Performance of non-invasive prenatal test in India

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Objective

Non - invasive prenatal testing has been a game changer in prenatal screening for chromosomal aneuploidies around the world. In India, the acceptance and adaption has been intermittent. To evaluate the performance of the test in an Indian setting, we conducted a research study.

Methods

Samples were collected after fulfilling preset inclusion and exclusion criteria and institutional ethical approvals from ten leading centers across India. The Panorama NIPT was performed in 516 pregnant women, who had previously tested intermediate to high risk on conventional first and second trimester screening (Combined and Quadruple screening). The samples were processed in the Medgenome laboratory located in Bengaluru, India. Results were communicated to the Clinicians for counselling the pregnant women. Both high and low risk results were confirmed, either by amniocentesis followed by fluorescence in situ hybridization and cytogenetic evaluation, or by clinical evaluation after birth.

Results

NIPT results were obtained in 97.7% samples. Of these, 480 (98.2 %) were low risk and 19 were high risk on NIPT. A sensitivity of 100% and specificity of 99.7 % was seen in all tested chromosomal abnormalities combined. Taken together, after confirmation, the positive predictive value for Trisomy 21, 18, 13 and Monsomy X was 84.2 %. The average fetal fraction was 8.2%, which was slightly lower than the average observed in studies elsewhere. NIPT results were available in 97.4% of obese women. The overall no - call rate was 2.3%. There were no false negatives.

Conclusion

This is the first systematic report of NIPT study conducted entirely in India. The results demonstrate comparable performance in all aspects of testing to the results published in other countries.