OVERVIEW

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Debate: How should NIPT be used: As primary screening to all patients

The introduction of non-invasive prenatal testing (NIPT) via cell-free DNA analysis has led to marked increases in the accuracy of screening for Down syndrome and other chromosome conditions compared to standard serum-based screening protocols. Studies have demonstrated sensitivity of >99% and an extremely low FPR (0.1%) for trisomy 21. While early clinical validation studies were focused on enriched patient groups with high risk of aneuploidy, recent studies have demonstrated this same performance in the general screening population. As such, careful consideration is appropriately being given to implementation of this technology both on an individual provider level and as part of a nationwide or public prenatal screening program. Rationale for the offering of NIPT as a first line screening to any woman opting for aneuploidy risk assessment will be discussed.

Debate: NIPT for microdeletion syndromes: Cons

New expanded non-invasive prenatal testing panels have been introduced that extend the use of cell-free DNA (cfDNA) technology to include assessment for specific fetal microdeletion syndromes as well as additional aneuploidies. If any, the performance data for these microdeletion syndromes, are derived from a very small number of samples, many of them generated in vitro. Available data suggests that false positive rates could be as high as 1% or greater and that detection rates may vary considerably based on deletion size. With relatively low disease prevalence and high false positive rates, screening in an average risk population is likely to have a low positive predictive value, with many more false positives than microdeletions detected. In addition, the residual risk for overall deleterious copy number variants (CNVs) after a 'negative' test is not predicted to change appreciably. Patients and physicians needs to carefully consider the uncertain performance of cfDNA for these conditions before widespread testing is implemented.