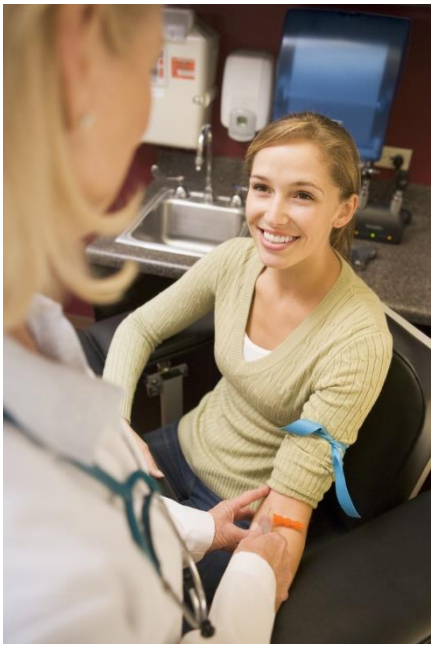
Genetic Tests in Pregnancy

Why offer genetic tests in pregnancy?

Most women are curious about their baby, and many like as much information as possible to help them make decisions. All babies carry a small chance of having a genetic condition. To help provide information, tests are available to help check that the baby is as healthy as possible.



Combined first-trimester screen

A 'combined first-trimester screen' uses the results of a blood test and an ultrasound test to provide more information about the chances a baby might be affected by a genetic condition such as Down syndrome (Trisomy 21).

The first component of the combined test is a **blood sample**. The sample is most accurate if taken at about ten weeks of gestation. It looks at two chemicals in particular – PAPP-A and free hCG – and compares the levels found to typical levels in women whose babies are not affected by Down syndrome.

The second component of a combined test is an ultrasound, usually done soon after twelve weeks. This measures the thickness of the skinfold at the baby's neck (the 'nuchal translucency') and look for the presence of the bone in the baby's nose (the 'nasal bone').

Cell-free DNA test

By ten weeks of pregnancy, almost one-tenth (10%) of all of the free DNA in a pregnancy mother's bloodstream comes from the placenta – and thus from the baby's cells. It is possible to take a blood specimen, then test for the DNA to determine whether a baby is likely to be affected by genetic conditions such as Down syndrome (Trisomy 21) and other conditions.

This is a screening test, and in a very small number of cases the results can be affected by other factors. However, it is very accurate in most cases.

Deciding to have a test – and knowing what to do with the results...

The decision whether to have a genetic screening test – or not to – is a very important one, and should not be undertaken lightly. It is important to remember that all 'screening' tests have the chance of being inaccurate. A test that suggests that a genetic condition might be present in the baby can cause stress and concern for a couple. In some cases, problems with the testing of specimens can mean that no result is available – this, too, can be very stressful and concerning.

Perhaps the most difficult aspect of testing is knowing what to do if you find out that your baby is affected by a genetic condition. It is worth thinking very carefully about what you and your partner would wish to do if you found out that your baby is affected by an important condition before having the test. Make sure you discuss this in detail with our doctor.