

What if the baby is found to be at increased risk of having a problem such as Down syndrome?

If you have been given an increased risk of your baby having a problem, you may consider whether or not you would like a further test to see whether the baby actually has a chromosome problem.

These tests are Chorionic Villus Sampling (CVS) or amniocentesis. Such testing most often shows that the baby's chromosomes are normal. Your screening test result and options for further testing can be discussed with your doctor or genetic counsellor.

When will the results be available?

The result of the nuchal translucency ultrasound scan may be available to you immediately or will be sent to your doctor.

If you have had the special blood test done prior to your ultrasound scan, you may also get an immediate risk figure based on the two tests. However, if you have your blood collected at the same time as your scan, 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.

The importance of counselling

Counselling before having a nuchal translucency ultrasound screen, with or without the special blood test, is important so you can discuss the implications of the testing. It is also important to have information and support if you get an increased risk result.

For more information please contact:

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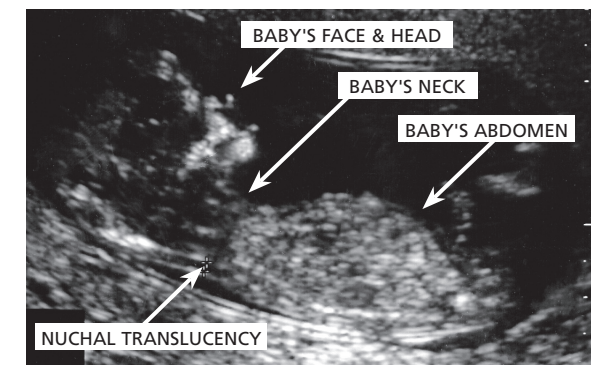
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Screening Tests for your Baby in Early Pregnancy



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What is a screening test?

A screening test can help identify babies with an increased risk or chance of having certain problems. These type of tests don't give a definite answer but may suggest the need to think about further tests.

Screening tests do not harm your baby

This brochure describes two screening tests that can be done early in pregnancy:

- an ultrasound screening test called nuchal translucency
- a special blood test

These tests are used to see if your baby is at an increased risk of having a chromosome problem.

The most well-known chromosome problem is Down Syndrome. Down syndrome is also known as Trisomy 21. It is a condition that occurs as a result of an extra copy of chromosome 21 in the body's cells. There are certain physical characteristics associated with this syndrome and varying degrees of intellectual disability.

Both tests are an option for all pregnant women, whatever their age. Discuss their availability in your area with your doctor.

The Nuchal Translucency Test

Doctors use the words nuchal translucency to describe 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 an ultrasound, the depth of the fluid at the back of the baby's neck can be measured.

The greater the depth of fluid, the greater the chance your baby has Down syndrome or, more rarely, another chromosome problem.

The nuchal translucency measurement, along with your age and stage of pregnancy is used to give a "risk figure" or chance of certain problems for your baby.

The Special Blood Test

This test measures the level of two proteins [called PAPP-A* and free-β(beta)hCG**] in the mother's blood. This test result, when combined with the nuchal translucency result, tells you the chance of your baby having a chromosome problem.

This special blood test is not offered on its own at present, but only combined with the nuchal translucency screening test.

You may choose to have the nuchal translucency screening test on its own, without the special blood test.

* PAPP-A: pregnancy associated plasma protein-A

** free-βhCG: free-beta human chorionic gonadotropin

When should the screening tests be done?

The measurement of nuchal translucency using 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 also having the special blood test, it is best done before the ultrasound but can also be done at the same time.

What will these tests tell me?

Both tests are screening tests - they don't give a definite answer. The result you receive will be based on your age, your stage of pregnancy and the test measurements. It will apply to this pregnancy only.

- **If you have the nuchal translucency ultrasound without the blood test:** About 75% of babies with Down syndrome and occasionally other problems will be picked up as being at "increased risk" by this test. Therefore about 25% of affected babies will be missed.

- **If you have both the nuchal translucency ultrasound and the blood test:** It is thought that when both test results are put together, around 80-90% of babies with Down syndrome will be picked up as being at "increased risk".

Therefore about 10–20% of affected babies will be missed.

What does a "low" 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 problem is low...** Most babies have less than a 1 in 300 chance of having Down syndrome or other chromosome problems (ie: 1 in 500 or 1 in 1000).

Please note that screening tests cannot identify all problems in babies. A congenital condition could still occur in your baby, even though the test indicated that the chance is low.

OR

- **The chance of a chromosomal problem 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 problems such as Down syndrome.

This does not mean that your baby is definitely affected.

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 the babies will not have a problem with their chromosomes.