عنوان مقاله:

The current applications of cell-free fetal DNA in prenatal diagnosis of single-gene diseases: A review

محل انتشار:

مجله طب تولید مثل ایران, دوره 20, شماره 8 (سال: 1401)

تعداد صفحات اصل مقاله: 14

نویسندگان:

Mohamad Mahdi Mortazavipour - *Department of Medical Genetics, Faculty of Medical Sciences, Tarbiat Modares University, Tehran, Iran.*

Reza Mahdian - *Molecular Medicine Department, Pasteur Institute of Iran, Tehran, Iran.*

Shirin Shahbazi - *Department of Medical Genetics, Faculty of Medical Sciences, Tarbiat Modares University, Tehran, Iran.*

خلاصه مقاله:

Prenatal diagnosis of hereditary diseases has substantially altered the way medical geneticists are helping families affected by genetic disorders. However, the risk of miscarriage and fear of invasive diagnostic procedures may discourage many couples from seeking prenatal diagnosis. With the discovery of maternal plasma cell-free fetal DNA, prenatal diagnosis has entered a new era of progress. Cell-free DNA is released during normal physiological functions as well as through the cell death programs of apoptosis and necrosis. It can be found in the plasma and other body fluids. Although this method has the advantage of being noninvasive, it is still rather expensive and requires advanced hardware and comprehensive data analysis. Promising implications of noninvasive prenatal diagnosis methods for the diagnosis of common trisomy disorders have paved the way for the development of more complicated assays of single-gene disorders. Relative mutation dosage and relative haplotype dosage are the most widely implemented assays for noninvasive prenatal diagnosis of single-gene disorders. However, each assay has its own advantages and disadvantages. Relative mutation dosage is based on the droplet digital polymerase chain reaction (PCR) technique which includes quantification features of real-time PCR assays. Relative haplotype dosage is based on next-generation sequencing that includes analysis of the maternal and paternal genome followed by sequencing of maternal plasma cell-free DNA. Co-amplification at a lower denaturation temperature PCR is another approach that is based on forming heteroduplexes between alleles to selectively amplify paternal mutations. In this review, we have described the most common noninvasive prenatal diagnosis approaches and compared their applications in genetic disorder diagnosis with different inheritance patterns.

کلمات کلیدی:

Cell-free nucleic acids, Prenatal diagnosis, Noninvasive prenatal testing, Single-gene diseases, Non-invasive techniques., DNA آزاد, تشخیص قبل از تولد, تشخیص غیر تهاجمی قبل از تولد, نقایص تک ژنی, تکنیک های غیرتهاجمی.