What we know about cell-free fetal DNA fragments

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Background:

The exploration of cell-free fetal DNA (cffDNA) fragments in maternal plasma represents a significant breakthrough in the field of non-invasive prenatal testing (NIPT). This lecture aims to comprehensively focus on the characteristic properties of cffDNA, such as its origin, kinetics, structure, fetal fraction, and the significance of these facts for prenatal diagnostics and understanding the mother-fetus relationship.

cffDNA primarily originates from the placenta and enters the maternal bloodstream through apoptotic processes of trophoblast cells. This origin offers a unique insight into the biological interaction between mother and fetus, while also highlighting the importance of the placenta as a source of cffDNA.

The kinetics of cffDNA refers to its dynamics during pregnancy, with its concentration in maternal plasma increasing with advanced gestational age and rapidly decreasing after birth. This fact points to the possibility of monitoring the health of the fetus and placenta through cffDNA analysis.

The structure of cffDNA fragments, typically short and double-stranded, plays a key role in their stability and allows for their effective detection. The size and form of these fragments are important for understanding the mechanisms of their release into the bloodstream.

Fetal fraction, the proportion of cffDNA relative to maternal DNA, is a critical parameter in the efficacy of NIPT. Factors influencing fetal fraction, such as maternal weight, gestational age, and chromosomal abnormalities, affect the sensitivity and specificity of cffDNA-based tests. Understanding these factors is essential for accurate interpretation of NIPT results and the development of more robust diagnostic tools.

Conclusion:

This lecture will provide a comprehensive overview of cffDNA properties and their relevance for the advancement of prenatal diagnostics, improving understanding of the fetomaternal relationship, and potential clinical applications.