HEALTH BASICS

Genetics

Genetics explains how genes and inherited biological differences can affect health

WHAT IT IS
The study of genes, inheritance, and biological variation

WHERE IT COMES UP
Family history, pregnancy care, diagnoses, and genetic test results

WHY IT MATTERS
It can clarify health risks, diagnoses, and treatment choices

WHAT IT IS NOT
Not a disease or diagnosis

WHO TO ASK
A genetic counselor or clinician familiar with your health question

HOW IT IS CHECKED
Genetic testing, chromosome analysis, or family history assessment

Overview

Genetics is the study of genes, inheritance, and biological differences. It is not a disease or diagnosis.

Genes are sections of DNA, the chemical instructions used by the body's cells. Most genes help make proteins or control how cells work. Genes are packaged into structures called chromosomes.

You inherit DNA from both biological parents. This helps explain shared features among relatives and why some health conditions run in families. However, genes are only part of the picture. Age, environment, lifestyle, chance, and other biological factors also affect health.

A genetic difference is often called a variant . Most variants are harmless. Some affect features such as hair texture, while others raise the chance of a health condition, contribute to a condition, or influence responses to medicine.

Genetics may arise in pregnancy care, newborn screening, cancer care, medication planning, unexplained health problems, or after a relative receives a genetic diagnosis.

How it works

Most body cells contain 23 pairs of chromosomes, with one chromosome in each pair usually coming from each biological parent. A person's complete DNA is their genome . Their genetic makeup is sometimes called their genotype , while observable features are their phenotype . Phenotype can reflect genes, environment, age, and interactions among them.

Genetic conditions can follow different inheritance patterns:

Not every genetic finding is inherited. A new, or de novo , variant can arise in an egg, sperm, or early embryo. Genetic changes can also develop in certain cells later in life. For example, many changes found in cancer are limited to the tumor and were not inherited.

A variant does not always determine what will happen. Penetrance describes how often people with a variant develop its related feature or condition. Expressivity describes how strongly that feature appears. Both may differ even among relatives with the same variant.

Laboratories may classify variants as benign, likely benign, uncertain, likely pathogenic, or pathogenic. Pathogenic means evidence links a variant to disease; it does not necessarily mean the person currently has that disease. The gene, inheritance pattern, symptoms, and test limitations still matter.

- Autosomal dominant: one altered gene copy may be enough to affect health
- Autosomal recessive: both copies usually need relevant variants
- X-linked: the gene is on the X chromosome
- Mitochondrial: the variant is in DNA within mitochondria
- Multifactorial: many genes and non-genetic factors contribute

Why it matters for your health

Genetic information may help explain a diagnosis, identify a condition that runs in a family, or guide screening, treatment, or medication choices.

Tests may look for one variant, examine a group of genes, study chromosomes, or sequence large parts of the genome. The right test depends on the clinical question. A test for a known family variant differs from broad testing for unexplained medical or developmental concerns.

Results can matter to biological relatives because they share some DNA. However, one person's result does not establish another person's result. Relatives may need their own assessment or targeted testing.

Genetic risk is not certainty. A result may indicate a higher or lower chance of a condition

without predicting whether it will occur, when it may begin, or how serious it will be.

Some tests identify carrier status. A carrier has a variant associated with a recessive condition but usually does not have the condition. This information may be relevant when considering biological children.

What can go wrong

Gene or chromosome changes can contribute to conditions such as cystic fibrosis, sickle cell disease, Huntington disease, Down syndrome, and some inherited cancer syndromes. Common conditions such as heart disease and type 2 diabetes may also have genetic influences, usually alongside other factors.

Genetic tests have limits. A test may not examine every relevant gene or every type of change, so a negative result may not rule out a genetic cause. A positive result may need confirmation, especially if it came from a consumer test rather than a clinical laboratory.

A variant of uncertain significance (VUS) means there is not enough evidence to determine whether a variant affects health. It is not a positive diagnosis, and its classification may change as evidence develops.

Broad testing may uncover an unexpected secondary finding unrelated to the original reason for testing. Results may also reveal unexpected information about biological relationships. Discuss these possibilities before testing.

Privacy and insurance protections vary by country and insurance type. In the United States, the Genetic Information Nondiscrimination Act provides some protections for health insurance and employment, but it does not cover every setting or insurance product.

A family history or genetic report usually needs routine follow-up rather than emergency care. Arrange prompt clinical advice if a report says action is time-sensitive. Seek emergency care for current serious symptoms, such as severe breathing difficulty or loss of consciousness, not simply because a finding is genetic.

What you can do

You do not need genetic testing simply because genetics affects health. Testing is most useful when there is a clear question and the result could change care, clarify a diagnosis, or provide meaningful information.

Keep a family health record if possible. Include major diagnoses, approximate ages at

diagnosis, causes of death, and which side of the family was affected. Avoid guessing when details are unknown.

Before testing, consider asking:

A genetic counselor is trained to explain inheritance, test choices, uncertainty, and possible effects on relatives. Counseling does not require you to have testing.

If you already have a result, keep the complete laboratory report rather than only an app or portal summary. Record the laboratory, test date, gene, exact variant, and classification. A clinician or genetic counselor can check whether the interpretation still reflects current evidence.

Do not make major medical decisions from a direct-to-consumer result alone. A clinically important finding may need confirmation in an accredited clinical laboratory and interpretation alongside your personal and family history.

- What question is this test intended to answer?
- What could a positive, negative, or uncertain result mean?
- Could the result change my care?
- Might it reveal unrelated findings or information about relatives?
- Who will have access to the result?
- Would genetic counseling help before or after testing?

Common questions

Are genes my destiny?

Usually not. Some variants have strong effects, while others change risk only slightly. Environment, healthcare, age, and chance also influence outcomes.

Can two healthy parents have a child with a genetic condition?

Yes. Both parents may carry variants for the same recessive condition without having it. A new variant or chromosome change can also occur.

Does a negative genetic test mean there is no inherited risk?

Not always. A test may not cover every relevant gene or type of variant, and medical knowledge may be incomplete. Meaning depends on what was tested and why.

Can genetic results change over time?

Inherited DNA usually does not change, but its interpretation can. New evidence may lead a

laboratory to reclassify a variant. Keep your contact details current with the testing service when possible.

Are ancestry tests the same as medical genetic tests?

No. Ancestry tests primarily estimate genetic relationships and population origins. Some provide health reports, but their coverage and confirmation processes may differ from clinical testing.

Should relatives receive the same test?

Sometimes targeted testing is useful after a familial variant is clearly identified. The appropriate test and timing differ among relatives, so each person needs guidance for their situation.

Sources

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