

- There is small chance in about 1 in 100, that it will not give a clear result. This could be because it was not possible to analyse the sample in the laboratory or that the sample was analysed and the test gave an uncertain result (called Mosiacism)
- If the result is not clear, it may be necessary to offer you a repeat test.

## THE SUMMARY

Making a decision about having a diagnostic invasive procedure during pregnancy, can be difficult. You may be making this decision alone or with your partner. In making a decision about amniocentesis / CVS, it is important you have enough time and that you feel supported in your final decision. The final decision is yours. Only you can weigh up how much you want information about a disorder versus the slight risk that the procedure may lead to miscarrying a baby who may, or may not, have a disorder. You have the right to be fully informed about your health care and to share in making decisions about it. Your healthcare team should respect and take your wishes into account.

You need to fully read and understand the implications of this test and give a written consent to undergo. You will not know the sex of the fetus. You will be told only about an abnormality, if found.



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# PRE-NATAL DIAGNOSTIC PROCEDURES

Amniocentesis / Chorionic Villous Sampling





Why These Tests May be Necessary

Appointment helpline:

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## These tests may be necessary in pregnant women who:

- Have received a high-risk screening result from a screening test (FTS / Quadruple /Triple Test) for Down syndrome or similar problems.
- Advanced Maternal Age
- Have had a family or previous pregnancy history of a genetic disorder like Thalassemia
- Where a Fetal Structural Abnormality is found

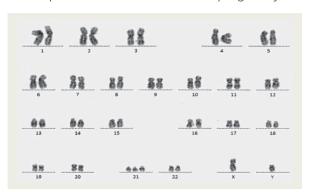
If your blood group is Rh negative, you will be advised to have an injection of anti-D immunoglobulin after the procedure to prevent you from developing antibodies against the baby's blood cells.

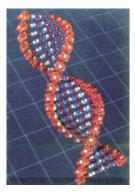
After the procedure you should rest in the clinic for at least half an hour before going home . You should take it easy the rest of the day. You could resume your normal life from the very next day.

## THE TEST

Amniocentesis is a procedure in which a small amount of the amniotic fluid surrounding the baby in the womb is removed by passing a fine needle through the mother's abdomen. Amniocentesis is performed after 15 weeks of pregnancy.

CVS is a procedure in which a small amount of fetal tissues from the placenta in the womb is removed by passing a fine needle through the mother's abdomen. CVS is performed at 11-13 weeks of pregnancy.





## THE PROCEDURE

Both Amniocentesis and CVS are outpatient procedures and will not need admission. In Amniocentesis, using an ultrasound probe for accurate guidance and to ensure a safe distance from the baby, a fine needle is pushed into skin, through abdomen and womb, under a local anaesthetic.

A small sample (15-20 mls) of the fluid surrounding the baby is removed using a syringe. This fluid is amber/yellow colour but may sometimes be stained with blood. The needle is then taken out and the baby's heartbeat is checked on ultrasound. The amniotic fluid, which contains some of the baby's cells, is sent to the laboratory for testing.

In CVS, a small amount of placental sample (Chorionic Villi) is removed using a syringe more or less in the same way.

For a very small group women having Amniocentesis/CVS, not enough fluid/chorionic tissues can be taken and the needle may need to be re-inserted.

## THE RISKS

Most women say that having amniocentesis / CVS is uncomfortable rather than painful, a bit like a period pain. Women describe a sharp stinging feeling when the needle goes in and a feeling of pressure when the needle comes out.

You may notice some cramping for a few hours afterwards. This is normal.

If you experience any unusual symptoms after the test, such as feeling shivery (as if you have flu), fluid loss, bleeding or contractions you should seek advice immediately.

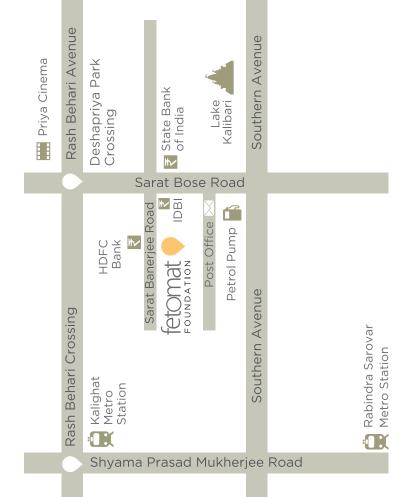
Every pregnancy carries a risk of miscarriage. Amniocentesis / CVS involves putting a needle through the wall of the womb. It may sometimes cause a miscarriage due to injury or infection in the womb. The additional overall risk of miscarriage is approximately 3 in 1000 Cores (0.3%).

### THE INTERPRETATION

For most women the laboratory test will give a definite 'yes' or 'no' answer. The result will let you know, one way or the other, whether the baby has the disorder the test was looking for.

- Many women who have amniocentesis
  /CVS will have a 'normal' result. In
  other words, their baby will be born
  without the disorder(s) the test was
  looking for.
- Some women will be informed that the baby has the disorder that the test was looking for. If the results are abnormal, these will be discussed

- fully with you. For the majority of disorders, there is no treatment or cure. You will need to consider what is best for you and the baby. This might be to:
- Terminate this pregnancy If you decide to end the pregnancy, you will be given full information about what this involves. It will depend upon how many weeks pregnant you are when you make the decision.
- Or continue with the pregnancy and use the information you have gained to help prepare for the birth and aftercare of your baby.





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### THE TEST

Congenital heart disease is a leading cause of infant morbidity and mortality from birth defects with an estimated incidence of 6 (8 to 9) per 1000 live births for moderate to severe forms. Most cases are not associated with known risk factors.

Fetal echocardiography is broadly defined as a detailed sonographic evaluation that is used to identify and characterize fetal heart anomalies before delivery.

The test is typically performed by specially trained Fetal Medicine Specialists.

A limited evaluation of the fetal heart is possible during regular obstetric scanning. However, Fetal Echocardiography is an important tool in detailed assessment of fetal heart, which is certainly recommended in many At Risk Pregnancy situations.

In Routine Low Risk Pregnancies, it can be an optional test to reasonably rule our major cardiac defect in the baby. Ultrasonography through maternal abdomen is the most common method used to evaluate the baby's heart. However, rarely Trans-vaginal Route (TVS) may be necessary.

### THE TIME

Fetal echocardiograms can be performed after 18 weeks gestation. However, if someone had previous normal Fetal Anatomy Scan, Fetal Echo is usually performed around 22-24 weeks.

### THE RESULT

At the end of the assessment, you will be told if no fetal cardiac problems are found. But, it is important to know that even in the case of a totally normal examination, not every heart problem can be ruled out by examination inside uterus. This is because the circulation in the fetus is different than after birth. Additionally, very small holes between the lower chambers of the heart are hard to see. However, considering the normal fetal circulation, your doctor can provide fairly definitive good news in the case of a normal fetal echocardiogram.

If a heart defect is found, you will be referred to a Paediatric Cardiologist for more detailed diagnosis and counselling regarding outcome, treatment and the need for heart surgery after birth.



## POINTS TO NOTE

- This test is not painful and causes no harm to the baby.
- The test can take longer than other Fetal Scans, depending on fetal position, pregnancy gestation, maternal body weight and complexity of the baby's heart problems, if any. A full bladder is not necessary for a fetal echocardiogram.
- It is always important to have as much information as possible when you
  come for your fetal echocardiogram, especially the details of why you
  were referred by your obstetrician, like any medical history in mother,
  any history of previous baby with heart problems. A four fold increase in
  congenital heart diseases is noted in fetuses of IVF pregnancies.
- There also may be a structure that is not seen as well as the doctor would like, and you may be asked to return even though the suspicion of a problem is low. Some problems such as maternal lupus may require more than one study even if the first one is normal.
- Certain heart defects may significantly increase the risk of genetic problems such as Down Syndrome or DiGeorge Syndrome. The finding of benign tumours in the heart makes the diagnosis of Tuberous Sclerosis, a genetic syndrome that has significant implications for abnormal brain development much more likely. These issues may have significant implications on the prognosis of the child and play a major role in helping you make decisions about your pregnancy. Thus, sometimes when a congenital heart disease is found, a genetic assessment of the baby may be needed, either after delivery or before by invasive testing like Amniocentesis.