissues. Deactivation means "to turn off." Some people may have slower enzymes. As a result, high levels of the drug may remain in their bodies for a long time. This means that they may have more side effects from the drug.

Besides pharmacogenomics, other factors may influence a person's reaction to a drug:

- Age and gender
- The cancer's stage
- Lifestyle habits, such as smoking and drinking alcohol
- Other diseases
- Medications taken for other conditions

## Benefits of pharmacogenomics

Pharmacogenomics offers important benefits:

- **It improves patient safety.** Severe drug reactions cause more than an estimated 120,000 hospitalizations each year. Pharmacogenomics may prevent dangerous drug reactions by identifying patients at risk.
- **It improves health care costs and efficiency.** Pharmacogenomics may help find appropriate medications and doses more quickly.
- **Challenges to pharmacogenomics.** Here are some challenges in the development and practical use of pharmacogenomics:
  - It can be expensive. It's particularly expensive if insurance doesn't cover the costs.
  - Not all tests may be widely available.
  - There could be privacy issues. This concern remains despite federal laws that make it illegal to discriminate based on genetic information.
  - There may be other, unresolved ethical and legal issues.

## Pharmacogenomic testing in practice

Here are some examples of pharmacogenomics testing in cancer care:

- **Colorectal cancer.** Irinotecan (Camptosar) is a type of chemotherapy. It is commonly used to treat colon cancer. In some people, genetic variations cause a shortage of the UGT1A1 enzyme. This enzyme is responsible for metabolizing irinotecan. Metabolism is the chemical reaction that helps the body process the drug. With a UGT1A1 shortage, higher levels of irinotecan remain in the body. This may lead to severe and potentially life-threatening side effects, especially if higher doses of the drug are used.

Doctors may use a pharmacogenomic test called the UGT1A1 test. It shows which patients have this genetic variation. For those who do, the doctor can prescribe a lower dose of irinotecan. Often, the lower dose is just as effective for these people.

- **Acute lymphoblastic leukemia (ALL).** Doctors use pharmacogenomic testing for children with acute lymphoblastic leukemia. About 10% of people have genetic variations

in an enzyme called thiopurine methyltransferase (TPMT). TPMT is responsible for metabolizing chemotherapy for ALL. To avoid severe side effects, children with lower TPMT levels receive lower doses.

- **Other cancer types.** Fluorouracil (5-FU) is a type of chemotherapy. It is used to treat several types of cancer, including colorectal, breast, stomach, and pancreatic cancers.

A genetic variation in some people causes lower levels of the enzyme called dihydropyrimidine dehydrogenase (DPD). DPD helps the body metabolize fluorouracil. Doctors may use a pharmacogenomic test to find this variation. If found, a lower fluorouracil dose helps prevent serious side effects.

## Questions to ask the doctor

Talk with your doctor about your treatment options and consider asking the questions below. You may also want to ask how your genetic makeup may affect your body's response to treatment.

- Would you explain my treatment options?
- Which treatment or combination of treatments do you recommend? Why?
- Do these treatments work differently in different people? If so, are there tests to find these differences?
- What are the possible side effects of this treatment?
- Is there a way to predict how my body will respond to this drug? To predict whether I might experience severe side effects?
- What are my options if the cancer does not respond to the drug? Or if I experience severe side effects?
- Whom do I call for questions or problems?

## More Information

[Genetics](#) [4]

## Additional Resource

American Medical Association: [Pharmacogenomics](#) [5]

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### Links

- [1] <http://www.cancer.net/navigating-cancer-care/how-cancer-treated/personalized-and-targeted-therapies/understanding-pharmacogenomics>
- [2] <http://www.cancer.net/about-us>
- [3] <http://www.cancer.net/node/24522>
- [4] <http://www.cancer.net/node/24864>
- [5] <http://www.ama-assn.org/ama/pub/physician-resources/medical-science/genetics-molecular-medicine/current-topics/pharmacogenomics.page?>