VCGS
Leaders in Genetic Health

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Diagnostic testing

What are diagnostic tests?

Diagnostic tests are different to screening tests. Screening tests give a risk result, while diagnostic tests are able to confirm (give a yes or no answer) that a pregnancy is affected by a certain condition. Diagnostic testing is offered when:

- There is a family history of a particular condition.
- A screening test has given a high chance result for a particular condition.
- An ultrasound has indicated a high chance for a particular condition.
- Maternal age is more than 35-37 years of age.

To perform a diagnostic test, a sample of the placenta or amniotic fluid is required. To obtain these samples, there are two diagnostic procedures available: chorionic villus sampling (CVS) and amniocentesis. Samples obtained from these procedures are sent to the laboratory for testing.

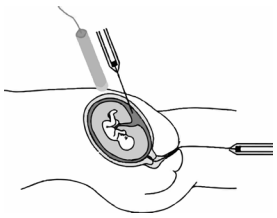
Invasive procedures

CVS and amniocentesis are invasive procedures used to collect a sample of fetal cells (containing the chromosomes).

A **CVS** is performed to collect a sample of the placenta between 11 and 13 weeks.

Under ultrasound guidance, a fine needle is inserted into the abdomen to collect a sample of the developing placenta. Occasionally, a CVS will be performed through the vagina.

A CVS is an outpatient procedure. The risk of miscarriage with a CVS is often reported as 1 in 100, but recent evidence suggests it could be as low as 1 in 500*

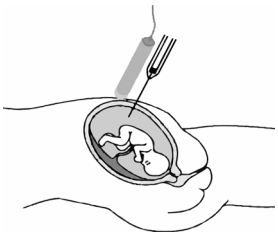


Chorionic villus sampling

An **amniocentesis** is performed to collect a sample of the amniotic fluid after 15 weeks.

Under ultrasound guidance, a fine needle is inserted into the abdomen to collect a sample of the amniotic fluid surrounding the baby.

An amniocentesis is an outpatient procedure. The risk of miscarriage associated with an amniocentesis is often reported as 1 in 200, with research suggesting it could be as low as 1 in 1000*.



Amniocentesis

Laboratory testing

These laboratory tests are considered diagnostic tests and are performed on the fetal cells collected by the CVS or amniocentesis procedures.

Karyotype: looks at the size, shape and number of chromosomes to determine if there are any extra, missing or abnormal chromosomes present. Results are usually available in 10-14 days.

FISH: stands for 'Fluorescence In Situ Hybridisation'. This test looks at whether the cells contain the correct number of chromosomes for the common conditions (chromosomes 13, 18, 21, X, Y). This is a rapid, preliminary test with results available in 1-2 working days.

Microarray (molecular karyotype): is a detailed test that provides more detailed information compared to the karyotype. This test looks for extra or missing segments of DNA in the chromosomes. Results are available in 10-14 days.

What happens after diagnostic testing?

Early FISH test results can be available in 1-2 working days, while a karyotype and/or microarray results can take up to two weeks.

If no abnormalities are detected then no further testing is required. If an abnormality is found you can choose to continue the pregnancy, or to terminate the pregnancy.

Your health care provider will offer specialist genetic counselling and refer you to a high risk pregnancy service.

Contact us

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* Akolekar.R et al. 2015. Procedure-related risk of miscarriage following amniocentesis and chorionic villus sampling: a systematic review and meta-analysis. Ultrasound Obstet Gynecol, 45:16.

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