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ACCEPTED MANUSCRIPT

Non invasive prenatal diagnosis of fetal aneuploidy using cell free fetal DNA

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Abstract

The discovery of cell free fetal DNA in maternal plasma has stimulated a rapid development of non-invasive prenatal testing. The recent advent of massively parallel sequencing has allowed the analysis of circulating cell-free fetal DNA to be performed with unprecedented sensitivity and precision. It is thus expected that plasma DNA-based non invasive prenatal testing will play an increasingly important role in the future of obstetric care. The present review summarizes recent advances in non invasive prenatal testing using cell free fetal DNA. The importance of genetic counseling, the current guidelines for the use of cffDNA screening in pregnancy, as well as specific maternal conditions that can affect the performance of non invasive prenatal testing are also discussed in this review.

Keywords: prenatal diagnosis NIPT cell free fetal DNA aneuploidy

Introduction

Approximately 3% to 5% of pregnancies are complicated by birth defects or genetic disorders. (1) Chromosomal abnormalities are present in approximately 1 in 150 live births, (2) and congenital malformations remain the leading cause of infant death and a leading cause of childhood death. (3) These chromosomal abnormalities include aneuploidy (defined as having one or more extra or missing chromosomes), translocations, duplications, and deletions. The most common chromosomal disorder is trisomy 21 (Down syndrome), with an incidence of 1 per 800 live births. (4) Trisomy 13 (Patau syndrome) and 18 (Edwards syndrome) can also result in live births, though with a significantly lower incidence (1 per 7500 live births for Trisomy 13 and 1 per 15000 live

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