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The future of cell-free DNA analysis for prenatal diagnosis

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Widespread clinical adoption of cell-free DNA analysis for prenatal assessment began in 2011 when screening tests for fetal chromosomal aneuploidies became commercially available. Applications of cell-free fetal DNA testing have continued to expand and include the detection of rare autosomal trisomies, subchromosomal aneuploidies, zygosity of twins, fetal inheritance of single gene disease mutations and fetal de novo mutations. Recent studies are starting to uncover the biology related to cell-free DNA production and degradation which in turn provide insights into molecular perturbations associated with fetal and/or maternal diseases. These developments may drive uses of cell-free DNA analysis beyond prenatal diagnosis and into antenatal monitoring instead.