HEALTH BASICS

# Genes

Genes are sections of DNA that help cells build and regulate the body.

**WHAT IT IS**
DNA instructions used to build and regulate the body

**WHERE IT COMES UP**
Family history, genetic tests, diagnoses, and medication decisions

**WHY IT MATTERS**
Can influence traits, disease risk, and treatment response

**WHAT IT IS NOT**
A disease or a diagnosis

**WHO TO ASK**
A genetic counselor or medical genetics clinician

**HOW IT IS CHECKED**
DNA sequencing or chromosome tests when clinically relevant

## Overview

Genes are sections of DNA, the chemical material that carries biological instructions. They help cells make proteins or functional RNA molecules used for growth, communication, repair, and specialized tasks.

Genes are not diseases or diagnoses. Everyone has them. Differences in genes contribute to ordinary traits, such as blood type, as well as some health conditions and differences in medication response.

Most DNA is packaged into chromosomes. Most human cells contain 23 chromosome pairs, with one chromosome in each pair usually inherited from each biological parent. A small amount of DNA is also found in mitochondria, which help cells produce energy.

People usually have two copies of most genes. Different versions are sometimes called alleles, while a DNA sequence difference may be called a variant. Most variants are harmless. Some affect how a gene works, and others have uncertain effects.

Genes are only part of health. Age, environment, infections, nutrition, activity, healthcare, chance, and other factors also influence whether a condition develops.

## How it works

DNA uses four chemical building blocks, represented by the letters A, C, G, and T. Their order forms a sequence that cells can use as instructions.

Some genes provide instructions for proteins that form tissues, transport substances, or control chemical reactions. Other genes produce RNA that functions directly. DNA around and within genes helps control when, where, and how strongly a gene is used.

Cells do not use every gene at once. Liver cells and nerve cells usually contain the same DNA but activate different sets of genes, allowing them to perform different jobs.

When eggs and sperm form, chromosomes are mixed and divided. Siblings can therefore inherit different combinations of variants from the same biological parents.

Inheritance patterns vary. A condition may involve one altered gene copy, two altered copies, the X chromosome, or mitochondrial DNA. Many common conditions result from the combined effects of many genes and non-genetic factors.

Not every genetic change is inherited. A variant present in an egg or sperm can be passed to a child and is often called a germline variant. Somatic variants arise in particular cells during life. They are common in cancer cells but are not usually passed to children.

## Why it matters for your health

A gene variant may cause a genetic condition, change the chance of developing one, affect medication response, or have no known health effect.

For example, certain variants in CFTR can cause cystic fibrosis when a person inherits an affected copy from each biological parent. Some variants in LDLR can cause familial hypercholesterolemia and very high cholesterol. Not every change in these genes causes disease.

Some variants influence risk without determining what will happen. A person may carry a risk-related variant and never develop the associated condition. Penetrance describes how often people with a particular variant develop its related feature or condition.

Pharmacogenetic testing examines selected variants that may help explain medication effectiveness or side effects. Results are only one part of prescribing and do not automatically show that a medicine is safe, unsafe, or suitable for you.

Family history can suggest inherited risk because biological relatives share some genes. However, relatives may also share environments and habits, so family history does not prove that a gene is responsible.

## What can go wrong

A pathogenic variant is a DNA change supported by evidence as causing or contributing to disease. Examples include variants associated with sickle cell disease, cystic fibrosis, or Lynch syndrome. "Pathogenic" describes the evidence about a variant, not how severely a particular person will be affected.

Larger chromosome changes may involve missing, duplicated, or rearranged DNA. Down syndrome, for example, usually involves an extra copy of chromosome 21 rather than a variant in one gene.

Cancer can develop as changes accumulate in genes controlling cell growth, DNA repair, or cell death. Most arise in cancer cells during life. Some people inherit variants that increase susceptibility to certain cancers, but increased risk is not the same as having cancer.

A variant of uncertain significance (VUS) means there is not enough evidence to classify a change as harmful or harmless. It should not be treated as confirmation of a genetic condition, and its classification may change as evidence improves.

Testing can also reveal unexpected information, including biological relationships or risks unrelated to the original reason for testing. Possible findings depend on the test's scope.

## What you can do

You do not need genetic testing simply because everyone has genes. Testing is most useful when it addresses a specific medical or family-history question.

If genetics may be relevant to your care:

A genetic counselor or medical genetics clinician can explain inheritance, test limitations, possible results, and family implications. A cancer genetics service may help with inherited cancer questions. Other specialists may interpret findings related to their field.

Consumer and clinical genetic tests are not interchangeable. They may examine different variants or use different methods. A health-related consumer result may need confirmation in a clinical laboratory before medical decisions are made.

Genetic results are generally discussed through routine follow-up. The word "gene" on a report does not itself create an emergency. Follow the instructions from the service that ordered the test.

Urgent care depends on what is happening now, not on the general concept of genes. Seek emergency help for severe current problems such as collapse, major breathing difficulty, or

signs of a stroke, regardless of genetic testing history.

- Record major diagnoses in biological relatives and their ages at diagnosis
- Ask what question the proposed test is intended to answer
- Ask whether it examines one gene, several genes, or whole chromosomes
- Request the exact laboratory report, not only a verbal summary
- Ask how positive, negative, or uncertain results would affect your care
- Ask whether results could have implications for biological relatives

## Common questions

Does one gene control one trait?

Sometimes a gene has a major effect, but relationships are rarely that simple. One gene can affect several processes, while one trait may involve many genes plus environmental factors.

Are all gene variants harmful?

No. Genetic variation is normal. Most variants are harmless or have effects too small to notice. Only some are associated with disease or medication response.

Does a positive genetic result mean I will become ill?

Not always. Meaning depends on the gene, specific variant, inheritance pattern, and available evidence. Some results confirm a condition, while others indicate increased risk.

Does a negative test rule out a genetic cause?

Not necessarily. A test may not cover every relevant gene or type of DNA change, and current knowledge may be incomplete. Results must be interpreted alongside the reason for testing and the methods used.

Can genes change during life?

Inherited DNA is generally stable, but new changes can arise as cells divide or experience damage. These may remain limited to certain cells. Gene activity can also change without changing the DNA sequence.

Can lifestyle change my genes?

Lifestyle does not usually rewrite inherited DNA. It can affect how some genes are regulated and modify health risks related to genetic susceptibility, but it cannot remove an inherited pathogenic variant.

## Sources

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