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How long does it take to obtain the result?



Approximately 7 days.

What is the detection rate?



The detection rate for Down Syndrome is approximately 60-65%.

What happens if a screening test is positive?



As these are screening tests, an abnormal result does not necessarily mean that the fetus has an abnormality. It only means that there is a significantly higher risk of having an abnormality. If the screening test is positive, the couple would be offered diagnostic tests such as chorionic villus sampling (CVS) or amniocentesis to rule out chromosomal defects (refer to our brochure on 'Diagnostic Tests For Chromosomal Abnormalities').

Does it mean that my baby is normal if the test is negative?



A negative test result does not necessarily mean that the baby is normal in every aspect. Although the tests can detect some of the more common chromosomal abnormalities, it does not guarantee a normal baby. Since it is a screening test and only assess whether the baby is at a high risk of having chromosomal disorders, there are false negatives in all these tests as the detection rates are not 100%. They do not predict other forms of fetal abnormalities such as structural abnormalities.

The diagnostic tests, like chorionic villus sampling and amniocentesis, are extremely accurate in detecting chromosomal abnormalities. The possibility of a false result is less than 1 in 40,000 cases.



NUH Women's Centre
 Is My Answer

Screening for Chromosomal Abnormalities

Purpose of Screening Tests

Every pregnancy is associated with a small risk of chromosomal disorders, which are abnormalities involving the genetic makeup of the baby. The most common disorder is Down Syndrome where there is excessive genetic material from an extra chromosome 21. Likewise, excessive genetic materials from extra chromosome 13 and 18 result in Patau syndrome and Edward syndrome respectively. These disorders result in mental retardation and frequently, heart defects and other physical abnormalities. Screening tests are available to assess the risk of having the above disorders in each pregnancy.

Who should consider the tests?



Any pregnant woman may choose to go through these tests.

Types of Screening Test

A number of tests are available. They are done at different times of a pregnancy and have different detection rates.



1. Nuchal Translucency Screening
2. Nuchal Translucency and First Trimester Serum Screening (Combined Screening)
3. Triple Test or Triple-marker Test

1. Nuchal Translucency Screening

Nuchal translucency (NT) describes a fluid-filled (sonolucent) area at the back of the neck (nuchal region) of the fetus. Measuring the thickness of this area at a specific place is called NT measurement. This provides an assessment of the individual specific risk of having a baby with Down syndrome, Patau and Edward syndromes.

How is it done?



It is done through an abdominal ultrasound scan.

When should the test be done?

This test is done between 11 weeks, 3 days and 13 weeks, 6 days of pregnancy.

How long does it take to obtain the result?



The result is known soon after the ultrasound scan is completed. Individual risk depends on the age of the mother, the size of the baby and the NT measurements. A report of your individual specific risk will be produced.

What is the detection rate?



Using nuchal translucency screening alone, the detection rate for these chromosomal disorders is approximately 80%.

2. Nuchal Translucency and First Trimester Serum Screening (Combined Screening)



Nuchal translucency screening can also be done jointly with first trimester serum screening (blood test). It measures two hormones called the free beta HCG (human chorionic gonadotrophin) and PAPP-A (pregnancy-associated plasma protein-A). This is also sometimes known as OSCAR (OneStop Clinic for Assessment of Risk).

How is it done?



It is done through an abdominal ultrasound scan to measure the nuchal translucency together with a blood test (about 5ml) to measure the two hormones (free HCG and PAPP-A).

When should the test be done?



The test is done between 11 weeks, 3 days and 13 weeks, 6 days of pregnancy.

How long does it take to obtain the result?



Using a special analyser machine (Brahm's Kryptor machine), the blood test result is available in two hours. It is then analysed together with the ultrasound result. A report of your individual specific risk will be produced.

What is the detection rate?



The detection rate of combined nuchal translucency and first trimester serum screening for Down syndrome, Patau and Edward syndromes is approximately 90%.

3. Triple Test or Triple-marker Test

This is a screening test for Down syndrome. In this test, 3 hormones are measured – AFP (alpha fetoprotein), HCG and Estriol. Since the test includes AFP measurement, it also screens for risk of neural tube defects like spina bifida.

How is it done?



The mother goes through a blood test (about 5ml).

When should the test be done?



The test is usually done between 14 and 20 weeks of pregnancy.