The serum integrated test to screen women for fetal trisomy 21

This document prepares the clinician to discuss scientific data with the patient so they can make an informed decision together.

Presenting the serum integrated test to patients

What is this test for?
- The serum integrated test estimates the risk of carrying a fetus with trisomy 21 (Down syndrome). If the risk is high, the physician offers an amniocentesis to verify that the fetus has trisomy 21.
- This information lets the patient decide whether to end the pregnancy or prepare for a child with special needs.

How is the test performed?
- Two blood samples are taken:
  - 1st: between 10 and 13 weeks of pregnancy
  - 2nd: between 14 and 16 weeks of pregnancy

Who might consider being tested?
- Any pregnant woman.
- The risk of trisomy 21 increases with a woman's age. Other risks include a previous pregnancy complicated by fetal trisomy 21, having at least one major or two minor fetal structural anomalies in the current pregnancy and chromosomal translocation or inversion, or aneuploidy in the woman or her partner.

What is considered a high risk of trisomy 21?
- The cutoff level chosen to define a positive result (high risk) and distinguish it from a negative result is usually 1:500, which corresponds to the risk of miscarriage from amniocentesis.

Why do patient preferences matter when making this decision?
- There are pros and cons to taking this test:
  - PROS: 85% of fetuses with trisomy will be detected during screening. This may lead to unnecessary amniocentesis, with the risk of providing a miscarriage (1 miscarriage/300 amniocenteses).
  - CONS: 4% of women, the test will indicate a high risk of trisomy 21 where there is none. This may lead to unnecessary amniocentesis, with the risk of providing a miscarriage (1 miscarriage/300 amniocenteses).
- Both doing and not doing the test are both acceptable options, so we propose that:
  - the decision takes into account the patient's values and preferences
  - the clinician shares this decision with the patient