HEALTH BASICS

Chromosome

A chromosome is a package of DNA and proteins that carries genes in a cell.

WHAT IT IS
DNA packaged with proteins inside a cell

WHERE IT COMES UP
Genetic tests, prenatal screening, fertility care, and cancer testing

WHY IT MATTERS
Chromosome changes can affect development, reproduction, or cancer care

WHAT IT IS NOT
Not a disease or diagnosis

WHO TO ASK
Genetic counselor or clinician who ordered the test

HOW IT IS CHECKED
Karyotype, chromosome microarray, or targeted genetic testing

Overview

A chromosome is a long piece of DNA packaged with proteins. DNA contains genes, which are instructions that help cells grow, work, and communicate. Chromosomes also contain areas that control when and how genes are used.

A chromosome is not a disease or a diagnosis. It is a normal part of human cells.

Most human cells with a nucleus usually contain 46 chromosomes, arranged in 23 pairs. You inherit one chromosome in each pair from the egg and the other from the sperm. The first 22 pairs are called autosomes. The remaining pair is often called the sex chromosomes, usually described as X and Y.

Chromosome patterns can differ. For example, some people have a different number or arrangement of X or Y chromosomes. Chromosomes do not, by themselves, describe every aspect of a person's sex, gender, health, or development.

Egg and sperm cells normally contain 23 chromosomes rather than 46. Mature red blood cells have no nucleus and therefore do not contain these nuclear chromosomes. Mitochondria, the structures that help cells produce energy, also carry a small separate piece of DNA.

How it works

DNA is extremely long compared with the size of a cell. Proteins called histones help coil and package it so it fits inside the nucleus. This packaged DNA forms chromosomes.

Each chromosome has recognizable parts. The centromere is a narrowed area that helps the cell move copied chromosomes during cell division. Telomeres are protective sections at the chromosome ends. They help keep chromosome ends from being treated like broken DNA.

Before most cells divide, they copy their chromosomes. The copies are then separated so each new cell receives a set. Errors can sometimes occur while chromosomes are copied or divided.

Genes occupy specific places on chromosomes. However, a chromosome is not made only of genes. It also contains DNA that controls gene activity and DNA with structural or other functions.

Chromosomes can be examined in several ways:

No single test finds every type of genetic change. The method used should match the question being asked.

- A karyotype displays chromosomes by size and shape, allowing large changes or an unusual chromosome number to be seen
- A chromosome microarray looks for smaller missing or extra sections of DNA, called copy-number changes
- FISH , or fluorescence in situ hybridization, uses fluorescent markers to examine a specific chromosome area
- DNA sequencing reads DNA letters and may find gene-level changes, but its ability to detect chromosome changes depends on the test

Why it matters for your health

Most chromosome differences are harmless. Some affect no important genes or occur in DNA regions where extra or missing material has little known effect.

Other changes can influence health when they alter the number of copies of a gene, interrupt a gene, or change how a gene is controlled. Their effects depend on which chromosome area is involved, how much DNA is affected, and whether the change is present in all or only some cells.

Chromosome testing may be discussed after an unusual prenatal screening result, developmental or learning concerns, repeated pregnancy loss, fertility difficulties, certain physical findings, or a family history of a chromosome rearrangement. It may also be used to

classify some cancers and help guide cancer care.

A screening test estimates the chance of a condition. It does not confirm a diagnosis. For example, cell-free DNA prenatal screening may suggest an increased chance of a chromosome condition, but diagnostic testing is needed to determine whether that condition is present.

A laboratory finding may also be described as a variant of uncertain significance . This means there is not enough evidence to know whether the finding affects health. It does not mean that a health problem will develop.

What can go wrong

Changes involving whole chromosomes are called numerical abnormalities . An extra chromosome is a trisomy, while a missing chromosome is a monosomy. Named examples include Down syndrome, usually caused by an extra copy of chromosome 21; Turner syndrome, associated with one complete or partial X chromosome; and Klinefelter syndrome, commonly associated with an XXY pattern.

Structural changes affect part of a chromosome. They include:

A rearrangement can be balanced , meaning there is no major gain or loss of DNA. A person with a balanced rearrangement may have no related health problems. However, it can sometimes affect fertility or increase the chance of a pregnancy having an unbalanced chromosome pattern.

Mosaicism means a chromosome change is present in some cells but not others. Its effects can differ according to the cells involved and the proportion carrying the change.

Some chromosome changes are inherited. Others happen when an egg or sperm forms, early in embryo development, or later within particular tissues. Changes found only in cancer cells are acquired changes and are not automatically present throughout the body or passed to children.

A chromosome result is not usually an emergency. Unexpected findings generally need routine follow-up with the clinician who ordered the test or a genetics service. If someone is acutely unwell, urgent or emergency care should be based on their current symptoms rather than delayed while waiting for genetic review.

- Deletion: a chromosome section is missing
- Duplication: a section is copied
- Translocation: chromosome material moves or exchanges places

- Inversion: a section breaks and reconnects in reverse order
- Ring chromosome: chromosome ends join to form a ring

What you can do

There is no routine action needed simply because you have chromosomes. Chromosome testing is not required for everyone.

If you receive a chromosome report, keep the complete laboratory report rather than only a summary. The test name, sample type, exact chromosome notation, and laboratory interpretation all matter.

Useful questions include:

A genetic counselor can explain chromosome terminology, the limits of testing, and possible effects on relatives or future pregnancies. Counseling does not require you to have more testing.

For prenatal results, ask whether the sample came from a screening blood test, placental tissue obtained through chorionic villus sampling, or amniotic fluid obtained through amniocentesis. These tests answer different questions.

For cancer testing, ask whether the finding was identified only in the tumor or blood cancer cells. A separate test using blood, saliva, or another non-cancer sample may sometimes be needed to assess whether a finding was inherited.

- What type of test was performed, and what can it detect?
- Is this a screening result or a diagnostic result?
- Was the finding inherited, acquired, or not yet classified?
- Could it affect my health, pregnancy planning, or relatives?
- Would testing a parent, partner, tumor, or different tissue clarify it?
- Is genetic counseling available?

Common questions

Are chromosomes the same as genes?

No. A chromosome is a large package of DNA containing many genes and regulatory areas. A gene is one section of DNA with a particular function.

Does everyone have exactly 46 chromosomes?

No. Forty-six is the usual number in most nucleated human cells, but naturally occurring chromosome differences exist. Cancer cells may also gain, lose, or rearrange chromosomes.

Can a normal karyotype rule out every genetic condition?

No. A karyotype mainly detects chromosome-number changes and larger structural changes. It may not find small DNA changes, changes within one gene, or low-level mosaicism.

Can lifestyle change a chromosome test result?

An inherited chromosome pattern is not changed by diet, exercise, or supplements. Acquired changes can develop in particular groups of cells, especially cancer cells, but they are not the same as changing every chromosome in the body.

Are chromosome changes always inherited?

No. Some are inherited from a parent, while others arise for the first time when reproductive cells form or during cell division. Testing relatives may sometimes clarify the origin.

Does an unusual chromosome result always cause health problems?

No. Its meaning depends on the specific change, genes involved, test method, and clinical context. Some findings are harmless or remain uncertain.

Sources

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