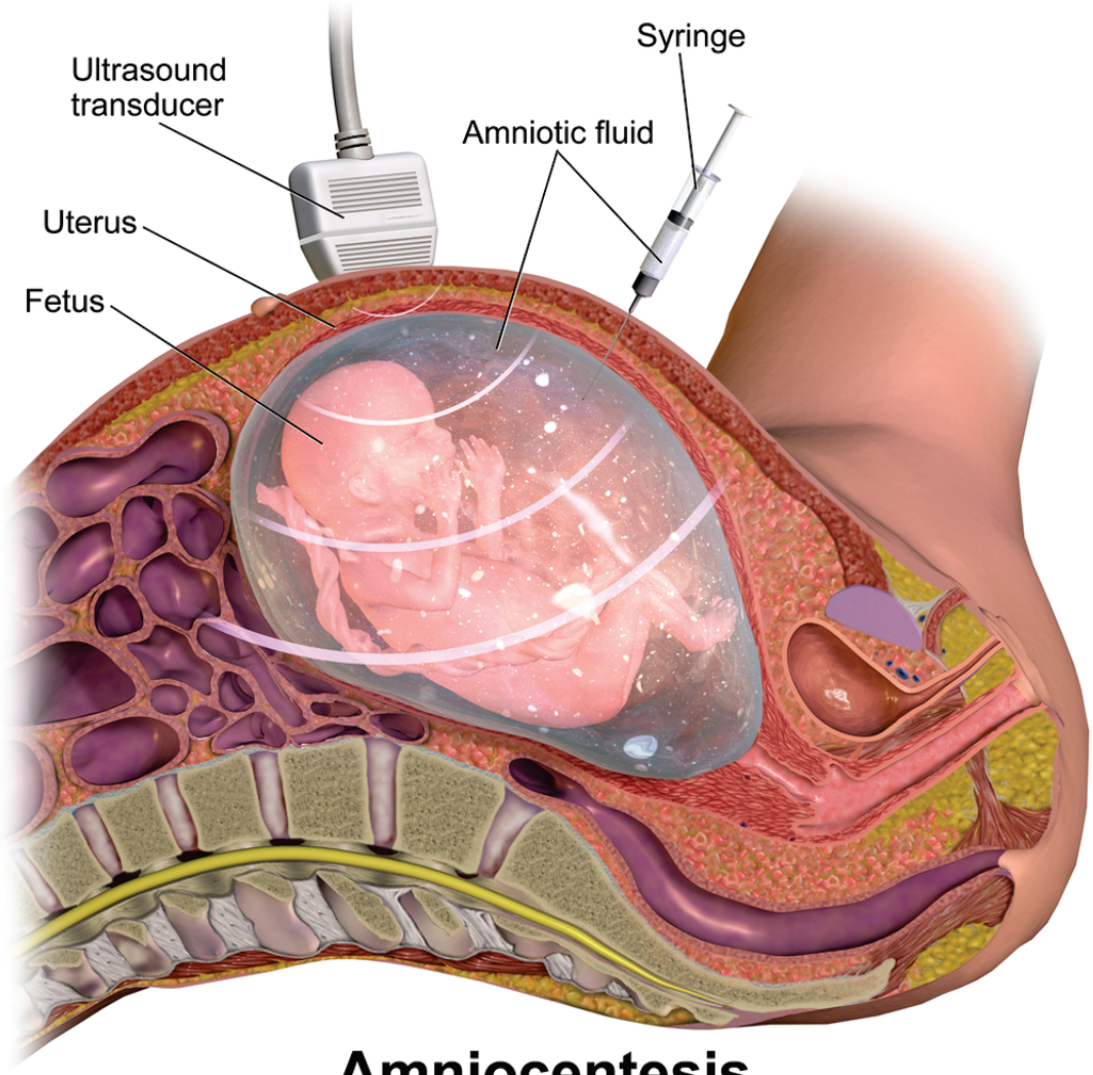

Amniocentesis

AMNIOCENTESIS

Introduction

Pregnancy can be a worrying time for some, but it is important to remember that most people have healthy babies. Certain women, however, are more likely than average to have a child with a genetic problem, and these women may be offered a procedure in pregnancy called an amniocentesis to detect certain genetic abnormalities.

Amniocentesis is a procedure in which a small amount of the fluid that surrounds the baby in the womb is taken for testing. This fluid is called amniotic fluid. It contains cells from the baby and substances from the pregnancy, which can be tested in the laboratory. The indications for doing an amniocentesis, together with the timing, safety and accuracy of the test are explained below.

What are the common reasons for having an amniocentesis?

The amniotic fluid obtained during an amniocentesis is most often used to detect chromosome disorders. Of these, Down syndrome is the most common. Amniotic fluid can also be used to detect most cases of spina bifida, a disorder in which the baby's spine does not form properly.

Other genetic disorders can also be detected, but the laboratory must be informed that there is a family history of a particular genetic problem so that specific molecular tests can be carried out.

Amniotic fluid can also be used help diagnose certain infections that your baby may have contracted in the womb. Furthermore, when an amniocentesis is done late in pregnancy, the amniotic fluid can indicate whether your baby's lungs are mature enough for your baby to survive outside of the womb.

Who can have an amniocentesis?

The final decision about having the test is yours. Amniocentesis may be offered in the following circumstances:

- The mother is 35 years of age or older at the time of conception. This is because the risk of having a baby with Down syndrome increases as a woman gets older, as shown in the table below.

Mother's age in years	Risk of having a baby with Down Syndrome
25	1 in 1250
30	1 in 900
35	1 in 300
38	1 in 150
40	1 in 90
45	1 in 25

- Women who have had a previous blood test indicating that there may be an increased risk for the baby having a chromosome disorder such as Down syndrome or another abnormality such as spina bifida.
- Couples who have already had a baby with certain medical conditions, or have a family history of specific genetic problems. A genetic counsellor must be consulted first to establish whether a particular disorder can be detected while the baby is in the womb.
- Women who have had a previous pregnancy terminated because the baby was diagnosed with a genetic problem.
- Women who have had something unusual found on ultrasound examination that may be associated with a chromosomal or chemical problem in the baby.

How is an amniocentesis performed?

Amniocentesis is an outpatient procedure, usually done at around 16 to 18 weeks of gestation (calculated from the first day of your last menstrual period). You do not have to be admitted to hospital or have a general anaesthetic. You can eat and drink normally on the day of the procedure.

An ultrasound scan is performed first, to check the stage of the pregnancy, and the position of the baby and the placenta (afterbirth). You will need a full bladder for this scan. The skin over the womb area is then cleaned with antiseptic solution, after which a fine needle is passed into the womb and a sample of the fluid (usually about 1 – 2 tablespoons) surrounding the baby is removed with a syringe. The needle is then removed and a dressing applied to the puncture site. The sample of amniotic fluid will be sent to the laboratory for analysis.

Is the procedure painful?

Most women report than an amniocentesis feels no more painful than having blood drawn or an injection. Occasionally some women feel a cramping or period-like pain during the procedure. Generally, women agree that the thought of it is worse than the actual procedure.

What happens after the procedure?

The procedure usually takes 25 to 35 minutes in total (including the ultrasound scan). Afterwards, it is suggested that you go home and rest for the remainder of the day, but you do not need to rest in bed. It is also a good idea to take things easy for a few days thereafter, and to avoid any heavy lifting or strenuous exercise. Sometimes women experience a "tightening" feeling in the womb for a short time and/or feel a little sore the next day. This is not unusual, but if you are worried about anything contact your doctor.

What are the risks of amniocentesis?

Amniocentesis is a well-established and widely available method for prenatal diagnosis. However, it is known that the procedure can sometimes cause a miscarriage. It is estimated that about 1 in every 150 to 200 women who have the procedure will miscarry as a result of it. Most of these miscarriages will occur within 2 weeks of having the procedure. You may also leak a very small amount of amniotic fluid after the procedure. This should stop within a week. Other than these uncommon events, amniocentesis is generally regarded as not being harmful to you or your baby.

When should I get medical advice?

You should contact your doctor if any of the following occur after the amniocentesis:

- You continue to leak fluid or bleed
- You have severe cramping that last for several hours
- You develop a fever (temperature higher than 38 °C) after the procedure

When do I get the results?

The chromosome test (mainly for Down syndrome) involves growing cells in the laboratory, and then analysing their genetic structure. The results are usually available within about 2 – 3 weeks. The spina bifida test is faster, and only takes about 2 days. The results will be sent to your doctor. In the event of a problem being found, the laboratory will contact your doctor by telephone and you will be offered the opportunity to discuss the findings with a genetic counsellor.

Very occasionally, the cells obtained from the amniotic fluid do not grow well in the laboratory and the amniocentesis will have to be repeated. If you are planning to be away within 2 – 3 weeks of having the amniocentesis, please inform your doctor.

What if the chromosome test results are abnormal?

If the chromosome test results show anything abnormal, your doctor will discussed the findings with you and clearly explain how the abnormality is expected to affect your baby. You can also obtain additional advice from a specialist genetic counsellor if you wish. Once you have been fully informed you and your partner can consider what the best course of action will be regarding your pregnancy.

Is the test reliable?

The chromosome analysis test is very accurate. The spina bifida test detects the majority, but not necessarily all, of pregnancies in which babies have an opening in the spinal column.

If you are having an amniocentesis for other genetic conditions, you should discuss the accuracy of the specific test that will be done with a specialist genetic counsellor.

In conclusion

After reading this pamphlet you may still have questions regarding amniocentesis. Please ensure that you raise these with your doctor or a specialist genetic counsellor before you decide to undergo this procedure.

Your doctor can also contact the laboratory at 011 358 0800 for any assistance from either the doctor-in-charge of the Cytogenetics laboratory, Dr Jennifer Rosendorff, or the specialist genetic counsellor, Dr Karen Milstein.

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