This fact sheet gives an overview of a number of ways that it may be possible to check the health of a baby before birth using a prenatal test.

In summary

- Prenatal tests are a way of checking the health of a baby before birth
- There is no test that gives a 100% guarantee of a healthy baby
- Some tests give a very accurate result about the health of the baby while others give an estimate or risk result
- Testing is always optional.

Every couple wants to have a healthy baby and for most, this will be the case. There are some couples however who will have a baby with a serious health problem. A health problem in the baby may occur for no apparent reason or it may come about due to a known risk factor.

Some of the factors which increase the likelihood of a baby having a health problem are:

- Where there is a close relative or a previous child with a serious health problem
- One or both parents of the baby has a health problem that may be passed on
- One or both parents of the baby are known ‘carriers’ of a particular faulty gene
- The mother is in her mid-30s or older (not necessarily her first pregnancy)
- There has been exposure to some chemical or other environmental factors during the pregnancy
- Where the parents are related by blood (e.g. first cousins).

WHAT ARE PRENATAL TESTS?

Prenatal tests can occur in a number of different ways and at certain stages of the pregnancy. In general, there are two main types of prenatal test – a screening test and a diagnostic test.

Screening tests give a risk or estimate of the probability that a baby has a health problem. These tests do not generally look directly at a sample from the developing baby and are therefore very safe.

Included in the group of screening tests are ultrasounds, non-invasive prenatal testing, first trimester screening and second trimester screening.

Each of these screening tests are described further in the diagram below.

Diagnostic tests provide a more accurate result since they are directly testing the baby. Because of this, some of the tests may cause a loss of the baby in a small number of cases.

Included in the group of diagnostic tests are ultrasounds, chorionic villus sampling (CVS) and amniocentesis. Each of these diagnostic tests are described further in the diagram and table below.

WHAT IF I DON’T WANT TO HAVE A PRENATAL TEST?

Testing during pregnancy is optional and should be discussed in full with your doctor, midwife or genetic counsellor. Making a decision to have a test or not is always up to you.

WHAT IF THE TEST SHOWS THE BABY HAS A HEALTH PROBLEM?

When a baby is found to have a health problem before birth, it can be a very emotional and difficult time for the parents and family. You will be given information about the health problem that has been identified and supported as you understand its impact on your baby’s health.

Depending on the stage of the pregnancy, you will be able to discuss options of planning for the birth of your baby or ending the pregnancy. Whether you decide to continue or end a pregnancy, you will be offered support and information.

IF A TEST RESULT IS NORMAL, WILL MY BABY BE HEALTHY?

There is no test during pregnancy that can give a guarantee that a baby will have no health or developmental problems. Even though diagnostic tests are more accurate than screening tests, they do not pick up every possible health problem.
PRENATAL TESTING TIMELINE

The prenatal testing timeline shows the different types of tests available at various stages of pregnancy. Each of these tests will give some information about the development of the baby. There is more detailed information about each specific test available in the fact sheets noted on the timeline below.

First trimester ultrasound
8-12 weeks of pregnancy
Fact Sheet 25

Screening Test
Non-Invasive Prenatal Testing (NIPT)
From 10 weeks onwards
Fact Sheet 27

First trimester screening test – Nuchal translucency
11.5-13.5 weeks
Fact Sheet 25

First trimester diagnostic test – chorionic villus sampling (CVS)
11-13 weeks of pregnancy
Fact Sheet 26

Second trimester screening test – Maternal serum test
15-18 weeks of pregnancy
Fact Sheet 25

Second trimester diagnostic test – amniocentesis
15-19 weeks of pregnancy
Fact Sheet 26

Second trimester ultrasound
18-20 weeks of pregnancy
Fact Sheet 25

Preimplantation genetic diagnosis (PGD)
Pre-pregnancy option using IVF
Fact Sheet 29