

What are genes?

Genes are segments of DNA that contain instructions for building the molecules that make the body work. Most of the molecules are proteins. Parents pass their genes to their offspring.

What is a genome?

A genome is all of the genetic material in an organism. It is made of DNA (or RNA in some viruses) and includes genes and other elements that control the activity of those genes.

Does everybody have the same genome?

The human genome is mostly the same in all people. But there are variations across the genome. This genetic variation accounts for about 0.001 percent of each person's DNA and contributes to differences in appearance and health. People who are closely related have more similar DNA.

Some of the variations between individuals result from epigenetic changes. These changes arise from chemical tags that attach to DNA and affect how cells read DNA's instructions. Epigenetic changes can be inherited.

Is genetic variation related to health and disease?

Many differences in DNA have no effect on health or disease risk. But some do. Because parents pass their genes on to their offspring, some diseases tend to cluster in families, similar to other inherited traits. Genetic variations can influence how people respond to certain medicines as well.

What does it mean to have a genetic risk?

Having a genetic risk means that a person has inherited the tendency to develop a certain illness. It does not mean that he or she will definitely develop the illness. Rather, it means there is a higher chance of developing it than if he or she did not have the risk.



DNA is shaped like a corkscrew-twisted ladder, called a double helix. The two ladder rails are referred to as backbones, and the rungs are pairs of building blocks called bases. Humans have about 3 billion base pairs in each cell. Credit: NIGMS.

What can a genetic test reveal?

A genetic test can identify genetic variations that studies have linked to the risk of developing a specific disease or of passing a disease gene on to descendants. A genetic test can also indicate how a person might respond to certain medicines.

Why does genetic research sometimes involve specific population groups?

Population groups often include people with the same ancestry and therefore very similar genomes. Plus, populations may share diets, environments and other characteristics that influence health. These genetic and environmental similarities make it easier for scientists to spot rare differences in a population group that could be related to health or disease.



The ability to determine the precise order of bases that make up DNA is speeding biomedical research. Credit: Stock image.

Such studies also can help explain why some medical conditions are more common in certain population groups.

Can researchers study someone’s genes without permission?

Scientists who conduct research with people follow strict rules. Among other things, they must obtain signed consent from participants. And before collecting blood, cheek swabs or other samples that contain DNA, researchers must tell participants the purpose of the study, how they will use the samples, and whether and for how long they will store the samples.

Why do scientists study the genes of other organisms?

All living things evolved from a common ancestor. Therefore, humans, animals and other organisms share many of the same genes, and the molecules made from them function in similar ways. For example, the human and mouse genomes are about 85 percent the same. Two-thirds of human genes known to be involved in cancer have counterparts in the fruit fly.

Researchers have found many genes that have been preserved in multiple organisms for millions of years. They can study these preserved genes and compare the genomes of different species to find similarities and differences that improve their understanding of how human genes function and are controlled. This helps researchers develop new strategies to treat and prevent human disease. Scientists also study the genes of bacteria, viruses and fungi to find ways to prevent or treat infection. Increasingly, these studies are helping them understand how microbes on and in the body affect human health, sometimes in beneficial ways.

What types of genetics research does the National Institute of General Medical Sciences support?

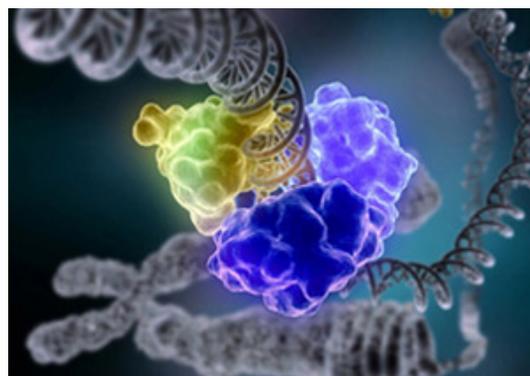
Increasingly sophisticated tools and techniques are allowing NIGMS-funded scientists to ask more precise questions about the genetic basis of biology. They’re working to better understand, for example, the factors that control when genes are active, the mechanisms DNA uses to repair broken or damaged segments, and the complex ways traits are passed to future generations. Another area of study focuses on tracing genetic variation over time to detail human evolutionary history and to pinpoint the emergence of disease-related attributes. [Read about specific efforts.](#) These basic research studies will continue to build a strong foundation for more disease-targeted studies.

Where can I learn more?

NIH’s National Human Genome Research Institute offers a range of resources on this topic, including a [glossary of genetic terms](#), [fact sheets about genetic and genomic science](#), and [FAQs on genetic and genomic medicine](#).

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>.

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The 2015 Nobel Prize in chemistry honored research revealing how our cells repair damage to DNA using molecules, such as the one shown here wrapping around DNA’s double helix. Without ways to fix damaged DNA, cells can malfunction, die, or become cancerous. Credit: Tom Ellenberger, Washington University School of Medicine in St. Louis, and Dave Gohara, Saint Louis University School of Medicine.

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[The New Genetics](#)
Booklet

[Circadian Rhythms](#) Fact Sheet

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Other Resources

[Genetics](#) (MedlinePlus, NIH)

[National Human Genome Research Institute](#) (NIH)