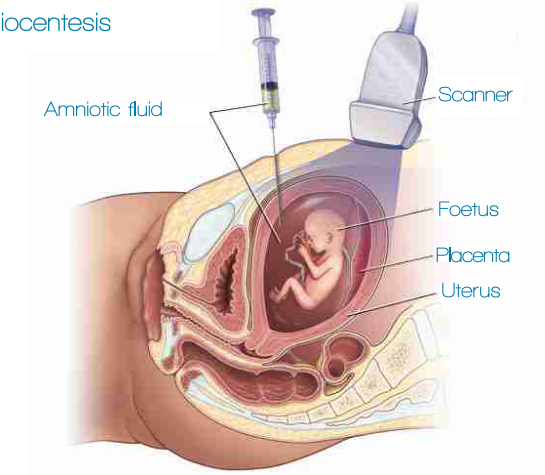
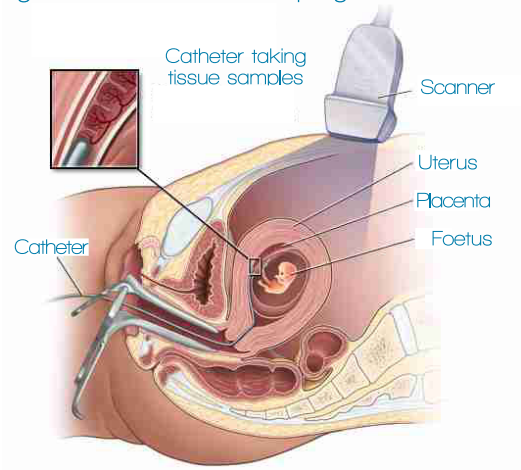
Amniocentesis



Transvaginal chorionic villus sampling



Prenatal Diagnosis

The aim of prenatal diagnosis is to detect congenital abnormalities resulting from chromosomal alterations (combined test, invasive tests) or foetal malformations (ultrasound) at an early stage. It allows the woman to decide whether to continue with her pregnancy in the event that some abnormality is found.

We currently offer what is known as the **first trimester combined test to all pregnant women,** irrespective of their age. The results of this test indicate the probability that the foetus has some type of chromosomal abnormality. When the risk is high, we recommend to confirm the diagnosis by way of so-called **invasive tests.**

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PRENATAL DIAGNOSIS

Gynaecology and Obstetrics Service

GENERALITAT VALENCIANA
CONSELLERIA DE SANITAT

HOSPITAL UNIVERSITARIO DE TORREVIEJA
DEPARTAMENTO DE SALUD TORREVIEJA

What do invasive tests involve?

They involve a sample of fluid or tissue being taken for subsequent analysis in a **chromosomal study.** The most common such tests are a chorial biopsy in the first trimester of pregnancy and an amniocentesis after week 15 of pregnancy.

We recommend such tests when the result of the first trimester combined test shows a **high risk** and in special circumstances, for example when there is an individual or family history of chromosomal abnormalities, previous pregnancies have been affected or suspected foetal disease.

What is a chorial biopsy?

This technique involves inserting a special instrument to obtain a sample of chorial villi from the placenta. This tissue has the same genetic composition as cells from the foetus. We perform this procedure vaginally, via the cervix. It is performed with ultrasound guidance. The main **advantage** of this technique is that it provides cytogenetic information at between weeks 11 and 14 of pregnancy, much earlier than amniocentesis, and with a diagnostic delay of only 2-7 days.

What is amniocentesis?

This technique involves inserting a needle through the mother's abdominal and uterine wall to extract amniotic fluid, which contains foetal cells that can subsequently be analysed to study the foetus' chromosome number (karyotype). It is performed with ultrasound guidance.

It is normally performed at between weeks 15 and 18 of pregnancy, although it can also be performed later.

What do the results of these tests mean?

These are diagnostic tests which, in most cases, allow your physician to rule out or confirm whether the foetus presents certain **chromosomal abnormalities.**

The procedure may sometimes fail due to insufficient sample or problems in the laboratory (e.g. lack of cell growth, ambiguous results, etc.).

Although, in most cases, the test results indicate the absence of any abnormality, as is the case with any prenatal test, this does not guarantee the birth of a healthy baby as it cannot be used to diagnose morphological defects or mental retardation or other defects of a congenital nature (biochemical, metabolic, etc.).

Are these procedures safe?

Although these techniques are **safe** complications may occasionally arise. For example, there is a risk of miscarriage, which varies depending on the procedure used and certain pregnancy-related characteristics, in around 1-2% of cases. There is also a risk of maternal infection and haemorrhage in the following days. In some cases, amniocentesis has been linked to premature birth, premature rupture of the amniotic sac and loss of amniotic fluid.

Are these tests painful?

In most cases they tend to cause discomfort rather than pain. After undergoing them, some women may note discomfort similar to period pains. It is also normal to feel some degree of anxiety and nervousness both before and after undergoing these tests.

We recommend that you share these feelings with your partner, family or friends, who can all be of help at such a time.

Are these tests compulsory?

These tests are completely voluntary. After receiving the appropriate information, you and your partner should assess and, together with guidance from the physician, take the decision whether or not to undergo these invasive tests.

As this is a medical examination, you must give your **consent for it to be performed** after being provided with written information. Make sure that you understand this information fully, and ask your gynaecologist if there is anything you are unsure about.

What to do after undergoing these tests...

Once you have undergone the test, we hope that you are satisfied with the treatment you have received and that the invasive technique has not been too uncomfortable for you. We recommend that you rest somewhat in the next 24-48 hours (not necessarily in bed) by avoiding physical exercise, sexual intercourse and standing for long periods of time. You may experience some abdominal discomfort, period-type pains or very light bleeding. This is relatively common and in most cases **is unimportant** as the pregnancy continues as normal. If you are not allergic, you can take one tablet of paracetamol to relieve this discomfort.

In the case of amniocentesis, you should remove the dressing in the morning before showering. If you experience continual rhythmic pain, heavier bleeding than a period, fever or loss of amniotic fluid, you should consult our unit or attend your nearest A&E Department. You will given your test results when you attend your next appointment or by telephone as soon as they are available.