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MedGenome concludes the Study on Non-invasive Prenatal Testing

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NIPT proves to be more effective, accurate and safer way to detect common chromosomal abnormalities such as Downs's syndrome, well before birth.

Genomics based diagnostics and research company, MedGenome along with ten other research collaborators conducted India's first systematic study on Non-invasive prenatal screening test (NIPT).

The study was conducted to analyse the performance of NIPT in Indian population at MedGenome's CAP certified Bangalore Laboratory. The main objective was to evaluate the performance of NIPT for trisomies 21,18,13, X & Y chromosome abnormalities and triploidy in a cohort of pregnant women with intermediate to high risk on conventional screening.

With a small blood sample taken from the mother's arm, the NIPT screening test analyses cell free DNA of the fetus to detect chromosomal abnormalities. The screening test is safe and non-invasive. With a detection rate of over 99% and a false positive rate of less than 0.03 % the screening test is highly accurate. In situations where NIPT has been implemented, a significant reduction of 50-70% in invasive procedures has been observed.

In the study, NIPT screening was performed on 516 pregnancies, who had tested intermediate to high risk on conventional screening test (Double marker, Quadruple marker test) in the first and second trimester.

The results were confirmed through invasive testing or clinical examination post birth. Over 98% were reported low risk and 2% tested high risk on NIPT for the conditions tested. The test showed that an abnormality detected by NIPT was much more likely to be actually present and therefore authenticating that NIPT test was much more likely to truly detect abnormalities as compared to conventional screening (Double Marker test, Quadruple marker test)

It is remarkable to note that those considered intermediate to high risk pregnancies through conventional screening were found to be low risk with NIPT. Meaning, a large number (98.2%) of the women could potentially avoid invasive procedures such as amniocentesis which cause emotional anxiety and come with a small risk of miscarriage.

Dr. Priya Kadam, Program Director- NIPT, MedGenome, emphasizing the importance of non-invasive screening process said, “NIPT is not gestation age specific and can be offered to all pregnant women to be assured of their baby’s health. Also, due to high accuracy, safety and low false positive rate NIPT is a viable option for any pregnant woman and can be performed from as early as the 9th week of a pregnancy.”

She further added, “NIPT is a revolutionary test and with increased awareness and adoption this test can help significantly reduce our country’s genetic disorder burden.

Currently, NIPT may be used in cases with high-risk results on conventional screening to avoid unnecessary invasive tests. Further reduction in cost and greater awareness would provide the bene?ts of this.

The leading hospitals and institutes participated in the study were: Sir Ganga Ram Hospital, New Delhi, Rainbow Hospital, Hyderabad, Amrita Institute of Medical Sciences and Research Center, Kochi, Sri Ramchandra Medical College, Chennai, All India Institute of Medical Sciences, New Delhi, Post graduate institute and Medical Research Center, Chandigarh, Jawaharlal Nehru Institute of Post graduate Medical Education and Research, Pondicherry, CIMAR Fertility Center, Kochi, Manipal Hospital, Bengaluru, Indraprastha Apollo Hospital, New Delhi.