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Dr Christopher talks about:
ANTENATAL RISK ASSESSMENT

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Antenatal Risk Assessment

What does antenatal risk assessment involve?

Down syndrome is the commonest chromosomal abnormality and its risk increases with increasing maternal age. As the aim of antenatal risk assessment (screening) is to identify women at high risk of having a Down syndrome baby and to then offer them invasive diagnostic testing using chorionic villus sampling (if < 13 weeks gestation) or amniocentesis (if > 15 weeks gestation), all women regardless of age are offered screening. These non-invasive screening tests are designed to identify women in whom a risk of Down syndrome is sufficiently high to justify invasive testing (as mentioned above) and to minimise the risk of miscarrying a healthy baby.

When is it usually performed?

There are several tests available. The most commonly offered is the OSCAR first trimester screening which involves a blood test for pregnancy-associated plasma protein-A and human chorionic gonadotropin. This is combined with an ultrasound scan to measure the space at the back of the baby's neck (nuchal translucency) and is performed between 11 to 13 weeks and 6 days. This measurement and blood tests are combined with the mother's age to calculate the risk of having a baby with Down, Edwards and Patau syndromes. If the risk level is low, first trimester screening can be of much relief and offer reassurance of a healthy pregnancy.

There are other newer tests called Non-Invasive Prenatal Test (NIPT) which are more sensitive and specific at screening for Down, Edwards and Patau syndromes but they cost more. They can even screen for certain sex chromosome abnormalities, triploidy, mircodeletions, DiGeorge syndrome, Angelman syndrome, Cri-du-chat syndrome and Parder-Willi syndrome. The sex of the baby can also be determined. The DNA of the baby can be detected by performing a simple blood test on the mother from 9 weeks into her pregnancy as some of the fetal DNA crosses into the mother's blood stream. It takes 7-10 days for the results.

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If the risk of a particular genetic defect is found to be high, what is the next step?

As the OSCAR and NIPT are all non-invasive screening tests, they can only tell us whether the baby is at high or low risk of having a chromosomal abnormality. Women who receive a high risk result will be offered confirmatory diagnostic invasive testing like amniocentesis or chorionic villus sampling (CVS).

Amniocentesis is a procedure in which a small amount of the amniotic fluid surrounding the baby in the womb is removed by passing a fine needle through the mother's abdomen. It is usually safe to perform this invasive procedure after 16th week but before 20th week of pregnancy. About 0.3-0.2% of women who have amniocentesis from 15th week of pregnancy under ultrasound guidance miscarry as a result of the procedure. It takes a few days to about 3-4 weeks for the results to be available. If a disorder is diagnosed, genetic counselling is offered.

In CVS, cells are taken from the placenta called chorionic villi and are sent to a lab for genetic analysis. The main advantage of CVS over amniocentesis is that it can be performed earlier (between 11 and 12 weeks of pregnancy) so you can find out about your baby's condition sooner. The miscarriage rate associated with CVS is higher (between 0.5 to 1%). Results are usually available in a few days to about 7 to 10 days.