Non-invasive prenatal tests of monogenic diseases: a current status.

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Background:

Noninvasive prenatal testing (NIPT) for monogenic diseases represents a significant advance in prenatal care, offering safer alternatives to traditional invasive diagnostic methods like amniocentesis and chorionic villus sampling. Recent advancements in NIPT technologies (next-generation sequencing (NGS) and digital PCR) have enabled the early detection of a wide range of monogenic diseases with minimal risk to the fetus or mother.NIPT can now accurately detect not only paternal and de novo mutations but also those inherited maternally, covering a wide range of monogenic disorders including cystic fibrosis, sickle cell disease, β-thalassemiaand Duchenne muscular dystrophy. Moreover, studies have explored various methodologies to enhance the accuracy and applicability of NIPT for monogenic disorders. These include the development of a Bayesian model integrated with haplotype-based methods for the recovery of the whole fetal genome, which showed promising results in detecting single nucleotide variations (SNVs) and insertions/deletions (indels).

Conclusion:

The evolving landscape of NIPT for monogenic diseases underscores its potential to significantly impact prenatal care by providing a safe, accurate, and comprehensive approach to diagnosing a wide spectrum of genetic conditions. This advancement not only enhances the ability to manage pregnancies at risk of transmitting monogenic disorders but also broadens the scope to include both paternally and maternally inherited conditions, thereby offering families crucial information for informed decision-making regarding their pregnancies.