

New Advances in Cell-Free DNA Analysis of Maternal Blood (Special Topic Issue: Fetal Diagnosis and Therapy 2014)



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Special Topic Issue: Fetal Diagnosis and Therapy 2014, Vol. 35, No. 3 Prenatal screening by cell-free (cf) DNA in maternal plasma has opened a new landscape for fetal medicine. Large clinical series have demonstrated that cfDNA analysis of maternal blood can achieve trisomy 21 detection rate of 99% for false-positive rate below 0.1%, which is a great improvement upon the current screening strategies. However, there is a need to define the optimal way of combining cfDNA testing with other first trimester exams. Likewise, several commercial cfDNA tests have rapidly become available, but their main features are not always readily comprehensible for clinicians. In this special issue, we combine a practical overview from a clinical perspective with meta-analysis and theoretical models that help explain the impact of introducing cfDNA testing, along with a number of important recent research contributions. This issue offers clinicians involved in fetal medicine up-to-date and useful information about the current state of cfDNA testing and how to use it in their daily practice.

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