An overview of prenatal cf-DNA screening methods: clinical efficacy and scope.

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Background:

Non-invasive prenatal testing) was introduced over 12 years ago. Four different approaches have emerged as the predominant clinical analytic methods: massively parallel shotgun sequencing (MPSS), digital analysis of selected regions (DANSR), single-nucleotide polymorphism (SNP)-based analysis and rolling circle (RC) amplification. Each of these methods share the characteristic of analyzing cell-free DNA (cf-DNA) derived from maternal plasma sampled from 9 or 10 weeks but differ with respect to the use of enrichment of target sequencies, use of sequencing, the chromosome regions analyzed, bio-informatics, and criteria used to define a positive test. All methods have high detection rates and low false-positive rates for the common autosomal trisomies although there are differences. More importantly, they have different capabilities with respect to detecting other chromosome abnormalities, their ability to perform well at low fetal fraction, and provide testing in special situations such as multiple pregnancies.

Conclusion:

This presentation will review the overall reported performance of NIPT for the common autosomal trisomies, explore the differences between approaches, and consider how an individualized test choice would be optimal for at least some women.