Non-invasive Prenatal Testing (Green-Top Guideline No. XX)

Draft scope

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

1. Purpose and Scope
   The guidance would cover the responsibilities of healthcare professionals to provide tests that are known to be accurate to a level that is appropriate to the condition or impairment being tested for; the provision of accurate, balanced and non-directive information and support; result giving; and dealing with unanticipated or secondary findings and failed tests.
2. Introduction and background epidemiology
   - 2.1. Different tests for Screening for Trisomy 21, 18 and 13 (including contingent screening) and Diagnostic testing
   - 2.2. Uptake of screening in UK, trends in invasive diagnostic procedures. Performance of screening in England including detection rates, screen positive rates and predictive value (positive and negative)
   - 2.3. Evolution of NIPT
3. Identification and assessment of evidence
4. NIPT as a screening tool for chromosomal abnormalities
   - 4.1. What are the common indications for NIPT?
     (including primary screening, contingent screening and gestational ages)
   - 4.2. What are the current recommendations from Fetal Anomaly Screening Programme (FASP) around the use of NIPT?
   - 4.3. What do women need to know before choosing to have NIPT? (including what should be included in consent)
   - 4.4. What is the performance of NIPT as a screening tool (including low and high chance populations)?
   - 4.5. What is the implication of fetal fraction in interpretation of NIPT results, and is this always reported?
   - 4.6. What are the common factors affecting the performance of NIPT screening?
     (false positives and negatives - including body mass index, assisted conception, maternal solid organ transplants, women on biological treatments)
   - 4.7. What are the recommendations when NIPT test fails to yield a result?
   - 4.8. Is there any role of NIPT for detection of rarer genetic conditions?
     (including 22q deletion, whole genome sequencing)
   - 4.9. Is there a role of NIPT for detection of sex or sex chromosomal aneuploidies?
5. Women opting for NIPT: common clinical situations and recommendations for counselling
   - 5.1. When there is increased nuchal translucency, and
     - 5.1.1. Low chance NIPT result?
     - 5.1.2. High chance NIPT?
   - 5.2. Where there are structural abnormalities in first trimester scan and low chance NIPT?
   - 5.3. Where there are structural abnormalities in anomaly scan (for example, nuchal fold [≥ 6 mm]; ventriculomegaly [≥ 10 mm]; echogenic bowel [with density equivalent to bone]; renal pelvic dilatation [AP measurement > 7 mm]; small measurements compared to dating scan [significantly less than 5th centile on national charts]) and low chance NIPT?
   - 5.4. Where NIPT has been performed outside the recommendations of FASP
     (for example, primary screening, very early or very late in gestation or a low risk combined test or parental choice)

5.5. What is the role of NIPT in women with a previous diagnosis for Trisomy 13, 18, 21?
6. How to manage women with high chance NIPT (irrespective of indication of the test) who have opted against diagnostic testing?
7. NIPT for multiple pregnancy
    7.1. What is the performance of NIPT in both monochorionic and dichorionic twins?
    7.2. How to interpret NIPT results for twins pregnancies?
    7.3. Can NIPT be offered in a twin pregnancy with a single empty sac or vanishing twin?
    7.4. Is there a role of NIPT in triplets and higher order pregnancy?
    7.5. Is there a role for NIPT in multiple pregnancy discordant for NT or anomaly?
8. Follow-up care after NIPT results and counselling
    8.1. What is the optimal diagnostic test (chorionic villus sampling [CVS] or amniocentesis)? (Cross reference to RCOG Green-top Guideline on CVS and amniocentesis)
    8.2. How to manage suspicion of confined placental mosaicism CVS?
    8.3. How to manage discordant results between NIPT and CVS/amniocentesis results? (including maternal copy number variants and maternal malignancies, high risk NIPT and normal CVS/amniocentesis)
    8.4. What is the role of a detailed anomaly scan?
    8.5. What are the indications for fetal echocardiography?
    8.6. What are the indications for referral to Clinical Genetics?
    8.7. What is the role of support groups to empower women with their choice for NIPT?
9. Recommendations for future research
    9.1. Role of NIPT in primary screening?
10. Auditable topics
11. Useful links and support groups
12. Quick reference flow diagram: Management pathway – care of women opting for NIPT