Pregnancy - prenatal tests

Summary

- A range of tests is available to pregnant women, including tests to confirm pregnancy, pregnancy screening tests and pregnancy diagnostic tests.
- Tests to confirm pregnancy check for the presence of a pregnancy hormone called human chorionic gonadotropin (hCG).
- It is important to note that pregnancy diagnostic tests for higher risk pregnancies are sometimes invasive and carry a small risk of complications including miscarriage.

A range of tests are available if you are pregnant. These tests can confirm your pregnancy and also monitor your baby’s development in the womb. There are also optional tests that can provide information about whether a pregnancy is affected by a range of conditions.

Regular check-ups with your doctor or midwife are an important part of pregnancy care, including information and advice about what tests you and your baby will need.

The different kinds of tests available to pregnant women include tests:

- to confirm pregnancy
- to monitor the health of the mother and the baby
- for specific medical problems (screening and diagnostic tests).

Tests to confirm pregnancy

A missed menstrual period is usually the first sign of pregnancy. Tests to confirm pregnancy detect a pregnancy hormone called human chorionic gonadotropin (βhCG).

The various tests include:

- Home test kit – these are available from pharmacies. A typical kit includes special paper that is sensitive to the presence of βhCG in urine. It is important to use a home test kit according to the manufacturer’s instructions, or false results can occur. Always confirm your results with your doctor. Pregnancy tests taken by your doctor are rarely inaccurate.
- Urine test at the doctor’s office – the doctor can test your urine for βhCG.
- Blood test – a test of the βhCG levels in your blood can be accurate within one week or so of conception.

Tests to monitor the health of the mother and the baby

Pregnancy care involves regular appointments with your doctor or midwife to monitor your pregnancy and the baby’s growth. Care includes tests that check on the health of both you and your baby, and help to identify any problems with the pregnancy.

Routine prenatal tests may include:

- blood tests at different stages of the pregnancy, such as blood group, iron levels, checks for maternal diabetes and infections
- ultrasound (first trimester) – a painless, non-invasive scan done within the first three months of pregnancy. This can be used to confirm the number of babies and helps to calculate the date you are due to give birth. This is known as your estimated due date.
- ultrasound (second trimester) – this is usually performed between 18 and 20 weeks. It is used to check the
Tests for specific medical conditions in pregnancy

Tests for specific medical conditions (including Down syndrome, neural tube defects such as spina bifida, or genetic conditions known to be present in a family) are optional. A woman may or may not wish to proceed with these tests, and may like to discuss her options with her doctors or with a genetic counsellor before making a decision.

Screening tests and diagnostic tests in pregnancy

It is important to understand the difference between screening tests and diagnostic tests. Screening tests can provide information about how likely it is that a baby might have a particular health problem, but cannot provide a definite answer. For example, results from a screening test may suggest as increased risk or low risk, or provide an estimate of risk, such as one in 1,000.

A diagnostic test provides a more definite result.

Screening tests

Screening tests can provide an estimate of how likely it is that a baby is affected by a range of conditions but cannot make a diagnosis. If a screening test suggests an increased risk, further tests are available to confirm that result.

Examples of screening tests include:

- Combined first-trimester screening – this test combines information from a first-trimester ultrasound (for example, a measurement taken from the back of the baby’s neck known as the nuchal translucency) and a blood test from the mother (first-trimester maternal serum screening) to calculate risks for a group of chromosome problems, including Down syndrome.
- Second-trimester maternal serum screening – this is a maternal blood test that helps to determine the risk of some conditions that may affect the unborn baby, such as chromosome abnormalities (including Down syndrome) or neural tube defects (such as spina bifida).
- Non-invasive prenatal testing (NIPT, also known as cell-free DNA screening) – this is a relatively new blood test that looks at the baby’s genetic material (DNA), which can be found in the mother’s bloodstream. It can provide information about the risk of conditions such as Down syndrome and some other chromosome problems, and can be done any time from 10 weeks into the pregnancy. It is more accurate than combined first-trimester screening or second-trimester maternal serum screening, but it is more costly than the screening tests.

Diagnostic tests

Some pregnancies are known to have an increased risk for a particular condition. For example, there may be a family history of an inherited disorder, or the mother may be older (we know that the risk of having a baby with a chromosome problem such as Down syndrome increases with age).

In other cases, a pregnancy screening test may have suggested an increased risk for a particular condition. A diagnostic test can provide more definite information about whether a condition is present. Some of these diagnostic tests use a needle to collect a sample of placenta or amniotic fluid and carry a small risk of complications including miscarriage.

Diagnostic tests include:

- ultrasound – may be used to check the health of the baby in the case of unusual pregnancy symptoms, such as vaginal bleeding or lack of fetal movement
- chorionic villus sampling (CVS) – a test that checks for specific medical conditions by taking a sample of the placenta. The placenta was originally formed from the same cells as the baby, so testing a small sample of placenta can provide information about the baby. To collect the sample, the doctor inserts a slender needle
through the mother’s abdomen. The needle is guided with the help of ultrasound. The tissue (known as a chorionic villus sample) is then examined in a laboratory.

- amniocentesis – a doctor inserts a slender needle through the abdomen to withdraw a small amount of amniotic fluid, the fluid which surrounds the baby in the womb. This fluid sample contains some of the baby’s cells which have been shed as the baby grows, and these are then examined in the laboratory.

Making decisions about diagnostic testing in pregnancy

About one woman in 20 is told that there may be a complication in her pregnancy. As screening tests cannot provide definite information, many women who have an increased-risk result on a screening test will go on to have a normal pregnancy.

The decision to have further tests to confirm whether an abnormality is present is up to the woman and her family after discussion with her doctor or midwife.

Diagnostic tests may be considered for a range of reasons. For example, if a diagnostic test confirms the presence of a medical condition:

- Some abnormalities can be surgically corrected while the baby is still in the uterus.
- A woman and her baby may need specialist care before, during and after at the birth.
- Knowing in advance that the baby has particular special needs may provide a family with time to prepare.
- Depending on the condition identified in diagnostic testing, some women or families may decide not to continue a pregnancy and arrange a termination of pregnancy (abortion).

Where to get help

- Doctor
- Midwife
- Obstetrician
- Genetic counselling services – available at many large metropolitan public maternity hospitals, or call Victorian Clinical Genetics Services Tel. (03) 8341 6201.
- Paediatrician
- Local hospital maternity service
- Pharmacist
- Family planning clinic
- Family Planning Victoria Tel. (03) 9257 0100 or 1800 013 952
- The Maternal and Child Health Line (24 hours, 7 days) Tel. 132 229
- NURSE-ON-CALL Tel. 1300 60 60 24 – for expert health information and advice (24 hours, 7 days)

Things to remember

- A range of tests is available to pregnant women, including tests to confirm pregnancy, routine tests to monitor the health of the mother and the baby, and tests for specific medical conditions.
- Tests to confirm pregnancy check for the presence of a pregnancy hormone called human chorionic gonadotropin (hCG).
- It is important to note that pregnancy diagnostic tests for higher risk pregnancies are sometimes invasive and carry a small risk of complications including miscarriage.