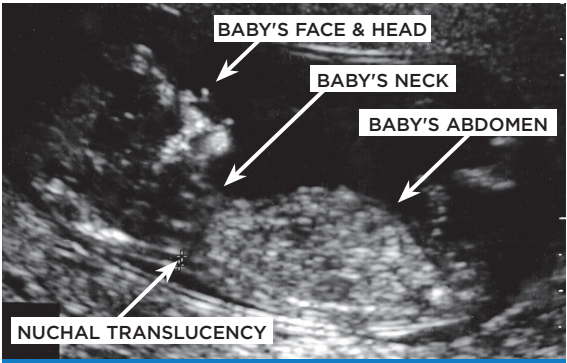
Screening Tests for your Baby in Early Pregnancy

Now Including Non-Invasive Prenatal Testing (NIPT)



Centre for Genetics Education | NSW GOVERNMENT Health

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Screening Tests for your Baby in Pregnancy

There are a number of different tests available to assess the health and development of your baby before birth. This brochure is about a number of screening tests available in pregnancy to check for some health problems. All tests during pregnancy are optional.

A screening test can help identify babies with an increased risk or chance of having some health and developmental problems.

Screening tests do not harm your baby

First Trimester Screening

Screening tests in early pregnancy are used to see if a baby is more likely to have a chromosome condition. The most well-known chromosome condition is called Down syndrome.

Down syndrome is also known as trisomy 21. It is a condition that occurs as a result of having an extra copy of chromosome 21 in the baby's cells. There are certain physical characteristics associated with this syndrome and varying degrees of intellectual disability.

- **The Nuchal Translucency (NT) Ultrasound.**

  Nuchal translucency describes a fluid-filled space at the back of a baby's neck which can be seen by ultrasound early in pregnancy. "Nuchal" (pronounced "new-cal") is a scientific word meaning "neck".

  During this test, the depth of the fluid at the back of the baby's neck is measured. The greater the depth of fluid, the greater the chance that the baby has Down syndrome or, more rarely, another chromosome condition.

The nuchal translucency measurement, along with your age and stage of pregnancy is used to give a “risk figure” or chance of your baby having certain chromosome conditions. The ultrasound is also used to look for other changes in the baby’s physical development.

- **The Combined First Trimester Screening Test (Nuchal Translucency Ultrasound and maternal blood test).**

  In addition to the nuchal translucency, a combined first trimester screening test includes a blood test for the mother. This blood test measures the level of certain pregnancy related chemicals in the mother’s blood.

  This test result, when combined with the nuchal translucency result is used to give a “risk figure” or chance of your baby having certain chromosome conditions.

When should first trimester screening be done?

The nuchal translucency ultrasound is done between 11½ and 13½ weeks after the first day of your last period. Only health professionals with special training should do this measurement.

If you are having the combined first trimester screening test, your blood is collected before the ultrasound but can also be done at the same time.

What will a first trimester screening test tell me?

Screening test results do not give a definite answer about the health of the developing baby.

The result you receive will be based on your age, your stage of pregnancy and the test measurements. It will apply to the current pregnancy only.

- **If you have the nuchal translucency ultrasound only:**

  About 75% of babies with Down syndrome and occasionally other conditions will be picked up as being at "increased risk". Therefore about 25% of affected babies will be missed.

- **If you have both the combined first trimester screening test (nuchal translucency ultrasound and the blood test):**

  About 90% of babies with Down syndrome will be picked up as being at "increased" risk. Therefore about 10% of affected babies will be missed.

What does a "low" risk and an "increased" risk result mean?

The chance of your baby having Down syndrome is given as a number as explained below. The result can mean that either:

- **The chance of a chromosomal condition is low...**

  Most babies have less than a 1 in 300 chance of having Down syndrome or another chromosome condition (ie: 1 in 500 or 1 in 1000).

  OR

- **The chance of a chromosomal condition is increased...**

A result that gives you a risk of 1 in 300 or greater (ie: 1 in 250, 1 in 100, etc) means your baby has an increased chance of having certain chromosome conditions.

This does not mean that your baby definitely has a chromosome condition, but further testing may be considered.

About 5% of women (or 1 out of every 20) screened will be given an increased risk result. It is important to note that most of these babies will not have a chromosome condition.

When will the results be available?

The result of the nuchal translucency ultrasound may be available to you immediately or will be sent to your doctor. If you have had the blood test done prior to your ultrasound, you may also get an immediate risk figure based on the combined first trimester screening test (nuchal translucency ultrasound and the blood test). However, if you have your blood collected at the same time as your ultrasound, it may be a few days before you have an answer.

Does everyone have a screening test early in pregnancy?

No. You have a choice whether or not to have the test. Some people prefer not to have information about a possible problem with their baby.

What if the baby is found to be at increased risk of having a chromosome condition?

If you have been given an increased risk of your baby having a chromosome condition, you may consider whether or not you would like to have further testing which can give a more accurate result.

Further testing may include:

1. A more accurate **screening** test for some chromosome conditions (NIPT)
2. A **diagnostic** test (Chorionic villus sampling (CVS) or Amniocentesis)

Non-invasive prenatal testing (NIPT)

NIPT is a screening test that it is more accurate for some chromosome conditions than the first trimester screening tests described above. The test can be offered from 10 weeks of pregnancy and requires a sample of the mother's blood. During pregnancy, some of the DNA from the baby (called foetal DNA) crosses into the mother's bloodstream. This foetal DNA carries the baby's genetic information. It is this foetal DNA in the mother's blood that is analysed and measured.

A negative, normal or low risk result indicates that the baby is unlikely to be affected by any of the chromosome conditions included in the screen.

A positive, abnormal or high risk result indicates that the baby is likely (but not definitely) to be affected by the specified chromosome condition.

NIPT is highly accurate for some chromosome conditions, however, since it is not a test which looks directly at the baby's cells like a prenatal diagnostic test (see below) does, the result will not be 100%. NIPT is a safe screening test and does not risk the pregnancy in any way.

If the NIPT result shows your baby might have a chromosome condition, then you may consider having a diagnostic test like chorionic villus sampling (CVS) or amniocentesis.

Prenatal diagnostic tests (Chorionic villus sampling (CVS) or Amniocentesis)

These diagnostic tests look for a specific condition that your baby might be at risk of. If you have been given an increased risk for a specific chromosome condition, the test will check the baby's cells for that condition directly.

CVS can be done around 11-13 weeks of pregnancy or you may choose to have an amniocentesis at 15-19 weeks of pregnancy.

While the CVS and amniocentesis give a very accurate result for the baby, they also can cause a miscarriage in some women (less than 1% of women will lose a pregnancy as a result of a prenatal diagnostic test).

Important points to remember:

Whether or not you decide to have first trimester screening and/or diagnostic tests, other standard pregnancy care should be followed as recommended and considered with your doctor or midwife.

Some of the tests mentioned in this brochure may not be part of standard care and you may incur personal financial costs. It is best that the cost of testing is considered as a part of your decision making before going ahead.

The importance of counselling

Counselling before having any prenatal test is important so you can discuss the implications and limitations of the result. It is also important to have information and support if you get an increased risk result.

For more information please contact:

NEW SOUTH WALES

Camperdown	Royal Prince Alfred Hospital Department of Ultrasound and Fetal Medicine Ph: (02) 9515 6042
Kogarah	St. George Hospital Women's and Children's Health KOGARAH NSW 2217 Ph: (02) 9113 3635
Liverpool	Liverpool Hospital Feto-maternal Unit LIVERPOOL NSW 2170 Ph: (02) 8738 5631
Newcastle	John Hunter Hospital Maternal and Fetal Medicine Unit NEWCASTLE NSW 2310 Ph: (02) 4921 4694
Penrith	Nepean Hospital Perinatal Ultrasound PENRITH NSW 2750 Ph: (02) 4734 2578
Randwick	Royal Hospital for Women Maternal Fetal Medicine RANDWICK NSW 2031 Ph: (02) 9382 6098
St Leonards	Royal North Shore Hospital Maternal Fetal Medicine Unit ST LEONARDS NSW 2065 Ph: (02) 9463 2370
Westmead	Westmead Hospital Fetal Medicine Unit WESTMEAD NSW 2145 Ph: (02) 9845 6802
ACT	The Canberra Hospital ACT Genetics Services WODEN ACT 2605 Ph: (02) 6174 7630

For information about the availability of testing and counselling in other areas, please contact:

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Please contact The Centre for further copies of this brochure.

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