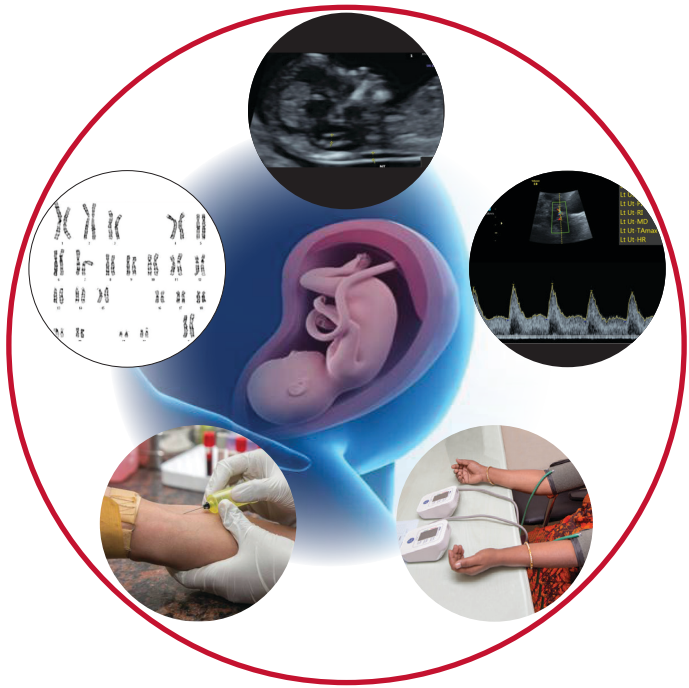
First Trimester
FETAL HEALTH CHECK
enhanced First Trimester Combined Screening (eFTS)



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Fetal Health Check

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. The vast majority of the babies are normal. However, there may be a serious health problem affecting 2-3% of babies. You may wish to find out early in your pregnancy, if your baby is at increased risk of a serious health condition.

Prenatal Screening

Prenatal Screening tests can help identify the mother at high risk of these problems. Screening has to be done ideally in the First Trimester of pregnancy i.e., $11\text{-}13^{+6}$ weeks. The Screening tests are either an ultrasound scan or a blood test or a combination of both.

First Trimester Screening

This health check requires your medical history, blood pressure measurement, ultrasound examination and a blood test.

Conditions to be Screened:

Birth defects or congenital malformations are problems that happen while a baby is developing in the mother's womb. Most birth defects happen during the first three months of pregnancy. A birth defect may affect how the baby looks or behaves or both. Birth defects can range from mild to severe. For most birth defects, the cause is unknown. However, birth defects may occur due to genetics, chromosomes, exposure to medicines etc.,

Neural tube defects are birth defects that involve the central nervous system. This system includes the brain and the spinal cord. It is formed from a structure in the developing embryo called the neural tube. As the embryo develops in the mother, the neural tube closes completely. However, if all or part of the tube fails to close, leaving an opening, the baby has a neural tube defect. Neural tube defect is the commonest birth defect in India.

The first-trimester scan helps to identify up to 50% of major birth defects. Along with a blood test, the detection rate of Spina bifida increases up to 95-99%.

Common Chromosomal abnormalities

All of us have 23 pairs of chromosomes. Each chromosome has 2 copies. The Chromosomal abnormality occurs in either the number or structure of a chromosome. Intellectual and physical development is affected as a result of alteration in the chromosomes.

All expectant mothers are at risk of having a baby with a chromosomal abnormality. Trisomy 21, 18, and Trisomy 13 are common chromosomal abnormalities.

Down 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 syndrome is about 1 in 700 live births. Down 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.

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.

Scan alone can predict up to 60-70% of chromosomal abnormalities. However, Scan and blood test together can predict up to 90%.

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.

Preeclampsia

It is a condition a woman can develop during pregnancy (at any time after 20 weeks of pregnancy) or up to six weeks after birth. This complication of pregnancy is marked by high blood pressure and protein in the urine. Left untreated, the mother and her baby are at increased risk of serious health complications, including growth restriction and prematurity. Early detection offers the opportunity to identify and treat women who are at risk.

This health check requires an ultrasound examination, blood pressure measurement and a blood test.

The chance of developing Preeclampsia can be estimated by ultrasound scan alone up to 60%, whereas a scan and a blood test together can assess the risk up to 90%.

How is the First Trimester Fetal Health Check Done?

First Trimester Fetal Health Check includes four components:

Maternal and Family History / Blood Pressure measurement / Ultrasound scan and a Blood test (eFTS)

After your registration at the Fetal Medicine Department, you will be taken to the prenatal screening room. Your doctor/nurse/midwife will ask you some questions (Medical History) about your health and your family's health. Your doctor will also check your height, weight and blood pressure. Afterwards, you will undergo a detailed ultrasound scan. Depending upon your baby's position and some technical issues, the scan may take approximately an hour. If your baby is not in a position, you may be asked to wait for a while or may be asked to come back for the next session.

As soon as your scan is over, you will be guided to the Clinical Laboratory (LAB) for a blood test.

eFTS: enhanced First Trimester Screening includes a simple blood test

Finally, in the laboratory a blood sample will be taken between 11 to 13^{+6} weeks, to look at some biochemical markers in your blood, which will help us complete the assessment. This blood test is called eFTS.

The laboratory will measure the relative concentrations of the biochemical markers present in an expectant mother's body (PAPP-A, free hCGß, PlGF and AFP). Any deviation from the normal range is indicative of some irregularity and may be a cause for concern.

Late Booking

What is the alternative test, if I miss the First Trimester Screening?

Second Trimester Screening: Ultrasound Scan and Blood Test

If you have missed the opportunity of First Trimester screening, you can opt for the Second Trimester screening which also involves scan and blood test (Quadruple test). However, the risk estimation for Down syndrome is lower than that of the first trimester (80% vs 95%).

Non-Invasive Prenatal 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 syndrome. The NIPT is sensitive up to 95% for Trisomy 21, 18 and Trisomy 13. If fetal DNA testing is positive for Down syndrome then you will have to undergo further confirmatory tests.

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 syndrome is required. These mothers can be followed with serial ultrasound scans.

A high-risk result requires further diagnostic tests. You will be guided by the Fetal Medicine experts.

For most women, the checks will assure that mother and baby are doing well and no further action is required. For a small group of women, there will be an early warning signal that indicates that the Doctor may need to request further tests or prescribe some medication for the remainder of the pregnancy to support mother and baby. By doing an assessment early in pregnancy, it enables a care pathway to be determined for you and your baby towards a happy outcome.

JUST REMEMBER
Most babies are born healthy.
Early and optimal screening ensures a happy and peaceful pregnancy.

FH/PIL/FMU/FetalHealthCheck/0087/Ver7/Jan2020