

Noninvasive prenatal testing – A new era of aneuploidy screening

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Abstract

Since ages maternal serum marker screening (double marker, quadruple test) and the respective antenatal ultrasound imaging (NT/NB scan and anomaly scan) have been the main stay of prenatal aneuploidy screening. Both methods, however, have large false positive rates, ranging from 2% to 7%. Since its inception in 2011, millions of pregnant women in more than 90 countries have used cell-free (cf) DNA-based Non-Invasive Prenatal Testing (NIPT) to screen for fetal chromosome abnormalities and gene mutations that cause a variety of genetic disorders.

NIPT is an advance method for detecting fetal chromosomal aneuploidies that examines cell-free fetal DNA in a pregnant woman's blood. While many professional associations advocate that NIPT be used as a screening approach rather than a diagnostic test, its excellent sensitivity (true positive rate) and specificity (true negative rate) make it a compelling alternative to serum screens and invasive procedures already in use.

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Biography

Dr. Khushboo Srivastav is a fetal medicine specialist and an obstetrician gynecologist working in Kathmandu, Nepal. She did her MBBS from Manipal College of Medical Science in 2008 followed by MS (Obstetrics and Gynecology) from BPKIHS Dharan, Nepal, in 2016. She went on to obtain a fellowship in Fetal Medicine from Bangalore, India.

She has more than five years' experience in Obstetrics and Gynecology and more than two years' experience in Fetal Medicine. She has presented in many reputed national and international conferences including RCOG (2017), ACOG (2018), RCOG (2019), Fetal Cardiac (2019).