HEALTH BASICS

Mosaicism

Mosaicism means that some cells in one person have a genetic change that other cells do not.

WHAT IT IS
Two or more genetically different cell groups in one person

WHERE IT COMES UP
Genetic tests, prenatal testing, cancer testing, or family risk discussions

WHY IT MATTERS
May change health effects, test interpretation, or family risk

WHAT IT IS NOT
Not a disease or diagnosis by itself

WHO TO ASK
Clinical genetics service or genetic counsellor

HOW IT IS CHECKED
Chromosome or DNA testing, sometimes in more than one tissue

Overview

Mosaicism means that a person has two or more groups of cells with different genetic information. The cells usually began from the same fertilized egg. A genetic change then happened as cells divided and grew.

The word comes from a mosaic artwork made of different pieces. In genetic mosaicism, the "pieces" are groups of cells.

Mosaicism is not a disease or a diagnosis by itself. Its importance depends on the genetic change, which cells carry it, and how those cells affect the body. Some mosaic changes have no known effect. Others are connected with a genetic condition, cancer, pregnancy finding, or family planning question.

A laboratory report may describe a result as "mosaic," "possible mosaic," or "low-level mosaic." These terms do not automatically show how serious the finding is. The sample type and testing method matter.

How it works

After an egg and sperm join, the resulting cell copies its DNA and divides. This continues as the body develops. If a genetic change happens during one of these divisions, cells descending from that cell may carry the change while other cells do not.

The timing helps determine where the changed cells are found. An early change may be present in several organs. A later change may be limited to one area of skin, one organ, the blood, or a tumour.

Several forms of mosaicism may appear in medical discussions:

The genetic difference may involve one DNA letter, a section of DNA, or a whole chromosome. For example, some cells may have an extra chromosome while others have the usual number.

A report may give a percentage or a variant allele fraction , often shortened to VAF. VAF is the proportion of DNA reads containing a particular change. It is not always the same as the percentage of cells affected. Chromosome copy number, sample quality, and the testing method can alter the result.

Mosaicism is different from chimerism . Mosaic cells usually come from one fertilized egg. In chimerism, genetically different cells come from more than one fertilized egg, or from another person through events such as a stem cell transplant.

- Somatic mosaicism affects body cells, such as blood, skin, or brain cells
- Germline or gonadal mosaicism affects some egg or sperm cells
- Constitutional mosaicism is present from early development and may involve several tissues
- Confined placental mosaicism is limited mainly or entirely to the placenta

Why it matters for your health

The effect of a mosaic change depends on more than its percentage. The specific gene or chromosome matters, as does the location of affected cells. A low level in a critical tissue may matter more than a higher level somewhere else.

Blood is commonly tested because it is easy to collect. However, a blood result may not represent the skin, brain, reproductive organs, placenta, or other tissues. A change found in skin but not blood can still be important. Likewise, a negative blood test may not rule out mosaicism elsewhere.

Mosaicism can help explain why people with the same named genetic condition have different features. Examples include mosaic forms of Down syndrome or Turner syndrome and conditions caused by mosaic changes, such as McCune-Albright syndrome and some

segmental overgrowth disorders. The diagnosis and health effects come from the specific condition, not from mosaicism alone.

A mosaic result can also affect family risk. A change limited to body cells may not be passed on. If some egg or sperm cells carry it, transmission may be possible even when a parent's blood test is negative.

In prenatal testing, the sampled tissue matters. Chorionic villus sampling examines placental tissue, while amniocentesis examines cells collected from amniotic fluid. A mosaic result from the placenta may not reflect the fetus. Cell-free DNA testing is a screening test based mainly on placental DNA; it cannot diagnose fetal mosaicism.

What can go wrong

A mosaic result can be misunderstood as a complete diagnosis. In practice, the laboratory finding must be connected with the person's medical history, examination, and the tissue tested.

A result may underestimate mosaicism if the changed cells are uncommon in the sample. It may miss a change that is absent from blood but present elsewhere. Testing another tissue, such as saliva, cheek cells, skin, or tumour tissue, may sometimes help, but an invasive biopsy is not routinely appropriate for every finding.

Results can also appear mosaic for reasons other than widespread constitutional mosaicism. A blood sample may contain an age-related group of blood cells with an acquired genetic change. This is called clonal haematopoiesis . Tumours also acquire genetic changes that may not be present in the rest of the body. Sample contamination and technical limits can sometimes produce uncertain results.

Specific situations associated with mosaicism include chromosome conditions, certain skin or overgrowth disorders, cancer, and confined placental mosaicism. Each has its own possible health effects and follow-up. Mosaicism itself does not have symptoms, a prognosis, or a treatment.

A report mentioning mosaicism is not usually an emergency. Arrange routine follow-up to understand the result and whether confirmation is useful. Seek urgent care for symptoms related to a known condition or pregnancy-such as severe pain, heavy bleeding, fainting, or serious breathing difficulty-rather than for the word "mosaicism" on a report.

What you can do

If mosaicism appears on a test report, keep the complete report rather than only a summary. Record what type of sample was tested, when it was collected, and whether the finding came from chromosome testing, sequencing, prenatal testing, or tumour testing.

Useful questions include:

A clinical geneticist or genetic counsellor can explain the result in context. An obstetric genetics or maternal-fetal medicine service may be appropriate for a prenatal finding. Cancer-related results may need review by the oncology team and, when relevant, a genetics service.

There is no home test that can map mosaicism throughout the body. Consumer DNA tests may miss low-level changes and usually cannot establish which tissues are involved. There is also no lifestyle change that removes a mosaic genetic change.

Testing relatives is not automatically helpful. It should be considered only when the specific finding and family situation make the result meaningful.

- What exact genetic or chromosome change was found?
- Which tissue was tested?
- Is the finding confirmed, possible, or uncertain?
- Could it be limited to blood, a tumour, or the placenta?
- Would testing another tissue change medical care?
- Does this result affect relatives or future pregnancies?
- Is follow-up based on a named condition rather than mosaicism itself?

Common questions

Does everyone have some genetic differences between cells?

Small acquired DNA differences build up as cells divide and age. Most are harmless and never appear on a medical test. Clinically reported mosaicism usually refers to a detectable difference that may help answer a health or reproductive question.

Does a higher percentage always mean a greater effect?

No. The percentage in one sample may not match other tissues. The gene or chromosome involved and the location of affected cells can be more important than the reported level.

Can mosaicism be inherited?

Many mosaic changes begin after fertilization and were not inherited from a parent. However,

a person with germline mosaicism may have egg or sperm cells carrying a change and may pass it on. The chance cannot be determined from the word "mosaic" alone.

Can a negative blood test rule it out?

Not always. Blood testing may miss a change limited to another tissue. Whether another sample is useful depends on the suspected condition and whether the result would change care.

Can the proportion change over time?

It can. Some cell groups expand, shrink, or remain stable. This is especially relevant in blood and tumours. A change in the measured level may also reflect a different sample or laboratory method.

Does mosaicism need treatment?

Mosaicism itself is not treated. Care, if needed, is directed at the specific condition or health issue associated with the genetic change. Some findings require only explanation and routine follow-up.

Sources

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