



# Non-Invasive Prenatal Testing: A Method to Detect Fetal Aneuploidy

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## DESCRIPTION

Fetal chromosomal, genetic and biochemical abnormalities are routinely detected by analysis of fetal cells obtained by invasive procedures such as amniocentesis and chorionic villus sampling. A small but definite risk of injury to mother and fetus is conferred by invasive sampling procedures. Another source of genetic material that accurately represents the condition of the fetus is circulating cell-free fetal DNA (ccfDNA), found in maternal plasma. The placenta is believed to be the major source of ccfDNA and consequent clearance of ccfDNA from maternal plasma occurs within hours of birth. Recently, the use of ccfDNA has enabled the introduction of Non-Invasive Prenatal Testing (NIPT) methods such as fetal RHD genotyping from maternal plasma and detection of fetal aneuploidy in high-risk in women. However, care should be taken to avoid an increase in circulating maternal DNA following phlebotomy (by lysis of maternal white blood cells), as quantitative applications, such as non-invasive detection of aneuploidy, may be affected by a relative decrease in Fetal Fraction (FF).

Typical fetal fractions range from 2% to 40% with an average of 10% total ccfDNA at different gestational ages. The minimum fetal fraction for accurate determination of trisomy 21 is 4%, as measured by the Fetal Quantifier Assay (FQA). Therefore, a small increase in maternal DNA could lower the fetal fraction below the 4%, especially in cases of low initial fetal fraction. No test results are reported below 4% providing a large population of mothers with access to non-invasive prenatal testing methods requires robust, validated blood collection equipment and processing protocols. To maintain a high fetal fraction, processing protocols for collecting maternal blood in standard EDTA tubes require cold storage of blood samples, followed by plasma preparation within 6 hours. The preparation of the plasma begins with a low-speed centrifugation of the maternal blood for the fractionation of the plasma from the blood cells. The plasma layer is then removed and centrifuged at a faster rate to pellet any residual debris from the plasma. Plasma processing

is cumbersome and therefore is performed at assembly sites only if absolutely necessary due to location or other circumstances.

The immediate demand for plasma processing for blood collected in standard EDTA tubes and the associated processing costs at collection sites would unnecessarily limit the availability of NIPT to a large population. To address these challenges, an ideal blood collection device would allow the shipment of whole blood at room temperature (6°C-37°C) and extend plasma processing times. Such a device would facilitate centralized processing and analysis. There are several alternatives to EDTA tubes for blood collection and plasma preparation for molecular diagnostic tests. There are three possible types of tubes for maintaining fetal fractions: tubes designed to create a physical barrier (gel cap or mechanical separator) between cellular and non-cellular blood components, tubes that provide reagents to keep maternal blood cells intact and active for a defined period of time and tubes containing cell retention reagents. A tube of cell storage reagents to prevent white blood cell degradation (maternal DNA release) and inhibit nuclease-mediated DNA degradation for up to 14 days at room temperature was recently introduced.

However, when the blood was stored in EDTA tubes for 72 hours, an increase in "long fragment" (maternal) plasma DNA was evident. The results indicated that in blood collected in EDTA tubes shipped with or without frozen ice packs, there was an elevated level of total plasma DNA after 72 hours. When whole blood is shipped in Streck BCT without ice packs (72 hours), the total DNA level remains unchanged, while shipment with frozen ice packs increases the total DNA level. This study aimed to evaluate the ability of Streck BCTs to maintain ccfDNA concentrations and inhibit nuclease-mediated DNA degradation under pragmatic conditions for routine use for clinical applications. Our assessment of the usefulness of the Streck BCT for maintaining the integrity of the fetal DNA fraction focused on four key variables: influence of storage time (up to 14 days), storage temperature (for 24 hours of storage), mechanical stress.

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