HEALTH BASICS

Translocation

A translocation is a chromosome change in which genetic material moves to another location.

WHAT IT IS
movement of chromosome material to a different chromosome location

WHERE IT COMES UP
chromosome tests, fertility care, pregnancy testing, and cancer testing

WHY IT MATTERS
may alter genes, reproductive outcomes, or cancer classification

WHAT IT IS NOT
not a disease or diagnosis by itself

WHO TO ASK
clinical geneticist, genetic counsellor, or ordering specialist

HOW IT IS CHECKED
karyotype, FISH, microarray, or sequencing selected for the question

Overview

A translocation is a change in the arrangement of chromosome material. Chromosomes are packages of DNA that contain your genes. In a translocation, part of one chromosome moves to a different place, often on another chromosome.

A translocation is not a disease or a diagnosis by itself. Its meaning depends on which chromosomes are involved, whether genetic material was gained or lost, and whether the change is present throughout the body or only in certain cells.

Some people are born with a translocation and never have related health problems. Others learn about one during fertility testing, pregnancy testing, an assessment of a child's development, or cancer testing.

The notation on a laboratory report may look like `t(9;22)` or `rob(14;21)`. These labels identify the chromosomes involved. They do not, by themselves, explain the health effects.

How it works

Most human cells usually contain 46 chromosomes arranged in 23 pairs. Egg and sperm cells usually contain one chromosome from each pair.

A balanced translocation rearranges chromosome material without an obvious overall gain or loss. A person with a balanced translocation is often called a carrier. Many carriers are healthy because they still have the expected amount of genetic material.

Balanced does not always mean harmless. A break can interrupt an important gene or affect how a nearby gene works. Some rearrangements that look balanced under a microscope may also contain DNA changes too small for that test to detect.

An unbalanced translocation causes extra or missing chromosome material. This can change the number of copies of many genes. The effects depend on the specific chromosome segments involved and cannot be predicted from the word "translocation" alone.

Two common patterns are:

A translocation may be constitutional , meaning it began in the egg, sperm, or early embryo and is present in many or all body cells. It may have been inherited or occurred for the first time in that person.

A translocation can also be acquired , or somatic. This means it developed later in a particular group of cells. Acquired translocations are important in some cancers and are generally not present in egg or sperm cells.

Different tests answer different questions. A karyotype provides a broad view of chromosomes and can identify many large rearrangements. FISH, or fluorescence in situ hybridisation, uses labelled DNA probes to look for a specific change. A chromosome microarray is good at finding extra or missing DNA but usually cannot detect a balanced translocation. Some sequencing tests can identify small breakpoints or gene fusions.

- Reciprocal translocation: segments from two chromosomes exchange places
- Robertsonian translocation: two specific chromosomes join near their centres, usually with loss of their short arms

Why it matters for your health

For a healthy person with a balanced constitutional translocation, the main concern may be reproduction rather than personal health.

During the formation of eggs or sperm, the rearranged chromosomes must separate. Some eggs or sperm may receive an unbalanced amount of chromosome material. Depending on the specific translocation, this can be associated with difficulty becoming pregnant, miscarriage, or

a pregnancy affected by a chromosome condition.

A child who inherits the same balanced arrangement may also be healthy. A child who inherits an unbalanced arrangement may have developmental, learning, growth, or physical differences. The possible outcomes are specific to the chromosome segments involved.

In cancer care, an acquired translocation can help identify or classify a cancer and may influence which additional tests or treatments are considered. For example, the translocation commonly written as `t(9;22)` creates the BCR::ABL1 gene fusion in chronic myeloid leukaemia and some acute leukaemias. Finding this change in cancer cells does not automatically mean it was inherited.

The laboratory report and the reason for testing are therefore essential. The same word can have very different meanings in prenatal testing, a child's chromosome assessment, or a cancer work-up.

What can go wrong

A translocation may relate to several named conditions or situations:

A test can also leave uncertainty. A karyotype may show a rearrangement without locating the exact DNA breakpoints. Conversely, a microarray may identify extra or missing DNA without showing how the chromosomes are arranged. Additional testing is sometimes needed to connect the findings.

Results can also be misunderstood when "balanced" is treated as a guarantee of no effect, or when a cancer-cell finding is assumed to be inherited. A genetics or cancer specialist can clarify which interpretation applies.

- Recurrent pregnancy loss or infertility: a balanced carrier may produce some eggs or sperm with unbalanced chromosomes
- Chromosome conditions: an unbalanced translocation can cause extra or missing genetic material
- Translocation Down syndrome: some cases result from a Robertsonian translocation involving chromosome 21
- Developmental or congenital disorders: these can occur if a rearrangement removes, duplicates, or disrupts important genes
- Blood cancers and solid tumours: acquired translocations can create abnormal gene fusions that help cancer cells grow

What you can do

There is no routine translocation screening for someone without a relevant personal history, family finding, pregnancy concern, or medical reason for testing. If none of these applies, there may be nothing you need to do.

If a report mentions a translocation, keep the full report rather than only the chromosome notation. Ask:

A clinical geneticist or genetic counsellor can review reproductive and family implications. Options may include testing during pregnancy with chorionic villus sampling or amniocentesis. Some people consider in vitro fertilisation with preimplantation genetic testing for structural rearrangements, called PGT-SR. These are individual choices, not required steps.

Screening tests, including cell-free DNA pregnancy screening, are not the same as diagnostic chromosome testing and do not reliably rule out every translocation.

A translocation result is not usually an emergency. Arrange routine follow-up when it comes from fertility care, family testing, or a non-urgent chromosome assessment. If it comes from an active cancer investigation, contact the ordering team promptly because the timing depends on that clinical situation. Seek emergency care for severe symptoms such as major bleeding, difficulty breathing, or collapse-not for the chromosome term alone.

- Is this balanced or unbalanced?
- Is it constitutional, inherited, newly occurring, or limited to abnormal cells?
- What chromosome segments and genes are involved?
- Would a karyotype, microarray, FISH test, or sequencing add useful information?
- Should biological parents, children, or other relatives be offered testing?
- Could this affect pregnancy planning or prenatal testing?

Common questions

Can a translocation be inherited?

Yes. A constitutional translocation can be inherited from a biological parent. It can also occur for the first time in an egg, sperm, or early embryo. Testing biological parents may help clarify which happened.

Does a balanced translocation affect life expectancy?

The label alone cannot answer this. Many balanced carriers have no related health problems.

Effects are possible if a breakpoint disrupts a gene or if additional DNA changes are present.

Does a translocation mean I have cancer?

No. Constitutional translocations and acquired cancer-cell translocations are different situations. The sample tested, the cells involved, and the exact finding determine its meaning.

Can a blood test find one?

Often, but the test must be appropriate. A blood karyotype can find many constitutional rearrangements. FISH or sequencing may look for a specific change. Cancer testing may use blood, bone marrow, or tumour tissue.

Can a translocation disappear?

A constitutional translocation generally remains part of a person's chromosome arrangement. An acquired translocation may become undetectable when the abnormal cell population responds to cancer treatment, but this is assessed with condition-specific tests.

Sources

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