

# Amniocentesis

## What is amniocentesis?

**Amniocentesis** is an invasive, diagnostic antenatal test. It involves taking a sample of amniotic fluid in order to examine fetal cells found in this fluid.

## Why is amniocentesis done?<sup>[1]</sup>

Pregnant women are offered amniocentesis or chorionic villus sampling (CVS) for prenatal diagnosis for a variety of reasons including a higher chance aneuploidy screening result, fetal structural anomaly, or a known risk of inherited genetic disease.

Implementation of national combined aneuploidy screening and increasing use of cell free DNA testing from maternal blood has resulted in a significant decline in screen positive results and therefore fewer invasive prenatal tests are being carried out.

However, both CVS and amniocentesis remain, at present, the only definitive diagnostic tests for aneuploidy in pregnancy. CVS, carried out to obtain placental villi for analysis, is usually performed between 11+0 and 13+6 weeks of gestation. If required, CVS can be performed between 14+0 and 14+6 weeks' gestation.

Individualised counselling of the merits of CVS versus amniocentesis should be provided for women considering CVS during this time period. A video demonstrating the recommended technique for transabdominal CVS is available online at [www.rcog.org.uk/gtg8](http://www.rcog.org.uk/gtg8).

Amniocentesis, performed to obtain amniotic fluid for analysis, is usually offered from 15+0 weeks. A video demonstrating the recommended technique for amniocentesis is available online at [www.rcog.org.uk/gtg8](http://www.rcog.org.uk/gtg8).

Informed written consent is advised prior to either procedure.

The Royal College of Obstetricians and Gynaecologists (RCOG) recommendations for amniocentesis and CVS include (see reference link for further details):

- Women should be informed that the additional risk of miscarriage following amniocentesis or CVS performed by an appropriately trained operator is likely to be below 0.5%.
- Amniocentesis should not be performed prior to 15+0 weeks' gestation.
- CVS should not be performed prior to 10+0 weeks' gestation. Where possible, to reduce the risk of technical challenges, CVS should be performed from 11+0 weeks' gestation onwards.
- Women with multiple pregnancies should be informed that the additional risk of miscarriage for twin pregnancy following CVS or amniocentesis performed by an appropriately trained operator is around 1%.
- Screening results for blood borne viruses, viral load and antigen test results should be reviewed when an invasive test is considered and individualised risk of viral transmission should be discussed.

## Genetic counselling

Genetic counselling should ideally be offered prior to any pregnancy, when there is a family history of a condition which might be diagnosed either by amniocentesis or CVS. It is clearly important to avoid unnecessary invasive testing in pregnancy where possible.

Diagnostic testing should be provided within the context of informed consent and autonomy, both about the conditions being tested and about the implications for the continuation of the pregnancy.<sup>[2]</sup> Pre- and post-test genetic counselling are both indicated.

## Indications for amniocentesis

It is important to remember that women choose whether or not to undergo amniocentesis. The list of indications becomes shorter with good pre-conceptual genetic counselling and earlier recognition of increased risk.

Most common indications for amniocentesis are:

- Positive antenatal screening tests, including for example:
  - Combined test for trisomy abnormalities.<sup>[3]</sup>
  - Abnormal fetal anomaly scan.
- A previous child with:
  - Chromosomal abnormalities.
  - Any other congenital abnormality.
- A history of:
  - Parent carrying a balanced chromosomal translocation (1 in 4-10 chance of a fetus being affected).
  - Risk of a recessively inherited metabolic disorder.
  - Mother carrying an X-linked disorder (to determine fetal sex).
- Analysis to detect specific conditions from:
  - DNA, if gene identified (for example, fragile X syndrome, sickle cell disease, cystic fibrosis).
  - Enzymatic activity in amniocytes (for example, Tay-Sachs disease).
  - Fluid biochemistry (for example, in congenital adrenal hyperplasia - 17-OH-progesterone).

Amniocentesis is also used much later in pregnancy to test for lung maturity.

## Procedure

- Most commonly performed at 15-16 weeks of gestation.
- Informed written consent should be obtained.

- Rhesus immunoprophylaxis should be given where appropriate (fetomaternal transfusion is a risk in amniocentesis and CVS).
- It should be performed under continuous ultrasound guidance.<sup>[1]</sup>
- A 22-gauge spinal needle is inserted through the maternal abdominal and uterine walls into the pocket of amniotic fluid within the amniotic sac
- 10–20 ml of fluid are aspirated (or approximately 1 ml per week of gestation).
- A cell filtration system may be used.
- Smaller volumes may be aspirated where advanced laboratory techniques require less material.
- If results are abnormal and the patient wishes it, termination of the pregnancy can be carried out at 18–20 weeks.

## Diagnostic testing of amniotic fluid

It is useful to know what is offered locally, as there is some variation between laboratories. The following tests can be performed:

- On the amniotic fluid:
  - AFP and acetylcholinesterase levels (for neural tube defects).
  - Bilirubin levels (for gestational assessment and to detect isoimmune haemolysis).
  - Tests of lung maturity (various - for example, lecithin-to-sphingomyelin ratio).
  - Enzyme analysis (many and varied, including for inborn errors of metabolism).

- On fetal cells extracted from amniotic fluid testing for genetic and chromosomal disorders:
  - Rapid testing (results in 24-48 hours).<sup>[4]</sup> In most areas this will identify specifically:
    - Down's syndrome (trisomy 21).
    - Edward's syndrome (trisomy 18).
    - Patau's syndrome (trisomy 13).
    - Turner syndrome.
    - Klinefelter's syndrome.
    - Other sex chromosome anomalies.
  - Chromosome analysis after cell culture (results take about two weeks). This will give full karyotyping but even this will not identify all chromosomal abnormalities.
- Other possible tests on fetal cells (appropriate genetic counselling may preclude the need):
  - Direct DNA analysis techniques (for example, for Tay-Sachs disease, phenylketonuria, Duchenne muscular dystrophy and cystic fibrosis).
  - Indirect DNA analysis (used, for example, to detect linkage disorders when the exact gene is not known).

Patients should be advised of how and when results of testing will be available and this may vary according to the tests being done and the laboratory used.

## **Risks and complications of amniocentesis**

- Discomfort (uterine cramping).
- Vaginal bleeding.
- Amniotic fluid leakage.

- Maternal rhesus sensitisation in susceptible pregnancies (also true for CVS).
- Amnionitis.
- Miscarriage risk:<sup>[5]</sup> <sup>[6]</sup>
  - An increased risk of mid-trimester miscarriage compared with the background risk has generally been quoted as about 0.5–1%.
  - A systematic review and meta-analysis, which included only studies published since 2000, therefore reflecting current practice, suggests the procedure-related risks are much lower at 0.1% and may even simply reflect the pregnancy characteristics of the women concerned rather than the procedure itself.
  - The procedure-related risk in twin pregnancies is higher than in singleton pregnancies and is about 1%.<sup>[7]</sup>
- Failure of cell culture from 1% up to 5% if performed under 12 weeks of gestation.
- Very experienced surgeons (more than 100 procedures per year) may have higher success rates and lower procedure-related miscarriages.<sup>[1]</sup>
- Anxiety for parents, due to lateness of diagnosis (this may make decisions about termination of pregnancy very difficult).

The small risks associated with the procedure mean that some women who are clear that they would not wish to terminate an affected pregnancy choose not to have the test.

## **Chorionic villus sampling**<sup>[8]</sup>

- Provides diagnosis in the first trimester.
- Is the technique of choice for invasive prenatal diagnosis before 15 weeks.
- Results are rapid.

- There is less risk of pregnancy loss compared with early amniocentesis (which is no longer recommended) but a greater risk of pregnancy loss compared with mid-trimester amniocentesis.<sup>[9]</sup>
- CVS before 10<sup>+0</sup> weeks of gestation is no longer performed, due to an association with fetal defects (limb reduction, oromandibular defects).<sup>[1]</sup>
- CVS is more technically demanding.<sup>[10]</sup>

Any benefits of earlier diagnosis with CVS must be carefully balanced against the slightly greater risk of pregnancy loss compared with second-trimester amniocentesis.<sup>[9]</sup> There appears to be no significant difference in long-term health outcomes between children who had transcervical CVS or amniocentesis for prenatal testing.<sup>[11]</sup>

***Dr Mary Lowth is an author or the original author of this leaflet.***

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## Further reading

- [Ogilvie C, Akolekar R](#); Pregnancy Loss Following Amniocentesis or CVS Sampling-Time for a Reassessment of Risk. *J Clin Med.* 2014 Jul 8;3(3):741-6. doi: 10.3390/jcm3030741.
- [Salomon LJ, Sotiriadis A, Wulff CB, et al](#); Risk of miscarriage following amniocentesis or chorionic villus sampling: systematic review of literature and updated meta-analysis. *Ultrasound Obstet Gynecol.* 2019 Oct;54(4):442-451. doi: 10.1002/uog.20353. Epub 2019 Sep 6.
- [Carlson LM, Vora NL](#); Prenatal Diagnosis: Screening and Diagnostic Tools. *Obstet Gynecol Clin North Am.* 2017 Jun;44(2):245-256. doi: 10.1016/j.ogc.2017.02.004.
- [Cruz-Lemini M, Parra-Saavedra M, Borobio V, et al](#); How to perform an amniocentesis. *Ultrasound Obstet Gynecol.* 2014 Dec;44(6):727-31. doi: 10.1002/uog.14680.
- [Daum H, Ben David A, Nadjari M, et al](#); Role of late amniocentesis in the era of modern genomic technologies. *Ultrasound Obstet Gynecol.* 2019 May;53(5):676-685. doi: 10.1002/uog.20113. Epub 2019 Apr 12.

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