Fact sheet **25**

SCREENING TESTS DURING PREGNANCY

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

IN SUMMARY

- Screening tests in pregnancy cannot tell if your baby definitely has a genetic condition but, they might suggest that further testing may 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 between the 4th and 6th months 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. Talking about prenatal testing options should be offered to you and your partner in a safe and understanding way. Seeking support at this time may help you with 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 pros and cons 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 in the usual way or could develop a condition, genetic counselling will give you the chance to talk about:

- 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 about disability or a health condition
- 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

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- 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
- Does not pose any health risk to you or your baby.

Non-invasive prenatal testing (NIPT):

- Is usually 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 estimate (although not 100%) of the baby having Down syndrome and/or some other chromosome conditions
- Does not provide information about the physical growth of your baby
- Does not pose any health risk to you or your baby.

First trimester screening test

- Can be done between 11—13.5 weeks of pregnancy
- Is a screening test for Down syndrome and some other chromosome conditions
- It includes 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
- When used with ultrasound, it identifies 95-100% of babies with a neural tube defect
- Does not pose any health risk to you or your baby.

Second trimester ultrasound:

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

If a screening test result shows that your baby may have a health or developmental condition, you and your partner will be given as much information as possible about the condition and what this might mean for your baby. You can also talk about whether any more testing will give you more information.

It is important to note that a result that shows that there is an increased chance that your baby will have a chromosome condition does not tell us that this is definite. Similarly, if your result shows that the chance for a chromosome condition in your baby is low, it does not mean that you cannot have a baby with a chromosome condition. Your doctor, midwife or genetic counsellor can inform you about the reliability of each screening test for Down syndrome and other chromosome conditions.

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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.