

## The serum integrated test to screen women for fetal trisomy 21

Probabilities of benefits and harms

Patient's preferences and values



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?

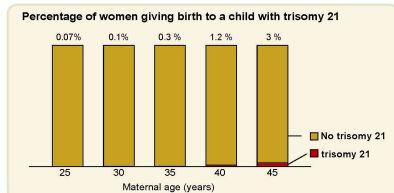
- Two blood samples are taken:**
  - 1<sup>st</sup>: between 10 and 13 weeks of pregnancy
  - 2<sup>nd</sup>: between 14 and 16 weeks of pregnancy

### 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 1/300, which corresponds to the risk of miscarriage from amniocentesis.<sup>1</sup>

### 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.<sup>1</sup>



Data source: Retrospective analysis of national registers of all live births (6 million) in England from 1969 to 1998.<sup>2</sup>

### 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.<sup>3,4</sup>

**CONS:** For 4% of women, the test will indicate a **high risk of trisomy 21 where there is none**.<sup>3,4</sup> This may lead to unnecessary amniocentesis, with the risk of provoking 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**



Serum integrated test

## State of knowledge – September 2011 Selection of best available studies

- Results from two prospective cohort studies were used to calculate benefits and harms of prenatal screening:
- SURUSS Study:** 47,000 pregnant women mostly from the UK (16% older than 35 years old) from 1996 to 2000.<sup>3</sup>
  - FASTER Study:** 37,000 pregnant women from the United States (22% older than 35 years old) from 1999 to 2002.<sup>4</sup>

### Benefits of screening

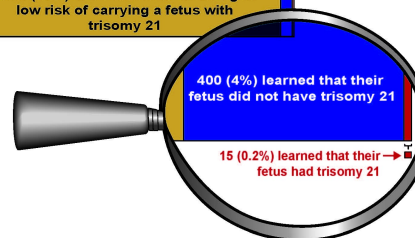
- Knowing about fetal health**
  - For each 10,000 women screened, **415 (4%)** were identified as being at **high risk** of carrying a fetus with trisomy 21.
  - These women had **amniocentesis** to verify the results of the screening, and **15 (0.15%)** were actually **carrying a fetus with trisomy 21**.
- Reassurance**
  - For each 10,000 women screened, **9585 (96%)** were identified as being at **low risk** of carrying a fetus with trisomy 21. These women were **reassured**.

### Harms of screening

- False reassurance**
  - Of the **9585** women identified as being at low risk of carrying a fetus with trisomy 21, **2** were **actually carrying a fetus with trisomy 21**. These women had been **falsely reassured**.
- False alarm**
  - Of the 415 women identified as being at high risk of carrying a fetus with trisomy 21, **400** were **not carrying a fetus with trisomy 21**.
    - Many of these women experienced anxiety.
    - Of these 415 women, **1** experienced a **miscarriage** following "confirmatory" amniocentesis (miscarriage rate 1/300).<sup>2</sup>

### For each 10,000 women who underwent screening:

**9585 (96%)** were identified as being at **low risk** of carrying a fetus with **trisomy 21**



### Less false alarms:

Using **nuchal translucency** in combination with the serum integrated test reduces the number of women identified as being at high risk when in fact they are not carrying a fetus with trisomy 21 from **400 to 80**.<sup>3,4</sup>

1 woman experienced a **miscarriage** following "confirmatory" amniocentesis

### How much confidence can we have in these results? **High**

These figures are founded on two observational studies<sup>3,4</sup> that used high-quality methods.

### Questions to identify the patient's decision making needs:

- Do you have any questions about the benefits and harms of each option?
- Which benefits and harms matter most to you?
- Do you feel sure about the best choice for you?
- Who will support and advise you in making a choice?