St George's University Hospitals NHS
NHS Foundation Trust

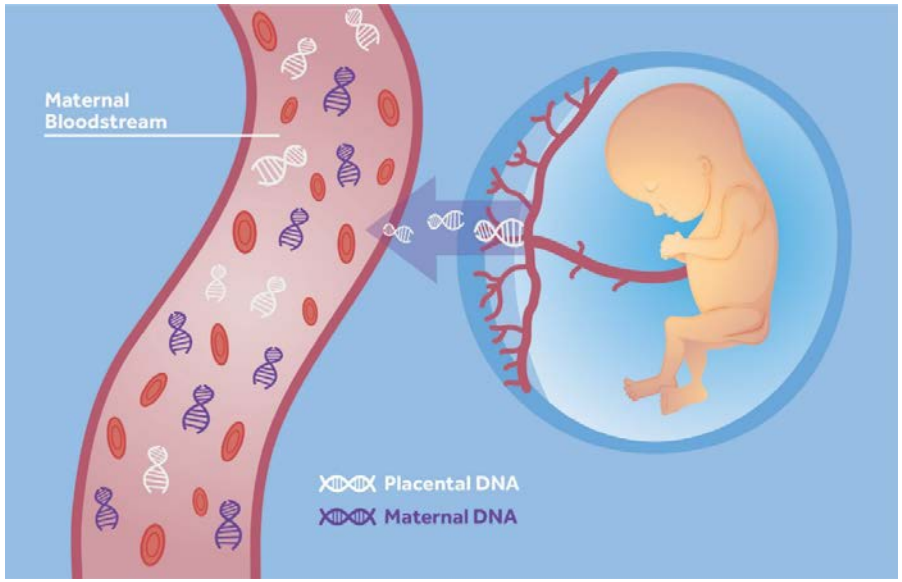
Module 1:
NIPT cell-free DNA introduction

Module 1 will cover:

- What is feto-placental cfDNA?
- How is cfDNA used to screen for chromosomal aneuploidies?
- How accurate is cfDNA screening?
- NIPT recommendations for use within the NHS

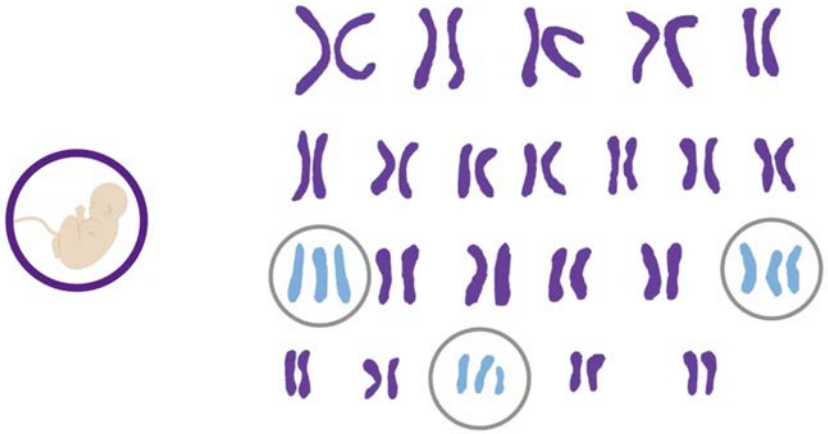
the SAFE test
St George's Antenatal Fetal Evaluation

What is placental cfDNA?



- During pregnancy the placenta and fetus leak cell-free DNA
- The majority of the cfDNA originates from the cells that make up the placenta
- Maternal blood contains a mixture of feto-placental and maternal cfDNA
- Approximately 10% of the circulating cfDNA comes from the feto-placental unit.

How is cfDNA used to screen for trisomies?



- Trisomy 21 (Down's Syndrome)
- Trisomy 18 (Edwards' Syndrome)
- Trisomy 13 (Patau's Syndrome)

- Chromosomes are made up of long strands of DNA that contain information that determine the baby's growth and development
- Trisomic syndromes occur when a person has an extra chromosome over and above the normal two strands
- During NIPT small fragments of feto-placental cfDNA are sequenced and counted
- Extra chromosomal material indicates the presence of a trisomy

How accurate is cfDNA screening?

- NIPT has a detection rate of greater than 99%, significantly reducing false negative results (<1%)
- NIPT has a specificity of greater than 99%, significantly reducing the false positive rate (<1%)
- This compares to a detection rate of approx. 85% and a false positive rate of 3% in the first trimester combined test
- NIPT enables greater clarity in the management of pregnancy

What are the recommendations for use within the NHS?

- The National Screening Committee (NSC) has recommended that NIPT is offered to women who have a high chance result (≥1:150) of trisomy on combined or quadruple testing
- The NHS is expected to roll-out NIPT to these high-risk women during 2018
- The recommendation is on the basis of the effectiveness of NIPT at avoiding invasive prenatal diagnosis and the healthcare cost savings.
- As a healthcare professional, it is important to understand who is eligible for the test, its risks, benefits and limitations