

Pharmacogenomics

What is pharmacogenomics?

Pharmacogenomics (sometimes called pharmacogenetics) is a field of research that studies how a person's genes affect how he or she responds to medications. Its long-term goal is to help doctors select the drugs and doses best suited for each person. It is part of the field of precision medicine, which aims to treat each patient individually.

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Your genes determine a lot about how you look. They also play a key role in how medications work in your body. Credit: NIH.

What role do genes play in how medicines work?

Just as our genes determine our hair and eye color, they partly affect how our bodies respond to medicine.

Genes are instructions, written in DNA, for building protein molecules. Different people can have different versions of the same gene. Each version has a slightly different DNA sequence. Some of these variants are common, and some are rare. And some affect health, such as those gene variants linked to certain diseases.

Scientists know that certain proteins affect how drugs work. Pharmacogenomics looks at variations in genes for these proteins. Such proteins include liver enzymes that chemically change drugs. Sometimes chemical changes can make the drugs more—or less—active in the body. Even small

differences in the genes for these liver enzymes can have a big impact on a drug's safety or effectiveness.

One liver enzyme, known as CYP2D6, acts on a quarter of all prescription drugs. For example, it converts the painkiller codeine into its active form, morphine. There are more than 160 versions of the CYP2D6 gene. Many vary by only a single difference in their DNA sequence. Others have larger changes. Most of these variants don't affect how people respond to the drug.

Typically, people have two copies of each gene. However, some people have hundreds or even thousands of copies of the CYP2D6 gene. Those with extra copies produce too much of the CYP2D6 enzyme and process the drug very fast. As a result, their bodies may convert codeine to morphine so quickly and completely that a standard dose can be an overdose. In contrast, some variants of CYP2D6 create an enzyme that doesn't work. People with these variants process codeine slowly, if at all, leading to little, if any, pain relief. For them, doctors can prescribe a different drug.

How is pharmacogenomics affecting drug design, development, and prescribing guidelines?

The Food and Drug Administration (FDA) monitors drug safety in the United States. It now includes pharmacogenomic information on the labels of [around 200 medications](#). This information can help doctors tailor drug prescriptions for individual patients by providing guidance on dose, possible side effects, or differences in effectiveness for people with certain gene variants.

Drug companies are also using pharmacogenomics to develop and market medicines for people with specific genetic profiles. By studying a drug only in people likely to benefit from it, drug companies might be able to speed up the drug's development and maximize its therapeutic benefit.

In addition, if scientists can identify genes that cause serious side effects, doctors could prescribe those drugs only to people who do not have those genes. This would allow some individuals to receive potentially lifesaving medicines that otherwise might be banned because they pose a risk for other people.

How is pharmacogenomics affecting medical treatment?

Currently, doctors prescribe drugs based mostly on factors such as a patient's age, weight, sex, and liver and kidney function. For a few drugs, researchers have identified gene variants that affect how people respond. In these cases, doctors can select the best medication and dose for each patient.

Additionally, learning how patients respond to medications helps to discern the different forms of their diseases.

What role does the National Institutes of Health (NIH) play in pharmacogenomics research?



Pharmacogenomics can help doctors pick the right treatment option and dose for each

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For many years, [NIH-funded scientists](#), through the Pharmacogenomics Research Network (PGRN), have studied the effect of genes on medications relevant to a wide range of conditions, including asthma, depression, cancer, and heart disease. The research findings are collected in an online resource called [PharmGKB](#). In addition, the [Clinical Pharmacogenetics Implementation Consortium](#) (CPIC) was started as a shared partnership between the PGRN and PharmGKB to help lower the barrier to clinical use of pharmacogenetic tests. CPIC creates, curates, and posts freely available, peer-reviewed, evidence-based, updatable, and detailed gene/drug clinical practice guidelines. Another NIH-funded project, the [Clinical Genome Resource](#), aims to define the clinical relevance of genes and variants for use in precision medicine and research.

NIH takes seriously the ethical and legal implications of pharmacogenomic research. It works closely with researchers, clinicians, and patient advocates to ensure research participants' privacy. And it strives to maximize the benefits of pharmacogenomics research for individuals and society. An NIH initiative where people can participate and learn more is called [All of Us](#).

Important goals for NIH are to further pharmacogenomics research and ensure that doctors implement the findings. This represents part of a major initiative on precision medicine that aims to tailor treatments based on each person's genes, environment, lifestyle, and other characteristics.

NIGMS is a part of the National Institutes of Health that supports basic research to increase our understanding of biological processes and lay the foundation for advances in disease diagnosis, treatment, and prevention. For more information on the Institute's research and training programs, visit <https://www.nigms.nih.gov>.

Content updated October 2017



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