Prenatal tests

Your Obstetrician/Gynaecologist will request some tests to check that you are healthy. These include the following:

Routine tests

- Your ABO and rhesus blood group, to determine whether your baby is at risk of anaemia or jaundice (if there is an incompatibility between your blood group and that of the baby's father);
- Your haemoglobin levels, to determine whether you are anaemic;
- Whether there are antibodies against rubella (German measles), HIV, hepatitis B and syphilis, to determine whether your baby is at risk of a congenital infection;
- Your blood glucose levels (in some cases), to determine whether you are at risk of diabetes (which might be underlying, or brought on by the pregnancy);
- Your blood pressure, to determine whether you might be developing a condition called pre-eclampsia (high blood pressure brought on by the pregnancy);
- Your urine, to determine whether you have a bladder infection, or whether there is protein in your urine, which might also indicate pre-eclampsia;
- Whether you are a carrier of group B streptococcus, a bacterium which might be present in your vagina and could give rise to a pneumonia/meningitis in the baby (if attempting a normal vaginal delivery).

It is also possible to do some tests to determine whether your baby is at risk for certain physical or mental problems. These tests include prenatal screening tests (which give an assessment of risk) and prenatal diagnostic tests (which give a more conclusive answer on whether a baby, which is at risk of some problem, is definitely affected or not). The prenatal screening and diagnostic tests are optional, but provide valuable information for the management of the pregnancy.

It is important to realize that no test or combination of tests can ever guarantee a normal baby. On the other hand, the vast majority of babies are normal (even if no screening test had been done!). It is also important to make sure that your doctor is aware of any family or personal history of medical problems that might have an effect on the baby.

Screening and diagnostic tests

Screening tests are tests done on everyone. A screening test does not give a definitive answer about whether a condition is present or absent, but gives an indication of the likelihood (low, intermediate, or high risk). The majority of afflicted individuals (e.g. foetuses with Down syndrome) should be in the "high risk" group. The proportion of afflicted individuals that is detected by the test is called the sensitivity. The proportion of the normal individuals that is falsely labelled as high risk is called the false positive rate. Ideally, a screening test would have a high sensitivity (although it is never 100%), and low false positive rate (although it is never 0%).

A diagnostic test is a test done on someone at risk of a condition, and gives a definitive answer about whether the condition is present or absent. A diagnostic test has some features that makes it unsuitable for use as a screening test, such as risk or cost.

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The following screening tests are available in the three trimesters of pregnancy:

First trimester tests

The main aim is to detect foetuses at high risk of Down syndrome. Other benefits include detecting foetuses at risk of a chromosomal abnormality other than Down syndrome, dating the pregnancy accurately, detecting twins (or higher order multiple pregnancies) and diagnosing a miscarriage or some major foetal abnormalities.

Early combination test

The recommended first trimester screening test is the early combination test. This consists of a blood test (PAPP-A and free b-HCG) done at 8-12 weeks, followed by an ultrasound examination ("simple NT scan") done at 12 – 13 weeks. The name "NT" refers to the measurement of the nuchal translucency thickness, a patch of fluid behind the foetus' neck which is thickened in most foetuses with Down syndrome. The simple NT scan should be done by a practitioner (your obstetrician or someone else) who is accredited by the Fetal Medicine Foundation (the list of practitioners is available on https://fetalmedicine.org/lists/map/certified/NT) or an experienced/competent Obstetrician in NT scanning. The early combination test has a sensitivity of 85% for Down syndrome and 5% false positive rate.

If the early combination test demonstrates a low risk (<1:1000), no further testing is recommended. If it indicates a high risk (>1:100), a diagnostic test such as amniocentesis or chorionic villus sample (see below) is offered. If an intermediate risk (1:100 – 1:1000) is found, the possibilities would include a diagnostic test or further screening with an extended NT scan or cell-free DNA testing (see below).

Alternative screening tests

Alternatives to the early combination test in the first trimester include:

- First trimester biochemical screening, which entails the blood test for PAPP-A and free b-HCG which is done as part of the early combination test. Ultrasound examination is still needed to determine the pregnancy duration accurately and exclude problems such as a major fetal abnormality, multiple pregnancy or miscarriage. This has a 60% sensitivity and a 5% false positive rate.
- Simple NT scan only without biochemical screening. This has a 75% sensitivity and a 5% false positive rate.
- Extended NT scan, which entails a similar procedure to the early combination screen, but with a more detailed ultrasound evaluation including evaluation of the nuchal translucency and nasal bone, as well as the foetal blood flow. There are fewer practitioners accredited for this (https://fetalmedicine.org/lists/map/certified/DV). The sensitivity for Down syndrome is 90% and the false positive rate 2.5%.
- Cell free DNA testing, also called NIPT (non-invasive prenatal testing). This test is very accurate (>99% sensitivity and <1% false positive rate), but currently very expensive. It can be performed any time after 10 weeks, but also requires an ultrasound examination beforehand to exclude problems such as a major fetal abnormality, multiple pregnancy or miscarriage.

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In general, what you pay is what you get: the more expensive the test, the higher the sensitivity and the lower the false positive rate.

Diagnostic tests

A chorionic villus sample (CVS) can be done to confirm or refute the presence of Down syndrome, other chromosomal anomalies or specific genetic conditions. It entails aspirating cells from the placenta by putting a needle under ultrasound guidance into the womb. There is a risk of causing a miscarriage of around 1/200.

Second trimester tests

Anatomy scan (foetal anatomical survey)

The main aim is to detect foetuses at risk of physical problems. Other benefits include detecting foetuses at risk of a chromosomal abnormality (if this had not been done in the first trimester) and detecting pregnancies at risk of preterm delivery. Not all abnormalities can be detected. As a general rule of thumb, about 50% of serious abnormalities would be detected by this ultrasound examination (and 50% not). Some would not be detected because it is not easily seen on ultrasound and some because the conditions only develop later in pregnancy. Factors which influence the accuracy of this ultrasound examination include the training and expertise of the operator, the quality of the ultrasound unit, and factors which influence the ability to see the foetus on ultrasound, such as the mother's build, abdominal wall scarring, position of the foetus (or foetuses), pregnancy duration, amount of amniotic fluid and the position of the placenta.

Specialized foetal assessment

This is a more detailed ultrasound evaluation which is performed by a maternal-fetal specialist, or other practitioner with special training and expertise in diagnostic ultrasound. This is usually done where prior tests (screening tests or factors in the history) suggest a high likelihood of a foetal problem.

Screening for Down syndrome

Screening for Down syndrome is less accurate in the second than in the first trimester (with the exception of cell-free DNA testing, which is the same whenever it is done). If Down syndrome screening was not done in the first trimester, it can be done by means of:

- Second trimester biochemical screening (a blood sample to test AFP, HCG and estriol which is done between 15 and 19 weeks). This has a 65% sensitivity and a 5% false positive rate.
- Anatomy scan; which has a 40% sensitivity and a 5% false positive rate, or
- Genetic sonogram; a more detailed ultrasound examination to examine the ultrasound markers of a foetal chromosomal abnormality. This has a 75% sensitivity and a 10% false positive rate

Maternal serum Alpha-Feto-Protein (MS-AFP)

MS-AFP is a blood test done between 15 and 19 weeks. It is advisable to help detect foetal spina bifida (open spine), unless the practitioner who performs the anatomy scan is a well trained expert. MS-AFP can detect 80% of foetuses with spina bifida with a 5% false positive rate.

Diagnostic tests

Foetal cells can be obtained from the amniotic fluid by inserting a needle under ultrasound guidance into the amniotic cavity (amniocentesis) or from foetal blood by inserting a needle under ultrasound guidance into the umbilical cord (cordocentesis). These tests respectively have a 1:200 and 1:100 risk of causing a miscarriage.

Third trimester tests

Screening ultrasound ("growth scan")

The main aim of the growth scan is to detect foetuses that are not growing well. Other benefits include structural foetal assessment (to detect physical problems that only develop late in the pregnancy). The growth scan would usually be done by your obstetrician.

Specialized foetal assessment

This is a more detailed ultrasound evaluation which is performed by a maternal-fetal specialist, or other practitioner with special training and expertise in diagnostic ultrasound. Common reasons are to determine the cause of problems developing late in pregnancy (such as decreased fetal growth or increased amniotic fluid volume), or to evaluate a pregnancy at risk for foetal abnormalities (due to factors such as the family history, maternal medical problems or adverse events in the pregnancy.)

Diagnostic tests

The second trimester diagnostic tests (amniocentesis and cordocentesis) can be done in the third trimester as well.

Genetic counselling

These options can be confusing. If you require more information, and especially if you have a family history of genetic conditions, a session with a genetic counsellor can be arranged.

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Glossary

Sensitivity	The chance of detecting an abnormality if it is present (i.e. a 60% sensitivity for Down syndrome means that out of 10 babies with Down syndrome, 6 would show up as high risk, and 4 would show up as low risk)
False positive rate	The chance of a high risk result if the baby is normal (e.g. a 5% false positive rate would mean that 1 in 20 of normal pregnancies would be labelled as high risk)

Planning

I confirm that I have read the information, understand it and have had the opportunity to discuss my questions. I opt for the following:

Test	Yes	No	If required
First trimester biochemical screening			
Simple NT scan			
Extended NT scan			
CVS			
Cell-free DNA test			
Second trimester biochemical screening			
MS-AFP			
Anatomy scan			
Second trimester detailed foetal assessment			
Amniocentesis			
Cordocentesis			
Growth scan			
Third trimester detailed foetal assessment			
Genetic counselling			

Signed at.....on.....

Patient (name).....signature.....

Witness (name).....signature.....

Doctor (name).....signature.....

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