

Prenatal testing - Invasive tests for diagnosis of specific conditions

Invasive tests are for diagnosis of specific conditions and are available only to women who are at an increased risk for having a baby with that condition. These tests are called invasive because they carry a small risk of miscarriage from the procedure. They are also called diagnostic tests. These tests are offered when:

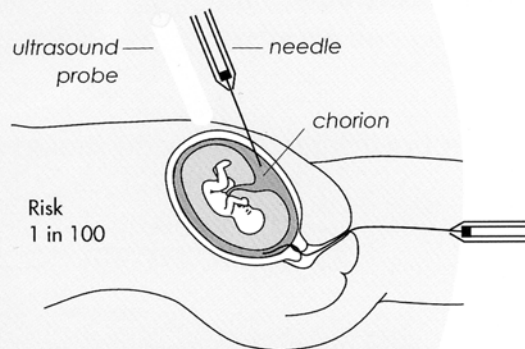
- o A woman is 37 years of age or older, or
- o There is a family history of a particular condition, or
- o Screening tests have indicated an increased risk of a particular condition, or
- o The fetal anomaly scan has shown that the risk of a chromosome abnormality is increased.

These specific diagnostic tests are done on samples of material taken from the placenta or the amniotic fluid. The samples of placenta are obtained by chorionic villus sampling (CVS). The samples of amniotic fluid are obtained by amniocentesis

Samples obtained by these procedures are then sent to a laboratory for chromosome testing for conditions such as Down syndrome and genetic (DNA) testing for conditions such as cystic fibrosis.

Chorionic Villus Sampling (CVS)

CVS is an outpatient procedure (ie you do not have to be admitted to hospital) generally done between 11 and 12 weeks of pregnancy. Under ultrasound guidance a fine needle is inserted through the woman's abdomen into the placental tissue and a sample of that tissue is withdrawn. (CVS is occasionally done through the woman's vagina). After this procedure the woman should rest quietly for the remainder of the day. A CVS is done early in the pregnancy, enabling more time for the parents to make decisions about the pregnancy. The risk of miscarriage after CVS is approximately 1% or 1 in 100 - greater than the natural risk of miscarriage at this time.. In some cases it is possible to get a preliminary result in 24 hours (FISH test) but the full results will usually take 2 weeks.

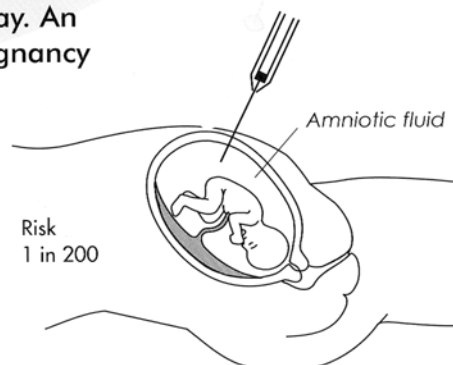


CVS

- Performed at:
- 11-13 weeks
 - 1% risk of miscarriage
 - 1% risk of CPM requiring amniocentesis to clarify
 - checks for major chromosome problems

Amniocentesis

Amniocentesis is an outpatient procedure generally done between 15 and 18 weeks of pregnancy. A sample of amniotic fluid is obtained by inserting a fine needle through the abdomen under ultrasound guidance. After this procedure the woman should rest for the remainder of the day. An amniocentesis is done later in the pregnancy but the risk of miscarriage is approximately 0.5% or 1 in 200 which is smaller than the risk of CVS. In addition, amniocentesis also provides information about neural tube defects. In some cases it is possible to get a preliminary result in 24 hours (FISH test) but the full results will usually take 2 weeks.



AMNIOCENTESIS

- Performed at:
- 15-17 weeks
 - 0.5% risk of miscarriage
 - checks for major chromosome problems
 - can help identify NTD (neural tube defects)