Cell-Free Fetal DNA Testing (Green-Top Guideline No. XX)

Final Scope

This is the first edition of this guideline, and has been produced jointly with the British Maternal and Fetal Medicine Society.

1. Purpose and Scope
2. Introduction and background
    - 2.1 Antenatal screening in the first trimester
    - 2.2 Screening tests for trisomies 21, 18 and 13
    - 2.3 Diagnostic testing with chorionic villous sampling and/or amniocentesis
    - 2.4 Policy and performance of aneuploidy screening in NHS England
3. Identification and assessment of evidence
4. Cell-free fetal DNA as a screening tool for chromosomal anomalies
    - 4.1 Cell-free fetal DNA
    - 4.2 What are the different approaches that can be used for screening with cell-free fetal DNA?
        - 4.2.1 Universal screening for trisomies 21, 13 and 18
        - 4.2.2 Contingent screening for trisomies 21, 13 and 18
        - 4.2.3 Based on clinical indication for pregnancies considered at a-priori high risk
    - 4.3 What do women need to know before choosing to have screening with cell-free fetal DNA?
    - 4.4 What is the screening performance of cell-free fetal DNA in low-risk and high-risk populations?
    - 4.5 How is fetal fraction defined, and what is its significance?
    - 4.6 What are the patient factors known to affect the performance of cell-free fetal DNA screening?
        - 4.6.1 Maternal factors
        - 4.6.2 Fetal factors
        - 4.6.3 Placental factors
    - 4.7 What is the recommended patient pathway in cases where cell-free fetal DNA screening fails to yield a result?
5. Expanding the use of cell-free fetal DNA screening beyond the detection of trisomies 21, 13 and 18
    - 5.1 Fetal sex
    - 5.2 Sex chromosomal aneuploidies
    - 5.3 Copy number variants
    - 5.4 Rare autosomal trisomies
    - 5.5 Single gene disorders
6. Potential indications for offering cell-free fetal DNA screening
    - 6.1 Women who have had the combined test with a high chance result for trisomy 21, 18 or 13 because of raised nuchal translucency or abnormal serum biochemistry
    - 6.2 Women who have had a high chance result from the quadruple test for trisomy 21
    - 6.3 Fetal anomalies detected by ultrasound
    - 6.4 Women with a previous diagnosis of trisomy 21, 13 or 18
    - 6.5 Women who have conceived with in vitro fertilisation/intracytoplasmic sperm injection and had pre-implantation genetic screening
    - 6.6 Women who have had cell-free fetal DNA screening with a low chance result and a subsequent finding of:
        - 6.6.1 Elevated nuchal translucency