Communication aid

Invasive prenatal genetic testing

This communication aid has been produced for clinicians to help support and guide conversations about chorionic villus sampling (CVS) and amniocentesis with their patients.

What is invasive prenatal genetic testing?

Prenatal genetic testing can be performed to find out if a pregnancy is affected by a genetic condition. Some prenatal tests are called 'invasive' because they involve taking a small sample from the pregnancy, using a fine needle.

This aid explains two types of invasive prenatal test: chorionic villus sampling (CVS) and amniocentesis.

When can testing be offered?

CVS or amniocentesis may be offered if:

- an ultrasound scan has shown a possible difference in a fetus' development; or
- a screening test in pregnancy (such as the combined screening test) has shown a higher chance of a genetic or chromosomal condition; or
- a previous pregnancy was affected by a genetic or chromosomal condition; or
- there is a known genetic condition in the family and the genetic cause (DNA change or variant) has been identified.

About CVS

CVS involves taking a small sample of the placenta, which contains the same DNA as the fetus. The sample is taken using a thin needle, guided by an ultrasound scan. The sample contains DNA, which can be tested.

CVS is usually performed between 11 and 14 weeks of pregnancy. The procedure carries a small risk of miscarriage, around 0.5% (1-in-200).

About amniocentesis

Amniocentesis involves taking a small sample of the amniotic fluid that surrounds the fetus. The fluid contains cells from the fetus, which contain DNA that can be tested. The procedure is carried out using a thin needle, guided by ultrasound.

Amniocentesis is usually performed after 15 weeks of pregnancy. The procedure carries a small risk of miscarriage, around 0.5% (1 in 200).

Sometimes, a blood sample is also taken from the mother. This helps the laboratory check that the DNA being tested does not include the mother's DNA. This ensures the test results are as accurate as possible.

Key terms

Placenta: The organ inside the womb that supports the fetus' growth
Amniotic fluid: The fluid that surrounds and protects a fetus during pregnancy.

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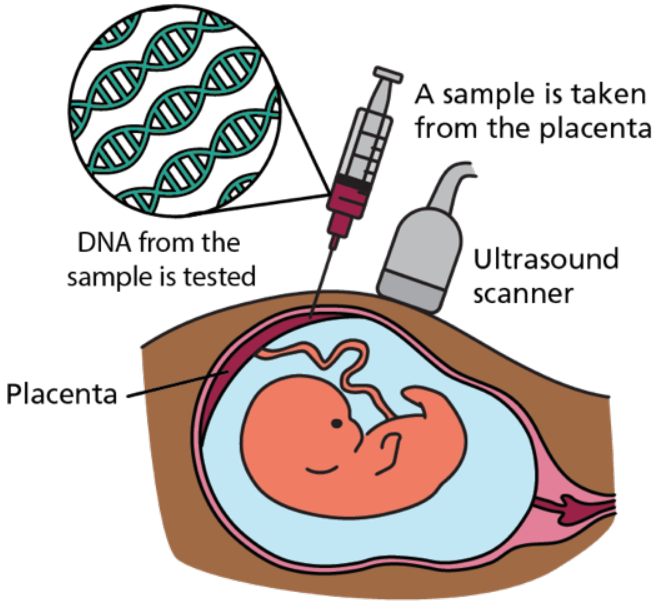
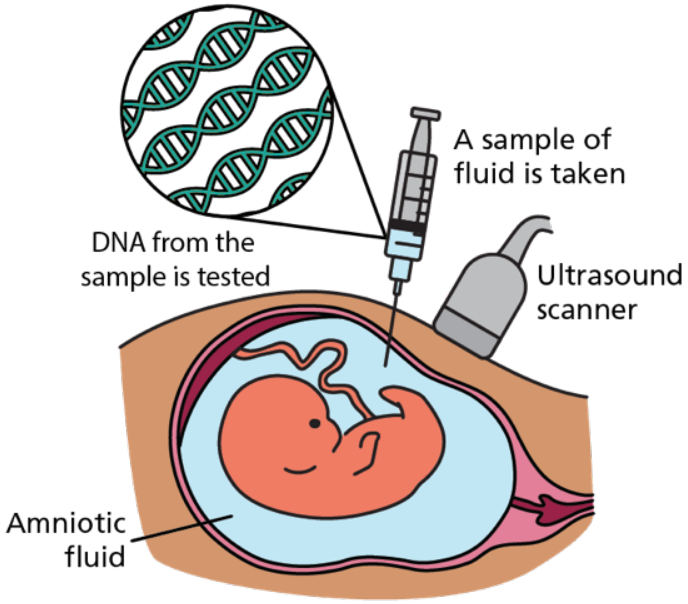


Chorionic villus sampling (CVS)
Usually between 11 and 14 weeks



Amniocentesis
Usually after 15 weeks

A sample of the mother's DNA may be needed to be sure that the CVS or amniocentesis result is from the DNA of the fetus.

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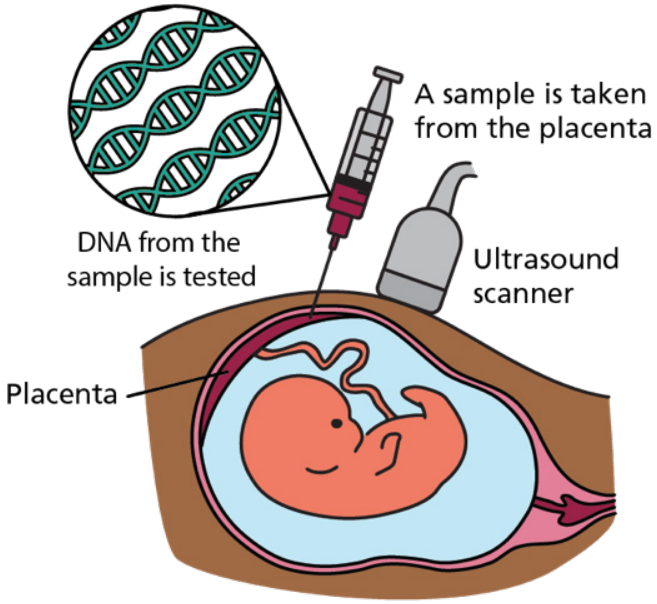

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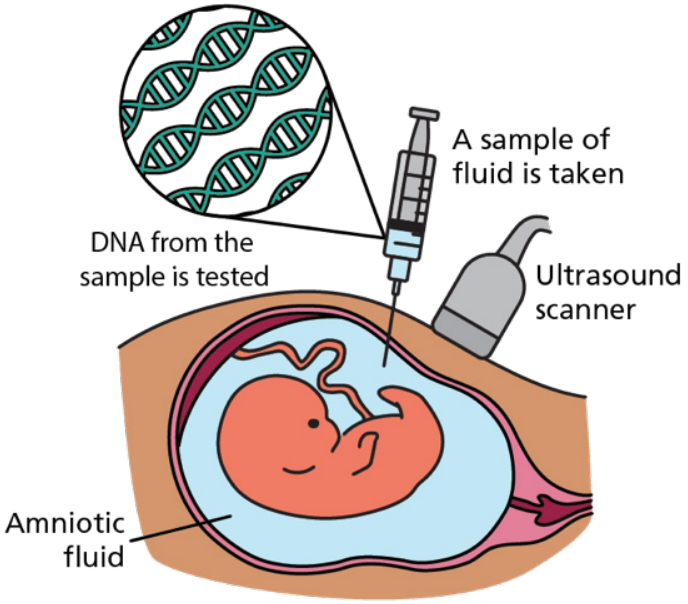

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Usually after 15 weeks

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