

of fluid behind the neck. The results of the Nuchal Translucency, along with blood test are combined with your age, weight and stage of your pregnancy to estimate the risk of your baby having Down's Syndrome. A risk can also be calculated for twin and triplet pregnancies.

About 90% of Down's Syndrome cases and about 95% of trisomy 18 and 13 cases are detected when results of the scan and blood test are combined. The scan alone may detect only about 70% pregnancies with Down's Syndrome.

SECOND TRIMESTER SCREENING

If you have missed the opportunity of First Trimester screening, you can opt for the Second Trimester screening which also involves a blood test and scan-the genetic sonogram. The detection rate for Down's Syndrome is lower in the 2nd trimester screening as compared to First Trimester Screening.

WHAT DO THE RESULTS OF THE PRENATAL SCREENING MEAN ?

It must be clearly understood that the results represent risks and not diagnostic outcomes.

A Low Risk result implies that no further test for Down's Syndrome is required.

A High Risk result requires further diagnostic test.

WHAT IS NON INVASIVE PRENATAL TEST (NIPT)?

It is a blood test which determines the presence of free fetal DNA in maternal blood. This fetal DNA when analyzed, gives us an estimate of the risk of the baby having Down's Syndrome then you will have to undergo confirmatory testing.

JUST REMEMBER :

Most babies are born healthy.

Early and optimal screening ensures a happy and peaceful pregnancy.



CIMS PRENATAL SCREENING



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PRENATAL SCREENING

Pregnancy is a wonderful time filled with excitement and anticipation of the addition of a new member in the family. The early weeks of pregnancy can be an anxious time. Like most expectant mothers, you seek reassurance that your baby will be in your arms after a smooth pregnancy. Read on to learn more about prenatal screening and why it is important for you to be tested early in your pregnancy.

WHAT IS PRENATAL SCREENING ?

International guidelines in countries such as UK, USA, Canada, Netherlands, Spain and France recommend that prenatal screening be offered to all pregnant women regardless of age.

Prenatal screening is done between 11 to 14 weeks of pregnancy and comprises two components:

1. Ultrasound scan, and
2. Your blood is tested to evaluate the levels of placental hormones.

Ultrasound screening detects the followings:

- Whether there are one or more babies
- Checks the beating of the baby's heart
- Measures the baby's length to confirm its age
- Whether there are any structural defects in the baby
- As part of screening for Down's Syndrome

The major purpose of prenatal screening is to check for Down's Syndrome in the baby. This is done by a combination of ultrasound and blood tests which is known as Combined Screening.



BENEFITS OF PRENATAL SCREENING

All expectant mothers are at risk of having a baby with chromosomal abnormality (T21, T18, and T13). The First Trimester screening test is performed to know whether your risk of having a baby with chromosomal abnormalities is increased or decreased. With expert counseling you will then be able to decide if you need a further diagnostic test. The diagnostic test gives definite information about the presence of chromosomal abnormalities.

WHAT ARE CHROMOSOMAL ABNORMALITIES?

All of us have 23 pairs of chromosomes. Each chromosome has 2 copies. Chromosomal abnormality occurs in either number or structure of a chromosome. Intellectual and physical development is affected as a result of alteration in the chromosomes. The common chromosomal abnormalities include Trisomy 21 (Down's Syndrome) Trisomy 18 and Trisomy 13.

WHAT IS DOWN'S SYNDROME ?

Down's Syndrome (Trisomy 21) is the most commonly occurring chromosomal abnormality. It is caused by the presence of an extra copy of chromosome number 21 in the cells of the developing baby.

The incidence of Down's Syndrome is about 1 in 700 live births. Down's Syndrome is the most common cause of severe learning disability in children and is often associated with physical problems such as heart defects (40 %) or difficulties with sight and hearing. It may also be associated with childhood leukemia.

Down's Syndrome can occur at any maternal age (not just elderly pregnant women) and any baby can be affected even if there is no family history of Down's Syndrome. This means that every mother in every pregnancy is at risk of having a baby with Down's Syndrome. So this emphasizes the need to offer prenatal screening to every pregnant woman. It is not a disease or a

hereditary condition and the extra chromosome could be from the mother or father. As yet, the cause of this extra chromosome has not yet been elucidated.

Trisomy 18 and Trisomy 13 are comparatively rare and severe forms of chromosomal abnormality. Trisomy 18 and Trisomy 13 are associated with severe mental and physical disability and also life threatening complications in the baby's first months and years.

No treatment is available for any chromosomal disorder as the extra chromosome cannot be removed from cells. Caring for such a baby is by counseling, support and rehabilitation therapy.

AVAILABLE PRENATAL SCREENING TESTS

- First Trimester – combined screening with Nuchal Translucency Scan (NT Scan) and Blood tests.
- Second Trimester – Quadruple serum screening.
- Second Trimester – Genetic sonogram.
- Non-Invasive Prenatal Testing – NIPT

HOW IS PRENATAL SCREENING PERFORMED

Various methods of prenatal screening are practiced. The screening may be done in the First Trimester, Second Trimester or in both trimesters. The screening takes into account your age, a blood test and a special ultrasound test.

FIRST TRIMESTER SCREENING

The ideal model of prenatal screening is the combined First Trimester screening test. It involves a simple blood test done when the fetus is between 11 weeks and 13 weeks of age. The blood is analyzed for 2 markers normally found in all pregnant women. The blood test is preceded by an ultrasound examination. The ultrasound confirms your baby's age and measures the amount of fluid behind the baby's neck (Nuchal Translucency scan or NT scan). The result of the blood test and the ultrasound combined estimates the risk of Down's Syndrome. Trisomy 18 and Trisomy 13. Babies with Down's Syndrome can have an increased amount