TEST

Buccal smear

A buccal smear examines cells scraped from inside your cheek for sex chromatin patterns.

WHAT IT MEASURES
Inactive X-chromosome patterns in cheek-cell nuclei

SAMPLE
Cells scraped from inside the cheek

FASTING NEEDED?
No

HOW LONG IT TAKES
A few minutes

RESULTS IN
Several days; laboratory turnaround differs

USED FOR
Supporting diagnosis of some sex chromosome differences

MAIN LIMITATION
Cannot reliably define the full chromosome pattern

Overview

A buccal smear examines cells collected from the lining of your cheek. A healthcare professional gently scrapes the inside of the cheek, spreads the cells onto a glass slide, and examines them under a microscope.

The traditional test looks for sex chromatin , usually an inactive X chromosome called a Barr body . When a cell contains more than one X chromosome, additional X chromosomes are usually condensed into structures that may be visible near the edge of the nucleus.

The pattern provides limited information about X chromosomes but does not show the complete chromosome set. Buccal smears are now used infrequently because chromosome analysis and newer genetic tests provide more detail. The term may also refer loosely to other tests using cheek cells, so confirm what the laboratory will examine.

Why the test is ordered

A buccal smear may support an assessment of a possible sex chromosome difference.

Historically, it was used when a newborn's genital appearance did not clearly indicate the underlying chromosome pattern or when physical development suggested a difference involving the number of X chromosomes.

Today, the test may provide an initial clue while more detailed chromosome or genetic testing is arranged, but it is rarely enough to make a diagnosis.

A standard sex-chromatin smear does not diagnose mouth cancer or infections, establish parentage, or provide a complete DNA profile. Those purposes require different tests, even if cheek cells are collected similarly.

Who should have the test

You may be offered a buccal smear when a specialist needs a quick, limited assessment of sex chromatin. It may form part of a broader evaluation involving medical history, physical development, hormone tests, imaging, and genetic testing.

It is not routine screening for the general population or a standard prenatal test. Depending on the clinical question, your care team may instead recommend chromosome analysis, chromosomal microarray, or another genetic test.

How to prepare

Fasting is not needed. You can usually take your regular medicines and drink water as normal.

The clinic may ask you not to eat, drink coffee, chew gum, smoke, or use mouthwash for a short time beforehand. This helps prevent food debris and other material from affecting the sample.

Tell the person collecting it if you have a painful mouth ulcer, recent dental work, bleeding, or an injury inside your cheek. They may use the other cheek or delay collection.

Follow the laboratory's instructions, especially if the sample will also be used for DNA analysis.

What happens during the test

You will usually sit upright and open your mouth. A healthcare professional uses a small spatula, brush, or similar device to rub or scrape the inside of one cheek.

Collection takes a few seconds. It may feel rough or mildly uncomfortable but should not be painful.

The cells are spread onto a glass slide, fixed, and stained. A laboratory specialist examines their nuclei for sex chromatin and checks that enough intact cells are available. Another sample may be needed if the first contains too few usable cells.

Understanding your results

The report may describe sex chromatin as present, absent, or not assessable. It may also state how many cells were examined and whether the sample was adequate.

In general, cells containing two X chromosomes may show one Barr body, while cells containing one X chromosome usually show none. More than one Barr body may be visible when a cell has additional X chromosomes. This relationship is not exact enough to establish a complete chromosome pattern.

An absent result does not prove that every cell has one X chromosome. A present result does not identify all chromosomes or explain how they affect the person's body or development.

There is no universal numeric reference range. Interpretation depends on the laboratory, sample quality, and clinical context.

An inadequate result means that too few intact cheek cells were available to answer the question. It does not mean a chromosome difference was found.

What affects the results

Although chromosome patterns are stable, several factors can make a smear unclear or misleading:

Mosaicism is an important limitation. A pattern present in blood or another tissue may not appear in enough cheek cells to be detected, or the reverse may occur.

A recent blood transfusion does not usually alter cheek-cell chromosomes, although blood contamination can complicate some tests. A stem-cell or bone-marrow transplant can complicate DNA testing because donor blood cells may mix with your cheek cells. Tell the genetics team if either applies.

Exercise, time of day, kidney function, and ordinary dietary changes do not meaningfully alter sex chromatin. Pregnancy and most medicines do not usually change the pattern either.

- Too few cells or too many damaged cells
- Food, mucus, bacteria, or blood obscuring the sample

- Recent cheek injury, inflammation, or dental treatment
- Poor spreading, preservation, or staining
- Examining too few cells
- Mosaicism, in which different cells have different chromosome patterns

Accuracy and limitations

A buccal smear can identify a visible Barr body when enough well-preserved cells are available, but it cannot reliably determine a complete chromosome pattern. Accuracy depends on collection, staining quality, the number of cells examined, and the chromosome difference being investigated.

The test may miss low-level mosaicism because it examines only a small sample of cheek cells. Debris, damaged nuclei, or staining artefacts may also resemble sex chromatin.

A normal-looking smear cannot rule out:

A karyotype shows chromosome number and general structure. Chromosomal microarray can detect smaller gains or losses of genetic material. Tests such as FISH or gene sequencing may be more suitable when a particular chromosome region or gene is being investigated.

No single test answers every genetic question. Results must be interpreted alongside clinical history and other findings.

- Small missing or duplicated chromosome sections
- Changes within individual genes
- Some structural chromosome rearrangements
- Low-level or tissue-specific mosaicism
- Differences involving hormone production or response

Risks and complications

The test has very little physical risk. Your cheek may feel mildly sore briefly, and minor surface bleeding is possible if the lining is already irritated.

No anaesthetic, injection, or radiation is involved.

What happens next

Your clinician will explain what the result does and does not show. More detailed chromosome or genetic testing may still be needed before reaching a conclusion.

An unclear or inadequate result usually leads to repeat collection or a different test. This is not an emergency.

A buccal smear result does not itself cause urgent symptoms. If testing is part of a newborn assessment and the baby develops repeated vomiting, poor feeding, unusual sleepiness, dehydration, breathing difficulty, or collapse, seek urgent medical help rather than waiting for the result.

Common questions

Can I drink coffee before a buccal smear?

Fasting is not required, but coffee can leave material in your mouth. Follow the laboratory's instructions; you may need to avoid drinks other than water briefly before collection.

Does the time of day matter?

No. Sex chromatin does not change during the day.

Can I exercise beforehand?

Yes. Exercise does not change your cheek-cell chromosome pattern.

Does a buccal smear hurt?

It may feel like firm rubbing or scraping. Discomfort is usually mild and lasts only a few seconds.

Is this the same as a DNA cheek swab?

Not necessarily. A traditional buccal smear examines stained cells under a microscope. A DNA cheek swab extracts genetic material for analysis.

Can the result determine my sex?

Not on its own. It provides limited information about inactive X chromosomes and does not fully describe chromosomes, reproductive anatomy, hormone function, physical traits, or gender identity.

Sources



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