

This fact sheet describes the types of tests available during pregnancy to screen for certain genetic health and developmental problems in the baby.

In summary

- **Screening tests cannot tell if your baby definitely has a genetic condition but they might indicate that further testing needs to be considered (diagnostic testing)**
- **Regardless of your age or family health history, you can choose whether or not you have a prenatal screening test**
- **Prenatal screening tests include:**
 - ◊ **Ultrasound**
 - ◊ **Non-invasive prenatal testing (NIPT): testing of the mother's blood**
 - ◊ **First trimester (early pregnancy) screening: nuchal (pronounced new-cal) translucency ultrasound with or without testing of the mother's blood**
 - ◊ **Second trimester (mid-pregnancy) screening: testing of the mother's blood (maternal serum testing).**

Pregnancy is usually divided into three stages (trimesters). The first trimester refers to the first three months of pregnancy, the second trimester spans months four to six and the third trimester covers the final three months of pregnancy. Prenatal tests are done in either the first or second trimester of pregnancy.

PRENATAL 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. Below is a summary of the different screening tests, when they can be done, how reliable they are and also what information they provide.

It is important that you get as much information as possible before any testing is done. Discussions regarding prenatal testing options should be offered to you and your partner in a safe and understanding environment. Seeking support at this time can assist you in making informed decisions about the future of your pregnancy.

It is important to discuss the following prenatal testing issues:

- How and when the tests are done
- The advantages and disadvantages of each test
- Any risks to you or your baby that may result from each test
- Any further testing that might be offered and what it involves.

If the result of a prenatal test shows that your baby is not developing normally or could develop a problem after birth, genetic counselling will give you the opportunity to discuss:

- What the result means for your baby and your family
- The options available at this time such as further testing and what it involves
- Your thoughts and feelings towards people with disabilities
- Your thoughts and feelings towards termination of pregnancy.

TYPES OF SCREENING TESTS

First trimester ultrasound:

- Is done between 8-12 weeks of pregnancy
- Will confirm how many babies there are and how many weeks the pregnancy has progressed
- Is an important step if you are considering having testing later in your pregnancy
- Is a safe way of assessing the growth of your baby in the first trimester of pregnancy
- Does not pose any health risk to you or your baby.

Non-invasive prenatal testing (NIPT):

- Is done from 10 weeks of pregnancy onwards
- Is a screening test for Down syndrome and some other chromosome conditions
- It involves the mother having a blood test
- It provides a highly accurate risk (although not 100%) of the baby having Down syndrome and/or some other chromosome conditions
- Is a new test currently only offered through some specialist centres
- Does not pose any health risk to you or your baby.

First trimester screening test

- Can be done between 11.5–13.5 weeks of pregnancy
- Is a screening test for Down syndrome and some other chromosome conditions
- It involves a special ultrasound with or without a blood test (See *Figure 25.1*)
- The result is given to you as an estimate of the chance of your baby having a chromosome condition such as Down syndrome
- It identifies between 75-90% of babies with Down syndrome
- The ultrasound may provide further information on the development of your baby
- Does not pose any health risk to you or your baby.

Second trimester screening test:

- Can be done between 15-18 weeks of pregnancy
- Is a screening test for Down syndrome and neural tube defects
- It involves the mother having a blood test
- The result is given to you as an estimate of the chance of your baby having a chromosome condition such as Down syndrome
- It identifies around 60% of babies with Down syndrome
- Identifies 95-100% of babies with a neural tube defect
- Does not pose any health risk to you or your baby.

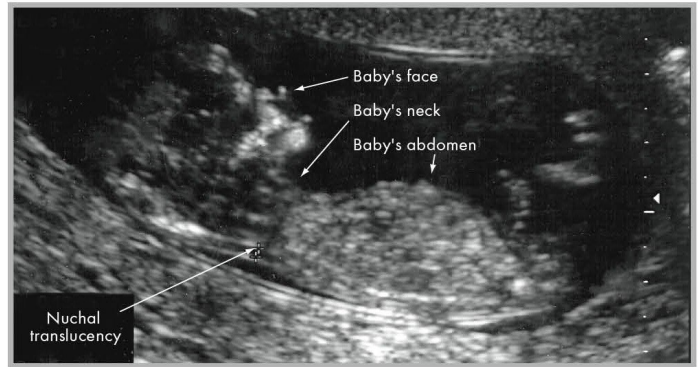


Figure 25.1: Photograph of an ultrasound scan taken at 13 weeks of pregnancy showing the nuchal translucency, the fluid filled space at the back of the baby's neck.

Second trimester ultrasound:

- Is done between 18-20 weeks of pregnancy
- Is a safe way of checking your baby's growth and development
- May identify 'soft markers' that, although do not indicate a physical problem with your baby, can increase the risk that the baby has a chromosome condition
- In a small number of cases may identify that your baby has a genetic condition or some other health or developmental problem
- Does not pose any health risk to you or your baby.

If a screening test result shows that your baby may have a problem, you and your partner will be given as much information as possible about the condition including the implications it might have for your baby's future health and development. You can also discuss whether any further testing may help you understand the situation.

It is important to note that an increased risk result does not mean your baby will definitely have a chromosome condition. Likewise, if your result is not in the increased risk category, your baby could still have a chromosome or other health condition. Your doctor, midwife or genetic counsellor can inform you about the reliability of each screening test for Down syndrome and other chromosome conditions.

Whether or not you decide to have a prenatal screening test, other standard pregnancy care should be followed as recommended and considered with your doctor or midwife.