Screening for Down's syndrome

Dr Deepti Gupta
MBBS, MS (OBS & GYNAE), MRCOG (UK)
Consultant Gynaecologist & Laparoscopic Surgeon

WHAT IS DOWN'S SYNDROME?

Statistics show that 1 in 700 babies have Down's syndrome, which is caused by the presence of an extra copy (three instead of the normal two) of chromosome 21 in each cell. This condition is referred to as "trisomy 21"
Down's syndrome is one of the most common causes of mental disability and can also be associated with congenital heart abnormalities or other physical abnormalities. There is no cure for Down's syndrome. It will affect someone throughout their life.

WHO SHOULD BE SCREENED?

Anyone can have a baby with Down's syndrome but a woman's risk increases as she gets older. The risk faced by 35-year-old women is about 1 in 350, but this risk increases exponentially after this age, rising to as high as 1 in 100 for 40-year-old women.

If you have previously had a baby with Down's syndrome, your risk of having another affected baby is increased. About 1 in 100 women with a previous baby with Down's syndrome will have another.

WHAT SCREENING TESTS ARE AVAILABLE?

Screening can take place in the 1st or second trimester of pregnancy. First-trimester screening consists of two parts: a blood sample and an ultrasound measurement of the thickness of the nuchal fold. High thickness measurements are associated more often with embryos with Down's syndrome.

Second-trimester screening is performed starting from the 14th week of pregnancy, when the 1st trimester screening is no longer an option. This screening involves a blood sample used for the analysis of 3 proteins.

WHAT DOES THE SCREENING TEST TELL YOU?

The screening test is only a risk assessment. It does not offer a diagnosis. For example, the test may show that there is a 1 in 1,000 risk of having a baby with Down's syndrome. This means that for every 1,000 pregnant women, one will have a baby born with Down's syndrome and 999 will have a baby born without Down's syndrome. So, this is quite a low risk. The higher the second number gets, the lower the risk becomes (the less likely you are to have a baby with Down's syndrome).

Be aware, however, that a normal result from 1st - or second-trimester screening offers no guarantee that a baby will not have Down's syndrome.

What is the difference between a screening test and a diagnostic test for Down's syndrome?

There are two different types of tests that can be done during pregnancy to look for Down's syndrome - a screening test and a diagnostic test.

As mentioned above, every woman has a risk of having a baby with Down's syndrome. A screening test is offered to all women during early pregnancy to look at the risk in this pregnancy of the baby being born with Down's syndrome.

It is important to understand that the screening test does not give a definite 'yes' or 'no' answer as to whether or not the baby does have Down's syndrome. If your screening test shows a high risk that the baby has Down's syndrome, you will usually be offered a diagnostic test. A diagnostic test is done before the baby is born (prenatally) to see if the developing baby actually does have Down's syndrome or not. There are two main prenatal diagnostic tests available: amniocentesis and chorionic villus sampling.

However, there is a small risk of complications if you have a diagnostic test. This includes miscarriage. This is why a diagnostic test for Down's syndrome is not offered to all women. (A screening test for Down's syndrome does not increase your risk of having a miscarriage.)

However, these tests are not compulsory. It is your choice as to whether you have the test or not.