CHROMOSOMAL SCREENING

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Prenatal screening helps to identify whether your baby may be at increased risk of having Down's syndrome (Trisomy 21), Trisomy 13,18 or other chromosomal abnormalities.

Down's syndrome is a common cause of intellectual and physical disability. It occurs overall in 1 in 650 pregnancies although the risk is higher as your age increases. At the age of 37 your risk is approximately 1 in 200 and at 40 years old 1 in 100.

Testing is voluntary and can help give you information concerning your pregnancy to allow decisions to be made regarding the future care of your pregnancy.

Testing takes two forms – either screening tests or diagnostic tests.

SCREENING TESTS

A screening test will not diagnose Down's syndrome but will identify a group of women who are at increased risk of having a Down's syndrome baby. Screening tests take the form of blood tests and/or an ultrasound. A diagnostic test, such as Chorionic Villus sampling (CVS) or Amniocentesis, needs to be performed to diagnose Down's Syndrome if the screening test places the women in an "increased risk" category. Usually only one of these screening tests are performed.

1) Non-Invasive Prenatal testing (NIPT)- A highly sensitive maternal blood test that can be performed after 10 weeks gestation to detect minute fragments of fetal DNA in the maternal bloodstream. The test uses FISH technology, which has the ability to test all 46 chromosomes, including chromosomes 13, 18, 21 and x and y sex chromosomes. The test sensitivity is over 99% in detecting Down syndrome.
2) Ultrasound for Nuchal Translucency. An ultrasound performed between 11 and 14 weeks of pregnancy can measure the thickness of the skin on your baby's neck. This can detect up to 75% of babies that have Down's syndrome.
3) Combined test. A blood test is usually performed prior to an ultrasound for Nuchal Translucency. The combined test needs to be performed prior to 14 weeks. The blood test measures two hormones - β-HCG and PAPP-A. By combining the blood test and the ultrasound up to 90% of Down's syndrome pregnancies can be detected.
4) Second Trimester Maternal Serum Screening. A blood test usually performed at 16 weeks gestation can detect approximately 70% of pregnancies with Down's syndrome.
5) Ultrasound at 18-20 weeks. This routine ultrasound will only pick up approximately 40-45% of Down's syndrome pregnancies.

The majority women who have an increased risk of having a baby with Down's syndrome actually do not have a Down's baby. The only way to diagnose Down's syndrome, if you are increased risk, is to have a diagnostic test.

DIAGNOSTIC TESTS

In contrast to screening tests diagnostic tests are more invasive and have a small miscarriage risk. These tests are usually offered if a woman will be 37 or older at the time of birth or if a screening test demonstrates an increased risk for a chromosomal abnormality.

1) Chorionic Villus Sampling (CVS). Under ultrasound guidance a biopsy of placental cells are taken and the chromosomes are examined. This is performed after 10 weeks and has a miscarriage risk of approximately 1%.
2) Amniocentesis. Under ultrasound guidance a sample of the amniotic fluid around the baby is taken and the chromosomes are examined. This is usually performed after 16 weeks and has a miscarriage risk of approximately 0.5%.