

Fetal DNA in Maternal Plasma: Biological and Diagnostic Implications

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Prenatal diagnosis has become an important part of antenatal care. Tests or procedures are carried out prenatally either as a screening procedure for relatively prevalent disorders, or as a diagnostic procedure for known familial conditions. However, at present, the sampling of fetal material is dependent on invasive procedures such as amniocentesis, chorionic villus sampling, and fetal blood sampling. These procedures, unfortunately, are associated with a finite risk of morbidity and mortality to the fetus. Consequently, prenatal diagnosis is currently offered on a limited basis to pregnancies where the benefit outweighs the risks. High risk pregnancies include those with significant family history, abnormal morphological scans, suspicious serum biochemical results and in the case of Down's syndrome, maternal age over 35 years. However, the current practice has limited effectiveness in that 80% of Down's syndrome infants are born to women under 35 years of age. Consequently, advances need to be made in reducing the risks associated with prenatal testing, so that more pregnancies could benefit from such procedures. In particular, there has been much recent research interest in the sampling of fetal material by non-invasive means. (HKJGOM 2000 ; 1 : 116 - 120)

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