



SOUND START

*Prenatal Diagnostics
Procedures*

What is an Amniocentesis?

Amniocentesis is a diagnostic procedure typically performed after 15 weeks of pregnancy. It involves extracting a small amount of amniotic fluid, which surrounds the baby in the uterus, to analyse for chromosomal abnormalities, genetic disorders, and certain infections.



Why is this procedure performed?

This procedure is recommended for pregnancies that are greater than 15 weeks in gestation. Some of the possible reasons for being referred for an amniocentesis include:

- High risk first trimester screening – Higher than normal risk for chromosomal abnormality detected on the Non-Invasive Prenatal test (NIPT) result
- Abnormal Nuchal translucency screening ultrasound
- Having a child with a chromosomal or genetic abnormality
- One or both Parents carriers of a genetic disease
- The parents elect to have this test for peace of mind
- Suspicion of infection in the pregnancy
- Identified structural anomaly in the fetus

What preparation is required for the procedure?

Please bring your current referral with you to your appointment, along with any previous scans or test results. It is not necessary to have a full bladder, but a small bladder can be helpful in locating the uterus.

How is the procedure performed?

This procedure is performed by a subspecialist who will first look at your baby using an abdomen ultrasound probe to confirm the gestation of your pregnancy, locate the position of your baby and plan where the sample of fluid will be taken from. During amniocentesis, a thin needle is inserted through the mother's abdomen into the uterus under ultrasound guidance to withdraw a sample of amniotic fluid. The procedure is relatively quick and generally well-tolerated, though some women may experience mild discomfort or cramping. The sample is then sent to a laboratory for analysis, and results are typically available within one to two weeks.

Risks and Considerations

While amniocentesis is a safe procedure, it does carry a small risk of complications, including miscarriage (approximately 1 in 400 procedures), infection, or injury to the baby. Our team will discuss these risks with you in detail and help you weigh the benefits and potential concerns based on your individual situation.

What is Chorionic Villus Sampling (CVS)?

Chorionic Villus Sampling is another diagnostic procedure offered at Sound Start Sydney, typically performed between 12 and 15 weeks of pregnancy. CVS involves collecting a small sample of cells from the placenta to test for chromosomal abnormalities and genetic disorders.

Why is this procedure performed?

CVS typically performed on gestations that are <15 weeks where there has been an early indication of a higher-than-normal risk during the first trimester screening process.

How is the procedure performed?

1. **Abdominal CVS:** A needle is inserted through the abdomen, similar to amniocentesis, to collect placental tissue. Local anaesthetic is usually given prior to the procedure to numb the abdominal tissue before a sample is collected.
2. **Transcervical CVS:** This procedure is performed by a subspecialist when the placenta is in a difficult to access position as seen on abdominal ultrasound. A needle is inserted through the abdomen, similar to amniocentesis, to collect placental tissue.

The choice of method depends on the location of the placenta and other factors. Our team will use ultrasound guidance to ensure the procedure is safe and accurate.

Risks and Considerations

CVS carries a slightly higher risk of miscarriage compared to amniocentesis, with an estimated risk of 1 in 100 procedures. Other potential risks include infection and, in rare cases, limb defects in the baby if performed very early in pregnancy. Our experienced team will discuss these risks with you and help determine whether CVS is the right choice for you.



What to expect after the procedure?

You may experience the following symptoms after the procedure:

- Mild discomfort or bruising at the injection site
- Mild pelvic pain/cramps, however this should subside within 48 hours.
- Small amounts of spotting if you have had a

When to see a doctor?

If you experience any of the following after your procedure, please contact your GP or go to the emergency department:

- Vaginal discharge
- Severe pain or cramps
- Contractions
- Fevers

There are 2 stages that the results are returned to us and your referring doctor

1. PCR – usually back within 48 hours
2. Karyotype and microarrays – usually back within 2-3 weeks