TEST

Karyotyping

Karyotyping examines the number and large-scale structure of chromosomes in a sample of cells.

WHAT IT MEASURES
Chromosome number and large changes in chromosome structure

SAMPLE
Blood, bone marrow, amniotic fluid, placental tissue, or other tissue

FASTING NEEDED?
No

HOW LONG IT TAKES
5-10 minutes for blood; longer for marrow or prenatal sampling

RESULTS IN
About 1-3 weeks, varying by laboratory and sample

USED FOR
Diagnosis, prenatal diagnosis, and cancer assessment

MAIN LIMITATION
Small DNA changes and low-level mosaicism may be missed

Overview

Karyotyping examines your chromosomes. Chromosomes are packages of DNA found in most cells. They contain genes, which provide instructions for how the body develops and works.

People usually have 46 chromosomes arranged in 23 pairs, although medically important differences can occur. A karyotype shows whether chromosomes are missing, added, or rearranged. It can identify large deletions, duplications, inversions, and translocations. A translocation happens when chromosome material moves to a different location.

The laboratory grows cells from blood, bone marrow, amniotic fluid, placental tissue, or another tissue. The chromosomes are stained to create light and dark bands, photographed under a microscope, and arranged into pairs.

Karyotyping is different from DNA sequencing. Sequencing examines much smaller sections of DNA. Chromosomal microarray testing can find smaller missing or extra pieces of chromosome material, but it may not detect balanced rearrangements that karyotyping can see.

Why the test is ordered

Karyotyping may be ordered to look for a chromosome difference that could explain certain health, development, fertility, or pregnancy findings.

It may help investigate:

In cancer care, the test looks for chromosome changes acquired by the abnormal cells. These findings can help classify a cancer and may guide treatment or monitoring. An acquired finding in cancer cells does not automatically mean the same change is present throughout your body or can be inherited.

- Developmental differences, learning disability, or multiple conditions present from birth
- Infertility or repeated pregnancy loss
- A prenatal screening or ultrasound result suggesting a chromosome condition
- A chromosome rearrangement already found in a relative
- Certain blood or bone marrow cancers, including some leukemias

Who should have the test

Karyotyping is most useful when there is a specific reason to look for a change in chromosome number or structure. A healthcare professional or genetic counsellor may discuss it with you based on your medical history, family history, examination, imaging, or other test results.

During pregnancy, diagnostic karyotyping may be offered after an abnormal screening result, an ultrasound finding, or identification of a chromosome rearrangement in a parent. Screening estimates chance; karyotyping of a prenatal sample is a diagnostic test for the chromosome changes it can detect.

Karyotyping is not a complete test for every genetic condition. If a small DNA change or a change within one gene is suspected, another test may be more suitable.

How to prepare

No fasting is needed for a blood karyotype. You can usually eat, drink coffee, exercise, and take your usual medicines.

Tell the testing team if you have recently had a blood transfusion or a stem cell or bone marrow transplant. These can affect which person's cells are present in a blood sample. Also mention

current cancer treatment, because it can affect cell growth in the laboratory.

Amniocentesis, chorionic villus sampling, bone marrow sampling, and biopsies need their own preparation. You may receive instructions about your bladder, medicines, or eating beforehand. Do not stop prescribed medicine unless the team performing the procedure tells you to.

What happens during the test

For a blood karyotype, a clinician places a needle into a vein, usually in your arm, and collects a small tube of blood. The appointment normally takes only a few minutes.

A bone marrow sample is usually taken from the back of the hip bone using a needle after the area is numbed. This takes longer and may cause pressure or brief pain.

During pregnancy, cells may be obtained by chorionic villus sampling, which takes placental tissue, or amniocentesis, which takes fluid surrounding the fetus. Both are specialist procedures guided by ultrasound.

In the laboratory, cells are encouraged to divide. Staff collect them when the chromosomes are most visible, then stain and examine multiple cells. Growing the cells is a major reason results take longer than many routine blood tests.

Understanding your results

There is no numeric reference range for a karyotype. The report describes the chromosomes seen and any differences found. Interpretation varies by sample, age, sex chromosome pattern, pregnancy status, medicines or treatment, and clinical context.

A result may be described as:

A balanced rearrangement may not cause health problems for the person carrying it. However, it can sometimes affect fertility or the chance of a pregnancy having an unbalanced chromosome pattern.

A typical result does not rule out every genetic condition. It may be followed by chromosomal microarray testing, a targeted gene test, or broader DNA sequencing if the clinical question remains.

Cancer karyotypes are interpreted differently from constitutional karyotypes, which assess chromosome patterns present from birth. The report may describe several groups of abnormal cells and how many cells showed each finding.

- Typical: No chromosome-number change or large structural difference was found
- Aneuploid: A chromosome is present in an extra or missing copy
- Structural: Part of a chromosome is deleted, duplicated, inverted, or moved
- Balanced: Chromosome material is rearranged without an obvious gain or loss
- Mosaic: The change appears in some tested cells but not others

What affects the results

The chromosome pattern itself is not changed by food, coffee, exercise, or time of day. However, several factors can affect what the laboratory is able to see.

Cells must be alive and able to divide. Delayed transport, poor sample quality, contamination, or too few cells may cause culture failure or an incomplete result. Some medicines and cancer treatments can reduce cell growth.

A recent transfusion can introduce donor white blood cells, although modern blood processing reduces this concern. A stem cell or bone marrow transplant can make a blood karyotype show the donor's chromosomes rather than your original chromosome pattern.

Mosaic changes may be present in one tissue but absent or uncommon in another. A blood result may therefore differ from skin, bone marrow, placental, or fetal cells.

In pregnancy, placental cells can occasionally have a chromosome pattern that differs from fetal cells. This is called confined placental mosaicism. A result from chorionic villus sampling may then need clarification with amniocentesis. Maternal cells mixed into a prenatal sample can also complicate interpretation.

Accuracy and limitations

Karyotyping is reliable for counting chromosomes and identifying large rearrangements when the sample contains enough suitable cells. Laboratories use quality checks and may examine additional cells or cultures before reporting an unexpected result.

The test can miss small deletions, duplications, and single-gene changes because they are below microscope resolution. The exact level of detail depends on the sample, chromosome banding quality, and laboratory method. Chromosomal microarray or DNA sequencing is often better for smaller changes.

Low-level mosaicism can be missed if none of the limited number of examined cells carries the

change. Results can also be misleading if the tested tissue does not represent other tissues.

A result can appear abnormal without reflecting the main tissue of interest. Examples include culture-related changes, donor cells after transplant, maternal-cell contamination, and chromosome changes limited to the placenta. A laboratory may recommend testing another sample or using a different method.

Targeted FISH testing can answer a specific chromosome question more quickly. Microarray is better at finding small gains and losses. Sequencing is better for changes within genes. None replaces every function of the others.

Risks and complications

A blood draw may cause a small bruise, soreness, or brief light-headedness. Serious complications are uncommon.

Bone marrow sampling can cause temporary pain and bruising, with smaller risks of bleeding or infection. A biopsy has risks related to the body area sampled.

Amniocentesis and chorionic villus sampling carry procedure-related risks, including bleeding, infection, fluid leakage, and pregnancy loss. Your specialist should explain the benefits and risks for your circumstances before you consent.

After an invasive prenatal or tissue procedure, seek prompt medical advice for fever, worsening pain, fluid leakage, heavy bleeding, or increasing redness or swelling. Call emergency services for severe bleeding, collapse, difficulty breathing, or another rapidly worsening symptom.

What happens next

A typical result may complete the chromosome investigation, or another genetic test may be suggested if the original concern remains unexplained.

If a chromosome difference is found, a genetic counsellor or specialist can explain what is known, what remains uncertain, and whether testing relatives could be useful. Some findings are inherited, while others arise for the first time or occur only in cancer cells.

Routine follow-up is appropriate for questions about the report, family testing, reproductive implications, or whether another test is needed. The karyotype result itself does not create an emergency. Urgent care is needed only for significant symptoms related to the sampling procedure or another current medical problem.

Common questions

Can I drink coffee before a karyotype blood test?

Yes. Coffee does not change your chromosomes, and fasting is not needed for an ordinary blood sample. Follow separate instructions if another procedure is planned at the same appointment.

Does the time of day matter?

No. Blood for karyotyping can usually be collected at any time the laboratory accepts samples.

Can I exercise beforehand?

Yes. Recent exercise does not affect a constitutional karyotype. If you are having a bone marrow sample, prenatal procedure, or biopsy, follow the procedure team's advice.

Why does the result take weeks?

The laboratory often needs to grow living cells until they divide. Chromosomes must then be stained, photographed, counted, and reviewed by trained staff.

Can karyotyping tell me every genetic condition I have?

No. It mainly detects changes involving whole chromosomes or large chromosome sections. Smaller changes may require microarray testing, FISH, or DNA sequencing.

Will a normal result rule out a chromosome condition?

Not completely. Small changes and low-level mosaicism can be missed, and the sampled tissue may not represent every part of the body.

Sources

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