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Important points

- Prenatal screening tests are done to see if the baby is at risk for having certain problems
- **First trimester screening** is a test in early pregnancy to see if the baby is at an 'increased risk' for having a chromosome problem, and consisting of a special ultrasound and blood test
  - The nuchal translucency test is done using ultrasound and can only be done between 11.5 and 13.5 weeks of pregnancy
  - A special test to look at proteins in the mother's blood may also be done between 10 and 13.5 weeks of pregnancy: the results of the blood test are combined with the nuchal translucency result. The blood test is not done on its own without the nuchal translucency test
- There is no risk to the baby
- Of all babies tested, a small number, about 5%, will receive an 'increased risk' result. The majority of these babies will **NOT** have a problem with their chromosomes
  - *If the nuchal translucency ultrasound test is done without the blood test:* about **75%** of babies who have Down syndrome will receive an 'increased risk' result. About **25%** of babies with Down syndrome will be missed using this test.
  - *If the nuchal translucency ultrasound result and the blood test result are considered together:* About **80-90%** of babies who have Down syndrome will have an 'increased risk' result. About **10-20%** of babies with Down syndrome will be missed using this test.
- **Maternal serum** screening test looks at the mother's blood later in pregnancy to see if the baby is at an 'increased risk' for having a chromosome problem or a neural tube defect such as spina bifida
- The test can only be done between 15 and 18 weeks of pregnancy
- There is no risk to the baby
- The result will give an 'increased risk' result for most babies who have a neural tube defect
- About 5% of babies tested will receive an 'increased risk' result for a chromosomal problem such as Down syndrome. The majority of these babies will **NOT** have a problem with their chromosomes
  - About **60%** of babies who have Down syndrome will receive an increased risk result. About **40%** of babies with Down syndrome will therefore be missed using this test

Every woman hopes for a healthy baby. In some cases, the baby may have a serious either physical or intellectual problem.

There are a number of different prenatal (meaning before birth) tests and procedures available to assess the development of the baby. Each has advantages, disadvantages and limitations.

The importance of counselling in association with prenatal testing

Counselling before any prenatal test is done, whether it is a screening test or a diagnostic test, is strongly recommended. It provides an opportunity to discuss:

- How and when the tests are done
- The advantages and disadvantages of each test
- Any risks to the baby that may result from each test
- Any further testing which may be offered and what this further testing may mean for the woman and the baby

Counselling before a test is done will help the parents decide which test, if any, is best for the woman and the baby.

This Fact Sheet discusses first and second trimester screening. Other related Genetics Fact Sheets are:

- Genetics Fact Sheet 17 – An overview
- Genetics Fact Sheet 17A – Ultrasound
- Genetics Fact Sheet 17C – CVS and Amniocentesis

1. First trimester screening

What is nuchal translucency?

Nuchal translucency is the term used by doctors to describe the appearance of a fluid-filled space at the back of a baby's neck that can be seen using an ultrasound early in pregnancy (*Figure 17B.1*).

At this early stage of the pregnancy, the baby's skin is very thin and fluid builds up in the space between the skin and the tissue at the back of the baby's neck. The depth of the fluid in this space at the back of the baby's neck can be measured using ultrasound.

The greater the depth of fluid, the more there is a chance that the baby has a problem with his or her chromosomes. A measurement which is higher than expected may indicate that the baby has a particular chromosome problem called Down syndrome (trisomy 21 - see Genetics Fact Sheet 28), or more rarely, another chromosome problem. The nuchal translucency test will give a tailored estimated chance of having a baby with these conditions.

The result is in the form of a risk number such as a chance of 1 in 100 or 1 in 500 that the baby has a problem. This number is worked out by the doctor using the thickness of the nuchal translucency and several other factors including the mother's age and stage of pregnancy.

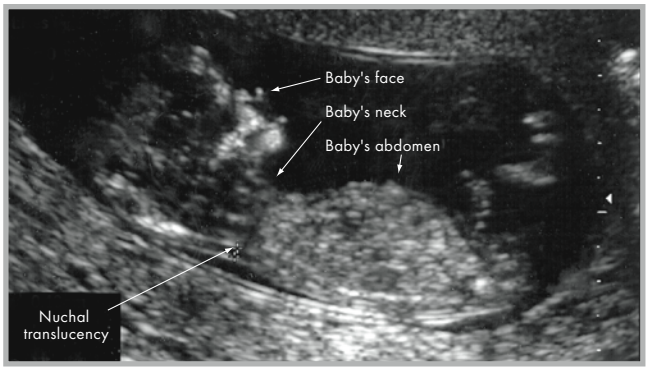

Figure 17B.1: *Photograph of an ultrasound scan taken at 13 weeks of pregnancy showing the nuchal translucency, the fluid filled space at the back of the baby's neck.*

The special blood test

This test measures the level of specific proteins in the mother's blood. This test result, when combined with the nuchal translucency result, tells the woman the chance of the baby having a chromosomal problem.

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

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

When should the test(s) be done?

The measurement of nuchal translucency using ultrasound is done between 11.5 and 13.5 weeks since the first day of the mother's last menstrual period. Only staff with special training should do this measurement.

The special blood test can be done before, or at the same time as the nuchal translucency.

What do the results of first trimester screening mean?

This is a screening test, therefore the result can indicate that either:

The chance of a problem is low

- Most babies have less than a 1 in 300 chance of having certain chromosome problems (eg. 1 in 500 or 1 in 1000)
- While the baby could still have a problem, it is less likely that this is so
- The nuchal translucency test with or without the special blood test cannot identify all problems in babies. A birth defect could still occur in a baby, even though the test result indicated a low risk

The chance of a problem is increased

A result which says that the risk is greater than 1 in 300 • (eg. 1 chance in 250, 1 chance in 100, etc) means that the baby has an increased chance of having certain problems

- This does not mean that the baby is definitely affected. Parents may consider further testing to see if their baby actually has a problem with his or her chromosomes such as Down syndrome. Such testing most often shows that the baby's chromosomes are normal
- The test result and options for further diagnostic testing can be discussed with the doctor or genetic counsellor

How reliable is the test?

The nuchal translucency test, done with or without the special blood test, is a screening test. This means that it does not give a definite answer, but only identifies those babies at increased risk of having a chromosome problem.

- About 5% of pregnant women screened (or 1 out of every 20) will be given an increased risk result. It is important to note that most of these babies will not have a problem with their chromosomes
- ***If the nuchal translucency ultrasound screen is done without the blood test:***
  - About 70%-80% of babies with Down syndrome and occasionally other problems will be picked up as being at 'increased risk' by this test
  - About 20-30% of Down syndrome babies will be missed and given a 'low risk' result
- ***If the nuchal translucency ultrasound and the blood test result are considered together:***
  - It is thought that when both test results are put together, around 80-90% of babies who have Down syndrome will be picked up as being at 'increased risk'
  - About 10-20% of Down syndrome babies will be missed and given a 'low risk' result
- The risk result the woman receives will be individually calculated, based on maternal age, how pregnant the woman is and the nuchal translucency measurement, with or without the blood test result

Does the first trimester screening test identify all birth defects?

No. The first trimester screening test does not ***identify*** most birth defects.

- Whether the nuchal translucency ultrasound test is done with or without the special blood test, chromosome abnormalities cannot be definitely identified
- The test can tell the woman and her doctor that there may be a greater possibility than average that the baby has a particular birth defect such as Down syndrome (see Genetics Fact Sheet 28)

When will the result of the test be available?

The measurement of the thickness of the nuchal translucency is combined with other factors such as the mother's age and how pregnant she is, and perhaps the result of the special blood test, to give a risk figure for the baby.

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

If the special blood test was done prior to the ultrasound scan, the woman may also get an immediate risk figure based on the two

tests. If however, the woman has the blood collected at the same time as the scan, it may be a few days before the woman has an answer.

What if the baby is at risk of having a problem?

If the baby has a higher than usual risk of having a chromosome problem (see Genetics Fact Sheet 6), the parents may consider whether or not she wants to have either a CVS or an amniocentesis test to give her a definite answer. Details about these two diagnostic tests are provided in Genetics Fact Sheet 17C.

What other tests are available during the pregnancy?

As the ultrasound scan used to measure the nuchal translucency is done early in the pregnancy, it is usually recommended that another ultrasound scan be done at 18 to 20 weeks to look carefully at the baby's physical development (see Genetics Fact Sheet 17A).

Does everyone have the nuchal translucency ultrasound test?

No. This test is optional and voluntary. Some couples would prefer not to have information about a possible problem with their baby. They may not wish to be faced with making a decision about having a diagnostic test during the pregnancy and will choose not to have the nuchal translucency test with or without the special blood test.

In Australia, the availability of the nuchal translucency test may be limited and the special blood test, in particular, may not be available.

2. Maternal serum testing: second trimester screening

What is maternal serum testing?

The maternal serum test is a screening test on the mother's blood (called maternal serum) in which the levels of special proteins produced during pregnancy are measured.

The levels of these special proteins, combined with the mother's age and other factors, can provide the woman and her doctor with an estimate of the risk that the baby has a problem with his or her chromosomes or a problem with the development of the spine.

Every woman has a risk of having a baby with a chromosomal abnormality such as Down syndrome (trisomy 21), or a problem with the development of the baby's brain and spine ie. neural tube defects (see Genetics Fact Sheets 28 & 59) .

This blood test will give the woman a tailored, estimated chance of having a baby with these types of birth defects in this pregnancy.

The risk, based on the blood test, may be higher than average. This does not mean that there is a problem in the unborn baby. Parents may, however, need to consider having further tests such as an ultrasound or an amniocentesis to determine if the baby actually has a problem. Parents can discuss these options with their doctor or genetic counsellor.

When should the test be done?

The blood test is done during the 15th, 16th or 17th completed week since the first day of the last menstrual period (that is between 15-18 weeks of pregnancy).

What do the results of second trimester screening mean?

Remember, this is a screening test. The result can indicate that either:

The chance of a problem is low

- Most babies have less than a 1 in 250 chance (eg. 1 in 300) of having certain problems
- While the baby could still have a problem, it is less likely that this is so
- The maternal serum test cannot identify all problems in babies. A birth defect could still occur in a baby, even though the blood test indicated a 'low risk'

The chance of a problem is increased

- A result which is greater than 1 in 250 (eg. 1 in 200, 1 in 100, etc) means that the baby has an increased chance of having certain chromosomal problems
  - This does not mean that the baby is definitely affected
  - Parents may consider further testing to see if their baby actually has a problem with his or her chromosomes such as Down syndrome. Such testing most often shows that the baby's chromosomes are normal
- The result may also indicate that the baby is at an 'increased risk' of having a neural tube defect such as spina bifida
  - In this case, the woman will be offered a detailed ultrasound which will examine the baby's spine very closely to see if an abnormality can be found

All test results and options for further diagnostic testing can be discussed with the doctor or genetic counsellor.

How reliable is the test?

The maternal serum test is a screening test. This means that it does not give a definite answer, but only identifies those babies at 'increased risk' of having a chromosome or spinal problem.

Most babies who have a neural tube defect will be detected using maternal serum screening alone. If maternal serum testing result is used together with a detailed ultrasound, the detection rate for spina bifida can be as high as 95% and 100% for anencephaly.

About 5% of all babies tested will receive an increased risk result. The majority of these babies will not have a problem with their chromosomes.

- About 60% of babies who have Down syndrome will have an 'increased risk' result
- About 40% of babies with Down syndrome will be missed using this test and given a 'low risk' result

The risk result the woman receives will be individually calculated risk based on maternal age, how pregnant the woman is, the levels of the hormones tested and some other factors.

Does the blood test identify all birth defects?

No. On its own, the blood test cannot definitely identify any birth defect.

It can tell the woman and her doctor that there may be a greater possibility than average that the baby has a particular birth defect such as a chromosomal problem or a neural tube defect such as spina bifida.

When will the test results be available?

Results are usually available in about one week following blood collection.

What other tests are available during the pregnancy?

Even if the maternal serum test result does not suggest a problem, it is still recommended that an ultrasound scan be done at 18 to 20 weeks to look carefully at the baby's physical development (see Genetics Fact Sheet 17A).

Does everyone have the maternal serum test?

No. This blood test is optional and voluntary.

Some couples would prefer not to have information about a possible problem with their baby. They may not wish to have to make a decision about having a diagnostic test during the pregnancy and will choose not to have the maternal serum test.

What if the test indicates that the baby has a problem?

In a small number of cases the test may show that the baby is at risk of having, a problem.

Discussing the situation with a doctor or genetic counsellor can help the parents to decide if they wish to have further diagnostic testing and which test would be best for the woman (see Genetics Fact Sheet 3).

Professional genetic counselling will provide an opportunity to discuss:

- What the result means for the parents and the family
- The options available at this time
- Whether the parents wish to have further testing if available
- The advantages and disadvantages of any further testing that is offered
- What course of action the woman will take
- The parents' attitude toward disabilities and to termination of pregnancy in a safe and understanding environment

Support will be offered to the parents at this time, no matter what they decide to do.

Other Genetics Fact Sheets referred to in this Fact Sheet: 3, 6, 17, 17A, 17C, 28, 59

Information in this Fact Sheet is sourced from:

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The Royal Australian and New Zealand College of Obstetricians and Gynaecologists [online]. Available from: www.ranzcog.edu.au. [Accessed June 2007]

Edit history

October 2004 (7th Ed)
Author/s: A/Prof Kristine Barlow-Stewart and Mona Saleh
Acknowledgements this edition: Gayathri Parasivam
Previous editions: 2002, 2000, 1998, 1996, 1994
Acknowledgements previous editions: Mona Saleh; Bronwyn Butler; Dr Warwick Giles; Dr Robert Robertson; Dr Debbie Wass; Prof Graeme Morgan