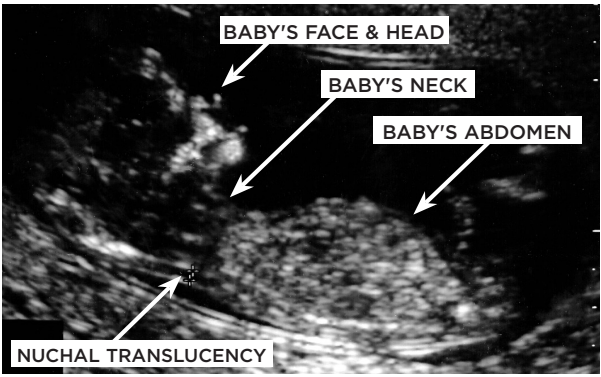
Screening Tests for your Baby in Early Pregnancy



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NSW GOVERNMENT

Centre for Genetics Education

Screening tests for your baby in pregnancy

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

A screening test may help to find out if a baby has an increased chance of having a chromosome condition. Chromosome conditions affect health and development.

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 features common in people with Down syndrome and different amounts of intellectual disability.

- **The nuchal translucency (NT) ultrasound.**

  Nuchal translucency describes the 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 thickness of the fluid-filled space at the back of the baby's neck is measured. The thicker the fluid-filled space, 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 check for unexpected 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 hormones, such as PAPP-A and HCG, in the mother’s blood.
  The blood test result, when combined with the nuchal translucency result, is used to give a “risk figure” or chance of your baby having some chromosome conditions.

- **The non-invasive prenatal test/screen (NIPT/NIPS)**
  NIPT (also known as NIPS) is a blood test of the mother which is used to measure and analyse the fetal DNA in the mother's blood. During pregnancy, some of the DNA from the baby (called fetal DNA) crosses into the mother’s bloodstream. The fetal DNA carries the baby’s genetic information.
  NIPT is a different screening test that is more accurate for some chromosome conditions, such as Down syndrome, than the other first trimester screening tests described above.

When should first trimester screening be done?

The nuchal translucency ultrasound is done between 11 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.

The NIPT/NIPS test can be offered from 10 weeks of pregnancy and requires a sample of the mother's blood.

What will a first trimester screening test result tell me?

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

The result given to you 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" or higher chance. Therefore about 25% of babies with these conditions 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 or higher chance. Therefore about 10% of babies with Down syndrome 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
  (for example, 1 in 500 or 1 in 1000).
  OR
- **The chance of a chromosomal condition is increased...**
  A result that gives you a chance of 1 in 300 or greater (for example, 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 in 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.

What will the NIPT/NIPS screening test result tell me?

- **If you have the NIPT/NIPS:**
  NIPT/NIPS is highly accurate for some chromosome conditions like Down syndrome, however, since it is not a test which looks directly at the baby’s cells like a prenatal diagnostic test (for example, chorionic villus screening or amniocentesis), the NIPT result will not be 100%.
  Results may be reported as a "low probability" or "low risk," "high probability" or "high risk" and rarely there may be no result

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 before your ultrasound, you may also get an immediate risk or chance 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.

For NIPT/NIPS it may take up to 2 weeks to get a result.

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 health condition with their baby.

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

If you have found out that your baby has a higher chance of 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 a prenatal **diagnostic test** such as:

- Chorionic villus sampling (CVS)
  OR
- Amniocentesis

Prenatal diagnostic tests: Chorionic villus sampling (CVS) and Amniocentesis

If you have been told that your unborn baby has a higher chance for a specific chromosome or genetic condition, then diagnostic tests like a CVS or amniocentesis in pregnancy may help to confirm the condition. In CVS, a small sample from the placenta is collected using a thin needle and tested. In amniocentesis a small sample from the amniotic fluid is collected using a thin needle and tested.

The diagnostic test will check the baby's cells for that specific condition as well as looking at the number of chromosomes in the cells to check for common chromosome conditions.

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, there is also a small chance that the test may cause a miscarriage in some women, in less than 1% of women (or less than 1 in 100), as a result of the 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 have a cost. It is important to speak with your healthcare provider before having any prenatal test.

The importance of counselling

Counselling before having any prenatal test is important so you can discuss whether or not you would like to find out about a possible condition in the baby during pregnancy by having the testing. It is also important to have information and support if you get an increased chance result.

For more information please contact:

NEW SOUTH WALES

Camperdown	Royal Prince Alfred Hospital RPA Women & Babies Fetal Medicine Unit Ph: (02) 9515 6042
Kogarah	St. George Hospital Women's and Children's Health KOGARAH NSW 2217 Ph: (02) 9113 3547
Liverpool	Liverpool Hospital Feto Maternal Unit LIVERPOOL NSW 2170 Ph: (02) 8738 5631
Newcastle	John Hunter Hospital Maternal Fetal Medicine 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 Maternal Fetal Medicine Unit WESTMEAD NSW 2145 Ph: (02) 8890 6802
ACT	ACT Genetics Service Canberra Health Services WODEN ACT 2606 Ph: (02) 5124 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.