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Non-Invasive Prenatal Testing:

Use of Cell-Free Fetal DNA in Down syndrome Screening

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ABSTRACT

Cell Free Fetal DNA can be used to determine higher chance of having disease state in fetus such as Down Syndrome (trisomy 21), trisomy 18(Edward syndrome), trisomy 13(Patau syndrome) or the abnormalities in the sex chromosomes .In this test sample of the mother's blood is taken after 10 weeks of pregnancy. The test measures the small fragments of fetal DNA in the mother's blood & can determine the chance of a chromosome problem based on the relative amount of DNA from chromosome 21,18,13 & sex chromosomes. Non- invasive prenatal testing (NIPT) is based on analysis of Cell Free Fetal DNA in maternal blood. This approaches the basis of NIPT for aneuploidy, a maternal blood test, that can be performed in early pregnancy to significantly refine the down syndrome risk & reduce need for invasive testing such as chronic villus sampling (CVS) or amniocentesis. Non-invasive prenatal testing (NIPT) based on cell-free fetal DNA (cffDNA) is highly accurate in the detection of common fetal autosomal trisomies. Aim of this project was to investigate short-term costs apnd clinical outcomes of the contingent use of cffDNA for prenatal screening of trisomies 21, 18, 13 within a national health service (NHS). In human pregnancy, the constant turnover of villous trophoblast results in extrusion of apoptotic material into the maternal circulation. This material includes cell-free (cf) DNA, which is commonly referred to as "fetal", but is actually derived from the placenta. As the release of cf DNA is closely tied to placental morphogenesis, conditions associated with abnormal placentation, such as preeclampsia, are associated with high DNA levels in the blood of pregnant women. Over the past five years, the development and commercial availability of techniques of massively parallel DNA sequencing have facilitated noninvasive prenatal testing (NIPT) for fetal trisomies 13, 18, and 21

Keywords :- Edward syndrome, Patau syndrome, Non- invasive prenatal testing (NIPT) , chronic villus sampling (CVS) .

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