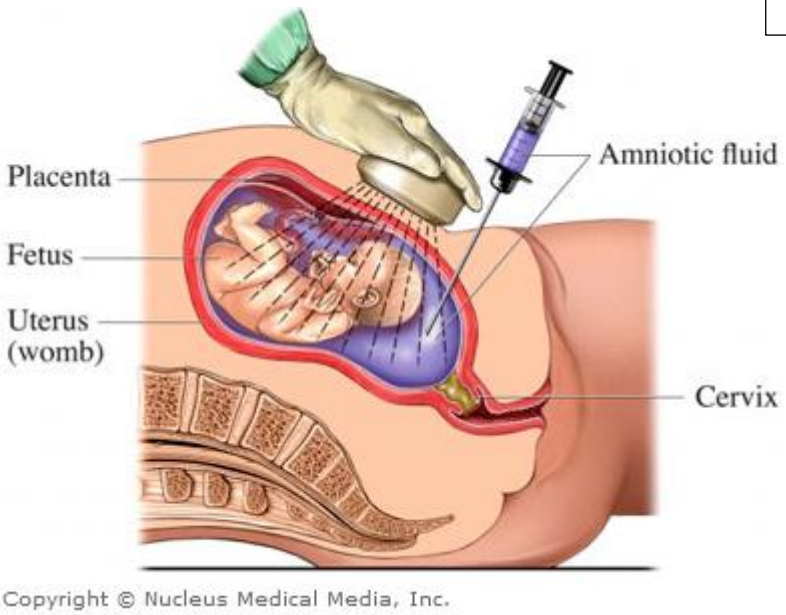
Chromosomal abnormalities : screening techniques

Genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. More than 1,000 genetic tests are currently in use, and more are being developed.

Amniocentesis



Amniotic fluid surrounds the baby during pregnancy. As the baby grows, cells from the baby are shed into this fluid. Amniocentesis is the removal of a small amount of the fluid for testing.

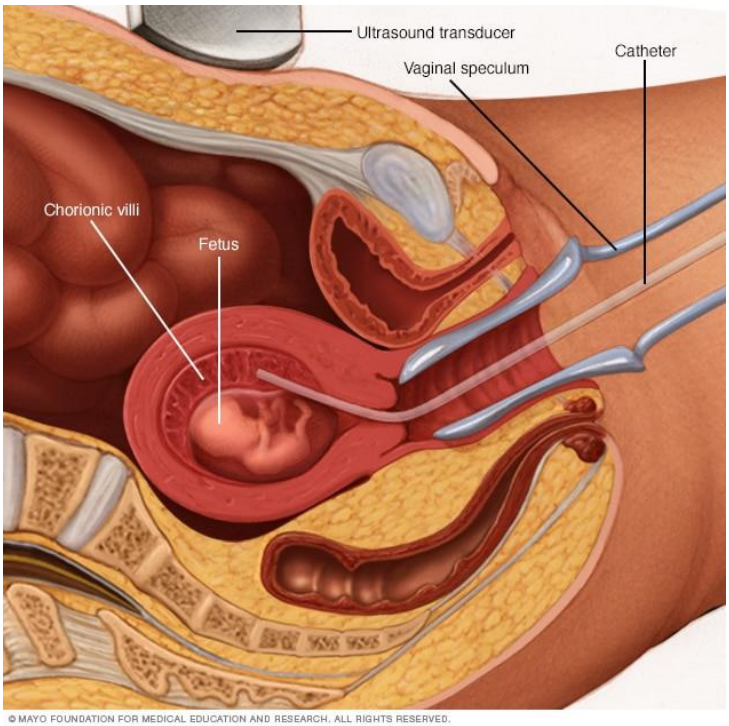
Cells found in the fluid can be used to examine the baby's genes. The test may be done if your baby has a high risk of a birth defect. They can also show if the baby is developing as expected. Depending on your risk factors, cells in the amniotic fluid are tested for: chromosome (DNA) abnormalities (number or structure), inherited genetic disease (cystic fibrosis, sickle cell disease…) , infection…

Chorionic villus sampling (CVS)

Chorionic villus sampling (CVS) is a prenatal test in which a sample of chorionic villi is removed from the placenta for testing. The sample can be taken through the cervix or the abdominal wall

The chorionic villi are wispy projections of placental tissue that share the baby's genetic makeup*. The test can be done as early as 10 weeks of pregnancy.

Chorionic villus sampling can reveal whether a baby has a chromosomal condition, such as Down syndrome, as well as other genetic conditions, such as cystic fibrosis. Although chorionic villus sampling can provide valuable information about the baby's health, it's important to understand the risks (increase of the probability of miscarriage by 0.5 to 1%) — and be prepared for the results.



Genetic makeup: genetic composition of an individual

Amniocentesis and CVS are called "invasive techniques" because they put the life of the baby at risk. Indeed, introducing needles or other tools in the uterus can lead to miscarriage.

(miscarriage = fausse couche)

Noninvasive Prenatal Testing - NIPT

NIPT is a prenatal screening, which looks at DNA from your baby's placenta in a sample of your blood to identify whether you're at increased risk of giving birth to a child with a genetic disorder. A screening like NIPT cannot, however, determine for sure whether your baby actually has a chromosomal disorder, only the *likelihood* of having that condition. But even though it can't tell for sure whether your baby has a genetic abnormality, it is highly accurate — 97 to 99 percent accuracy for three of the most common conditions.

The results of an NIPT screening can help you and your doctor decide next steps, including whether to have a diagnostic test like chorionic villus sampling (CVS) or amniocentesis ("amnio"). These genetic tests analyse a babies' own genetic material, collected from the amniotic fluid or placenta, to tell with 100 percent certainty whether a baby has a chromosome abnormality. However, they are invasive, which means they slightly increase the chance of miscarriage.