Non-Invasive Prenatal Testing Using Cell-Free Fetal DNA from Maternal Plasma: A Review

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Abstract

Nowadays, the use of cell-free fetal DNA (cfDNA) is a promising tool in clinical practice as a potential non-invasive prenatal testing (NIPT) method since it was discovered in early 1970s. The fetal DNA is approximately ~10% in a mixture of maternal DNA from maternal plasma and this amount will increase as gestation period increases. In recent years, the development of robust molecular analysis of fetal DNA namely via RT-PCR, next generation sequencing (NGS), digital PCR and massively parallel sequencing (MPS) helps the implementation of NIPT especially in fetal chromosomal aneuploidy detection. Thus, these analyses provide the alternative to the conventional invasive prenatal testing such as amniocentesis and chorionic villus sampling (CVS). The common fetal aneuploidy is trisomy 21 (T21) which is caused by an extra copy of all or part of chromosome 21 and known to be validated by amniocentesis approach as the gold standard method. Currently, the epigenetic detection of trisomy 21 had been introduced as a new non-invasive method that investigates the association between DNA methylation and gene expression in T21 fetal DNA. This review briefly summarizes the NIPT and invasive prenatal testing of fetal aneuploidy and recent molecular analysis study by using cfDNA from maternal plasma.

PDF

Published
2017-09-16

How to Cite
GHAFAR, Nurul Fatehah Abdul; ZAINUDDIN, Norafiza. Non-Invasive Prenatal Testing Using Cell-Free Fetal DNA from Maternal Plasma: A Review. **INTERNATIONAL JOURNAL OF ALLIED HEALTH SCIENCES**, [S.l.], v. 1, n. 1, sep. 2017. Available at: <http://journals.iium.edu.my/ijahs/index.php/IJAHS/article/view/79>. Date accessed: 03 nov. 2017.

Citation Formats
ABNT
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BibTeX
CBE
EndNote - EndNote format (Macintosh & Windows)
MLA
ProCite - RIS format (Macintosh & Windows)
RefWorks
Reference Manager - RIS format (Windows only)
Turabian

Issue
Vol 1 No 1 (2017): Special Issue: Enhancing Academic and Research Quality

Section
Articles

KRW2016/ORAL/AHSC2016/35

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ABSTRACT

Nowadays, the use of cell-free fetal DNA (cfDNA) is a promising tool in clinical practice as a potential non-invasive prenatal testing (NIPT) method since it was discovered in early 1970s. The fetal DNA is approximately ~10% in a mixture of maternal DNA from maternal plasma and this amount will increase as gestation period increases. In recent years, the development of robust molecular analysis of fetal DNA namely via RT-PCR, next generation sequencing (NGS), digital PCR and massively parallel sequencing (MPS) helps the implementation of NIPT especially in fetal chromosomal aneuploidy detection. Thus, these analyses provide the alternative to the conventional invasive prenatal testing such as amniocentesis and chorionic villus sampling (CVS). The common fetal aneuploidy is trisomy 21 (T21) which is caused by an extra copy of all or part of chromosome 21 and known to be validated by amniocentesis approach as the gold standard method. Currently, the epigenetic detection of trisomy 21 had been introduced as a new non-invasive method that investigates the association between DNA methylation and gene expression in T21 fetal DNA. This review briefly summarizes the NIPT and invasive prenatal testing of fetal aneuploidy and recent molecular analysis study by using cfDNA from maternal plasma.

KEYWORDS: NIPT, Trisomy 21, Non-Invasive Prenatal Testing, DNA Methylation, Invasive, Cell-Free Fetal DNA, cffDNA

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