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Awareness of non-invasive prenatal testing (NIPT) for aneuploidies in Mediclinic Precise setting: a reliable accurate prenatal non-invasive (CE-IVD) diagnosis (RAPID) using whole genome sequencing method

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Abstract:

Statement of the Problem: Fetal chromosome abnormalities, specifically aneuploidy, which is an abnormal number of chromosomes, are a common cause of reproductive failure, congenital anomalies, developmental delay, and intellectual disabilities. Aneuploidy affects approximately 1 in 300 live births, with much higher rates associated with miscarriage and stillbirth. The purpose of this study is to bring awareness among pregnant women and to understand their preferences for risk information and decision-making concerning prenatal examinations with emphasis on NIPT.

Methodology and Theoretical Orientation: Pregnant women of at least 10 weeks gestation across UAE will be offered either NIPT basic or genomewide at Mediclinic precise based on their maternal age and clinical history, NIPT screening includes common aneuploidies (chromosomes 21, 18, 13), all rare autosomal aneuploidies (RAAs), sex chromosome aneuploidies (SCAs), and partial deletions and duplications, referred to as copy number variations (CNVs), ≥ 7 Mb in size. Results for NIPT will be reported within 7–10 working days. Data on clinical findings and follow-ups with NIPT positive patients will be collected for future studies.

Findings: The pregnant women in this study were helpful for this evaluation. We generated one year NIPT data from different pregnant women's across UAE, which was evaluated based on different factors affecting NIPT results like advance maternal age, maternal weight, gestational age and %ff etc. Please see below table for our one year summarized evaluation.

Conclusion & Significance: Women are considered high-risk if they are over 35, or if they have received a positive result from a different screening test. NIPT is quite accurate in that case. The results of this study will make a significant contribution to policy decisions and implementation of NIPT for aneuploidies within the UAE. It will result in safer prenatal testing for aneuploidies by reducing the number of invasive tests required. The laboratory standards for testing and reporting, education materials and counselling strategies developed as part of the study are likely to reinforce the introduction of NIPT into Mediclinic Precise practice.

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NIPT Data-1 year	
Total no. of processed NIPT samples	**870**
% of Total positive women	1.38%
% of Down syndrome	1.03%
% of Edwards syndrome	0.23%
% of Patau syndrome	0.00%
% of Turner syndrome	0.11%
% Positives in genomewide screening	0.23%

Biography

Priyanka Tripathi is a Senior Laboratory Technologist and a section supervisor who has validated an in vitro diagnostic test developed by illumina called Veriseq NIPT Solution (CE-IVD), intended for use as a sequencing-based screening test for the detection of fetal aneuploidies in pregnant women. Presently, she has been employed in Mediclinic City Hospital, Dubai and she works as a laboratory supervisor for Mediclinic Precise department. She graduated from the National Institute of Technology, Raipur, India with Bachelor Of Technology in Biotechnology in 2008. She holds 12 years of hands on experience in various molecular techniques including RT PCR, Sanger sequencing and NGS assays. Her academic project was "Alcohol Production from Paddy Straw using "Saccharomyces Cerevisae NRRL 11857" at CIAE Bhopal, India using Fermentation technique.

About Research Topic

Our NIPT in vitro diagnostic test (Basic & Genome-wide Screen) takes advantage of powerful Illumina NGS technology to bring a whole-genome sequencing (WGS) approach to NIPT. Combining a broad test menu, accurate results, and low failure rates, our NIPT offers the most comprehensive screening of the fetal genome, enabling informed and timely pregnancy management decisions. NIPT can tell you whether your baby is at risk for certain genetic conditions, such as Down syndrome. Understanding the benefits and limits of NIPT testing can help you decide whether you want it. In general, prenatal testing with the use of cfDNA had significantly lower false positive rates and higher positive predictive values for detection of trisomies 21 and 18 than standard screening methods.

About Institution:

Mediclinic Precise is part of Mediclinic International, one of the world's leading private healthcare companies with operations in Southern Africa, the United Arab Emirates and Switzerland. Mediclinic Precise is one of the key players in NGS based diagnostics and currently providing various genetic testings in the Middle East region. We are based in Mediclinic city hospital, Dubai healthcare city. The research topic of an evaluation of NIPT for aneuploidies and bringing awareness among pregnant couples and society is the main goal of Mediclinic Precise.

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