PROCEDURE

Chorionic villus sampling

Chorionic villus sampling tests a small placental sample for specific chromosome or genetic conditions during early pregnancy.

PURPOSE
Diagnose specific chromosome or genetic conditions before birth

PROCEDURE TYPE
Diagnostic, minimally invasive ultrasound-guided placental sampling

WHERE IT IS DONE
Outpatient clinic or prenatal diagnosis unit

ANAESTHESIA
None or local anaesthetic, based on approach

HOW LONG IT TAKES
Usually 10 to 20 minutes

PREPARATION
Usually no fasting; bladder instructions and medicine review

RECOVERY
Same day; arrange someone to drive you home

MAIN RISKS
Miscarriage, bleeding, infection, inconclusive placental results

RESULTS
Rapid results in days; full testing often within two weeks

Overview

Chorionic villus sampling, or CVS, is a prenatal diagnostic procedure. It removes a very small sample from the placenta, the organ that supplies the pregnancy with oxygen and nutrients.

The sample contains chorionic villi. These tiny placental structures usually have the same chromosomes and genes as the developing baby. A laboratory can examine them for the conditions included in your agreed testing plan.

CVS is usually performed between 10 and 13 weeks of pregnancy. It provides diagnostic information earlier than amniocentesis, which is generally performed later. However, CVS cannot identify every genetic condition or physical difference, and a normal result cannot guarantee a healthy baby.

Why the procedure is performed

CVS may be offered when there is an increased chance of a chromosome or genetic condition. It can confirm or rule out a specific condition more reliably than a screening test.

Reasons you may be offered CVS include:

Common chromosome conditions tested for include Down syndrome, Edwards syndrome and Patau syndrome. If a particular inherited disorder is suspected, the laboratory may perform a targeted genetic test.

CVS does not test for all birth differences. In particular, it does not directly screen for open neural tube defects, such as spina bifida. These are assessed later using ultrasound and, in some care plans, a blood test.

- A screening test suggesting an increased chance of a chromosome condition
- An ultrasound finding that may be linked to a genetic condition
- A previous pregnancy or child affected by an inherited condition
- You or the other biological parent carrying a particular gene change
- A family history of a condition that can be tested during pregnancy

Who should and should not have it

CVS is an option rather than a requirement. It may be right for you if you want a definite prenatal result, understand what the planned laboratory test can and cannot show, and feel that earlier information would help you make decisions.

The procedure should not usually be performed before 10 completed weeks. Procedures done earlier have been linked to problems with limb development.

CVS may be delayed, avoided or replaced with another test when:

CVS can be performed in some twin or other multiple pregnancies, but sampling the correct placenta requires specialist experience.

Before consenting, ask what conditions will be tested, how reliable the result will be, what uncertain findings are possible and how the result could affect your pregnancy care.

- The pregnancy is too early
- The placenta cannot be reached safely
- There is active vaginal or cervical infection, particularly for a cervical approach

- Bleeding, uterine anatomy or another pregnancy complication increases the difficulty
- A bleeding disorder or blood-thinning medicine requires further planning
- The condition being investigated cannot be reliably diagnosed from placental tissue

How to prepare

Your treating facility's own instructions take priority because preparation differs according to the stage of pregnancy and the planned approach.

You can usually eat and drink normally. You may be asked to attend with a full bladder because this can improve the ultrasound view or help position the uterus. At other stages, you may be asked to empty your bladder.

Tell the team about all medicines and supplements you take, especially blood thinners like warfarin or apixaban . Do not stop a prescribed medicine unless the team responsible for the procedure tells you to do so.

Your team may confirm your blood group. If you have RhD-negative blood, you may be offered an anti-D injection afterward to reduce the chance of developing antibodies against RhD-positive blood.

Consider arranging for someone to accompany you and drive you home. Bring any questions about the testing and decide whether you want to know information such as fetal sex if it is revealed by the chromosome analysis and can legally be reported.

What happens during the procedure

An ultrasound is performed first to confirm the pregnancy's stage, check the baby's heartbeat and locate the placenta. The clinician then chooses one of two approaches.

For transabdominal CVS, the skin on your abdomen is cleaned. Local anaesthetic may be used to numb the area. A fine needle is guided through the abdomen and uterus into the placenta while ultrasound images are viewed continuously.

For transcervical CVS, a speculum is placed in the vagina, similar to a cervical screening examination. A thin tube is passed through the cervix into the placenta under ultrasound guidance. Local anaesthetic is not usually needed for this approach.

Gentle suction removes a small amount of placental tissue. The needle or tube does not intentionally enter the baby's body. Occasionally, more than one sample is needed if the first does not contain enough tissue.

The sampling itself usually takes only a few minutes, although the full appointment is longer because of preparation, ultrasound and checks afterward. The baby's heartbeat is usually checked again before you leave.

Recovery and what to expect afterward

Mild period-like cramps are common for a few hours. Light vaginal spotting can occur, especially after transcervical CVS. These symptoms should gradually settle rather than become stronger.

You can usually eat and drink normally immediately afterward.

Plan a quiet remainder of the day. Your facility may advise avoiding strenuous exercise, heavy lifting and sex for a short period. Follow its specific instructions rather than assuming prolonged bed rest is needed.

CVS does not usually involve sedation, but many facilities advise arranging a lift home because you may have cramps or feel emotionally unsettled. Do not drive if your facility has told you not to, or if pain or distress could affect your concentration.

Many people can return to desk-based work the next day if they feel well. You may need longer if your job involves heavy physical activity or if cramping continues.

Risks and possible complications

CVS has important limitations and risks to consider.

Miscarriage can happen after CVS. It can also happen naturally at this stage of pregnancy, making the exact procedure-related risk difficult to measure. The Royal College of Obstetricians and Gynaecologists states that the additional risk is likely below 0.5% when an experienced operator performs the procedure [1].

Bleeding, cramping or leakage of clear fluid can occur. Infection inside the uterus is uncommon but can be serious and requires prompt assessment.

Blood cells from the pregnancy may enter your circulation. This can lead to Rh sensitisation in someone with RhD-negative blood, which is why anti-D may be offered.

Rarely, the placental sample has a mixture of normal and abnormal cells. This is called confined placental mosaicism. It may reflect the placenta rather than the baby, so amniocentesis or further testing may be recommended.

The laboratory may also obtain too little tissue, find cells from the pregnant person in the

sample, or report a genetic change whose health effect is uncertain. These situations can delay or limit an answer.

Results and follow up

A rapid chromosome test may be available within several working days. It commonly looks for chromosomes 21, 18 and 13 and may examine the sex chromosomes. The exact panel is set by your laboratory and testing plan.

A fuller chromosome analysis, chromosomal microarray or targeted gene test often takes around two weeks, but complex testing can take longer. Ask whether the first result is preliminary and when the final report is expected.

Your healthcare team or genetic counsellor will explain whether the result is normal, identifies a condition, or is uncertain. A result may also show that more testing is needed. For example, amniocentesis can help clarify whether a chromosome finding limited to the placenta is also present in the baby.

Routine pregnancy care continues after a normal CVS result. This includes ultrasound assessment because CVS does not rule out structural differences, neural tube defects or conditions not included in the laboratory test.

Alternatives

You may choose a different test or no prenatal testing.

Non-invasive prenatal testing uses a blood sample from you to estimate the chance of certain chromosome conditions. It does not carry a procedure-related miscarriage risk, but it is a screening test rather than a diagnosis. It also covers fewer conditions and can produce false-positive or false-negative results.

First-trimester combined screening uses ultrasound and a blood test. It is non-invasive but gives a probability, not a definite answer.

Amniocentesis samples fluid surrounding the baby, usually from 15 weeks onward. It can diagnose many of the same chromosome and genetic conditions and can help assess open neural tube defects. It provides information later in pregnancy and also carries a small miscarriage risk.

You may decline prenatal diagnosis and continue with ultrasound care, or arrange testing after birth. This avoids an invasive pregnancy procedure but delays certainty and may reduce the

time available to prepare or consider pregnancy options.

When to call your healthcare team

Mild cramps and light spotting that steadily improve are usually part of expected recovery. Mention ongoing concerns at routine follow-up or contact the clinic during normal hours for advice.

Contact your maternity or procedure team urgently the same day if you develop:

Seek emergency care for very heavy bleeding, fainting, severe unrelenting pain, breathing difficulty or any situation in which you feel acutely unsafe.

- Increasing or severe abdominal pain
- Heavy or persistent vaginal bleeding
- Clear fluid leaking from the vagina
- Fever, chills or feeling significantly unwell
- Regular tightening or contractions

Common questions

Will I be asleep?

Usually not. You remain awake, and sedation is not normally used. Local anaesthetic may numb the skin during transabdominal CVS.

Can I drive home?

Arrange for someone else to drive if possible. Your facility may require this even though sedation is uncommon. Do not drive if you have significant cramps or feel unable to concentrate.

How painful is it?

Experiences differ. Transabdominal CVS may feel like pressure, stinging or period-like cramping. Transcervical CVS may feel similar to a cervical screening examination, followed by cramps.

When will I know the result?

Rapid results may be ready within several working days. Full chromosome or genetic testing often takes about two weeks, and specialised tests may take longer.

Can CVS tell me the baby's sex?

Some chromosome tests can identify sex chromosomes. Whether this information is tested or reported depends on the medical purpose, laboratory policy and local law.

Is CVS completely accurate?

It is diagnostic for the conditions tested, but no prenatal test is completely comprehensive. Placental mosaicism, a limited sample or an uncertain genetic finding may require further testing.

Sources

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