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SCI-TECH » HEALTH

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PRENATAL SCREENING

# Knowing early, and safely

divided on whether women need to undergo this expensive test.



Weekend Being health

risk of chromosomal defects

The idea of genetic disorders, an important health concern, and the third most common cause of mortality in newborns, need not baffle Indian parents-to-be anymore. Technology has now made it easier to detect problems in the early stages of pregnancy.

Many expectant mothers now access a non-invasive prenatal test (NIPT) that examines foetal DNA in the mother's blood to determine whether there is a high risk of chromosomal defects. But opinion is still

# Not a diagnostic test

Unlike the invasive amniocentesis, which is a prenatal test where a small amount of amniotic fluid is removed for testing from the sac surrounding the foetus by inserting a fine needle into the uterus through the abdomen under ultrasound guidance, or the Chorionic Villus Sampling, another prenatal test in which a sample of chorionic villi is removed from the placenta, NIPT is a blood test, according to Priya Kadam, Program Director, MedGenome NIPT, one of the players in the field.

Citing American College of Medical Genetics and Genomics (ACMG) guidelines that support the use of non-invasive prenatal tests as an optimal, initial option to screen for specific genetic conditions, she argues that NIPT should replace the conventional tests.

The test, which can be taken up anytime during pregnancy, will screen the blood sample for chromosomal disorders that include trisomy 21 (Down's syndrome), trisomy 18 (Edwards syndrome) and trisomy 12 (Patau syndrome). But this is only a screening test and not a diagnostic test. If the test shows a positive result, the doctor has to validate the defect through amniocentesis.

# Amniocentesis still required

Over the last two years, the test is being recommended by some doctors as a matter of routine in every case. While there are no figures available, the number of people getting tested is increasing, according to companies conducting the tests.

However, gynaecologists and foetal medicine specialists who youch for the safety of the test, are sceptical about the need for it.

Dr. Hema Divakar, who represents the Federation of Obstetric and Gynaecological Societies of India (FOGSI) at the International Federation of Gynecology and Obstetrics (FIGO), says with each test costing around Rs.30,000, only those cases that raise a red flag after the First Trimester Screening (FTS) need the NIP test.

"Even after getting the test done, the patient has to undergo an amniocentesis to get the result validated. This means the patient has to undergo two tests for the same purpose," she says.

Dr. C.N. Sheela, President of the Bangalore Society of Obstetrics and Gynaecology, says NIPT has several limitations. "The test results may not be as accurate in women who are obese or have had multiple pregnancies. If women in the low-risk category undergo the test, the positive predictive value is less," she says.

Genetic counselling plays an important role and the patient should be counselled that even if she undergoes NIPT, she may have to undergo amniocentesis.

Dr. N. Venkatesh, senior consultant and Head of the Department of Obstetrics and Gynaecology in Vikram Hospital, Bengaluru, says that despite some doctors being euphoric about the test, past history has shown that we need to take some universal tests and recommendations promoted by companies with a pinch of salt. "The ACMG guidelines are formulated for the western population that is genetically different from the Indian population," he says.

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