

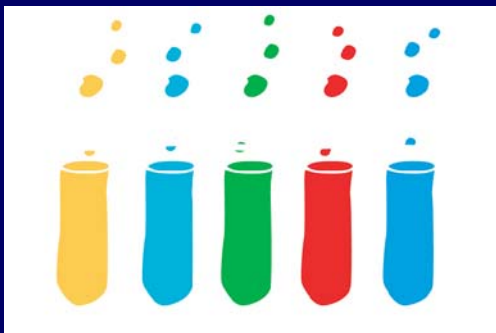
Screening Tests

Element D

Aims and Objectives

- List and discuss screening tests currently available
 - Examine markers used
 - Time of testing
 - Detection rates / False positive rates
- Tests of the future
- NSC Recommendations

Screening Tests



- Maternal Age
- Second Trimester
 - Double test
 - Triple test
 - Quadruple test
- First Trimester
 - Nuchal Translucency
 - Combined test
- First and Second Trimester
 - Integrated test
 - Integrated serum

Maternal Serum Screening for Open Neural Tube Defects

Anencephaly



AFP >5.0 MoM
hCG NORMAL
uE₃ <0.5 MoM

Open Spina Bifida



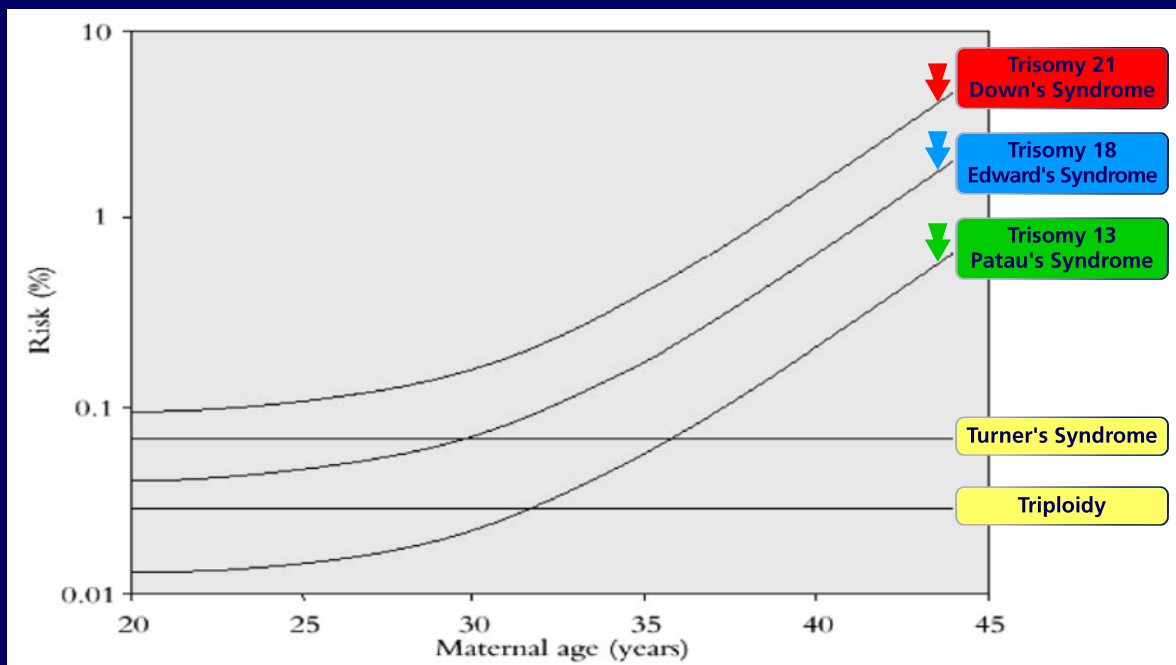
AFP >2.5 MoM
hCG NORMAL
uE₃ NORMAL

Maternal Serum Screening for Open Neural Tube Defects

- Raised maternal serum AFP
- Optimum time of testing 16-18 weeks
- Cut off usually 2.2 - 2.5 MoMs
- False Positive - Bleeding/incorrect GA
- Diagnosis is made by USS
- IUGR / stillbirth association
- Detection rate 99% anencephaly, 90% open NTD (minor lesions may not be detected)

(RCOG 2000)

Maternal age-related risk for chromosomal abnormalities

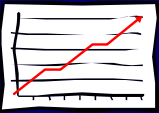
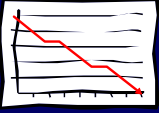
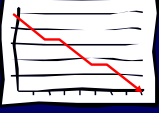
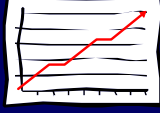



Second Trimester Screening

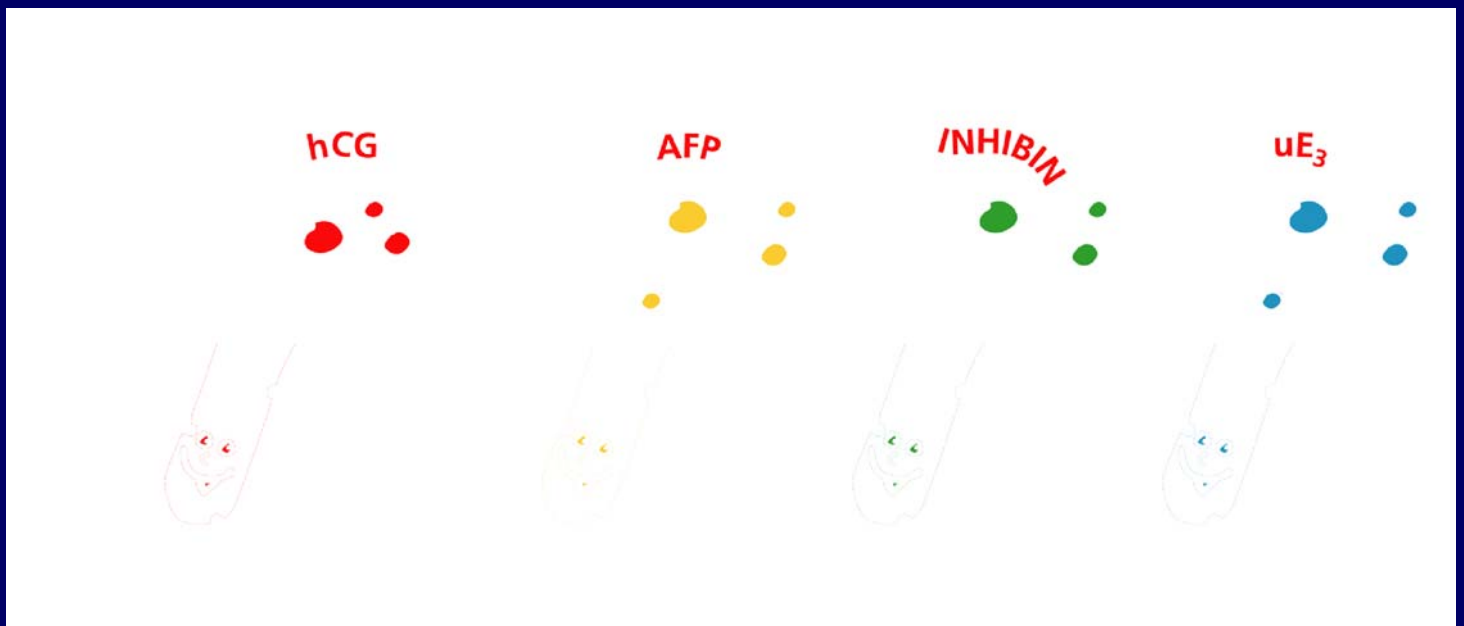
Test	Biochemical Markers
Double Test	AFP + hCG
Triple Test	AFP + hCG + uE ₃
Quadruple Test	AFP + hCG + uE ₃ + Inhibin A

Second Trimester Screening

What happens normally with gestational age?

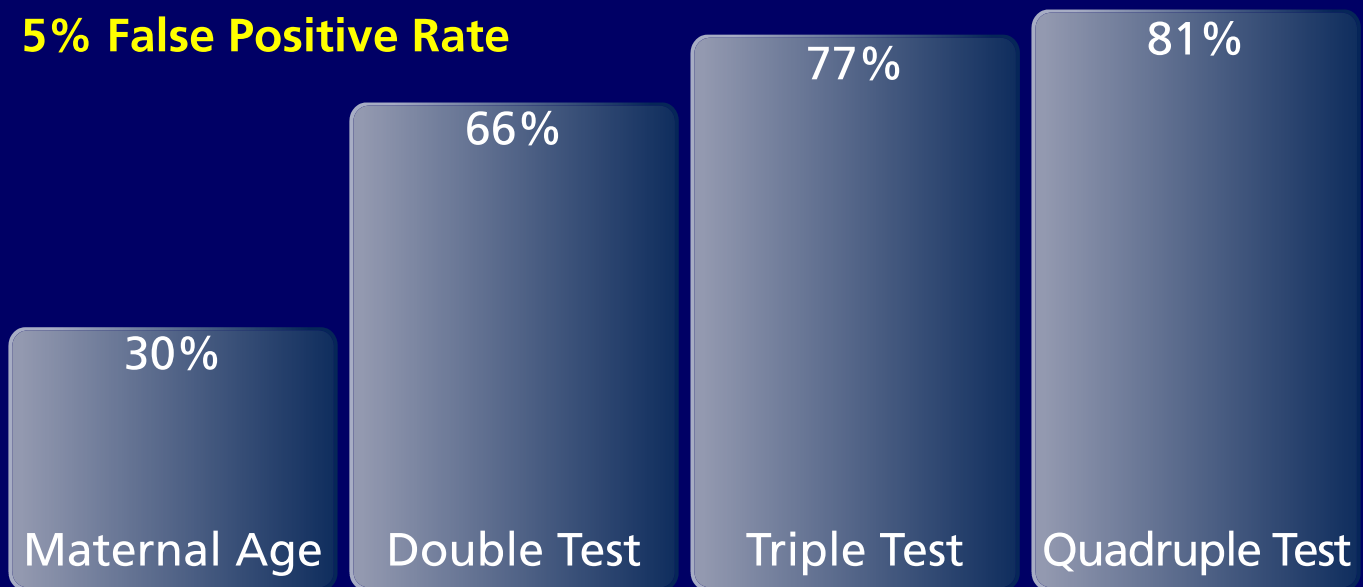
- AFP } 
- uE₃ }
- hCG 
- Inhibin A  until 17 weeks 
- Inhibin levels are 40% higher in smokers 

Median Marker Levels in Down's Syndrome Pregnancies



Second Trimester Detection Rates

5% False Positive Rate

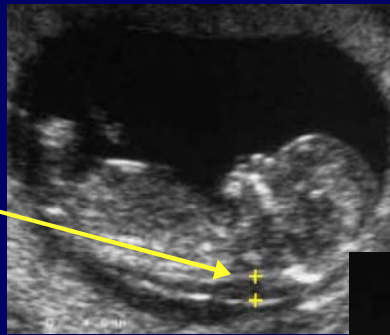


How does this test perform locally?

(SURUSS 2003)

First Trimester Screening

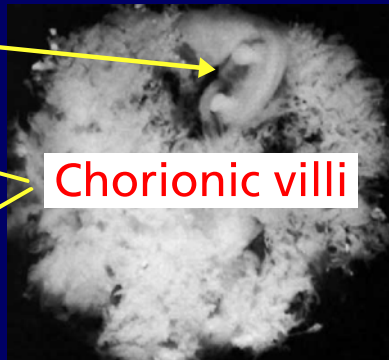
Nuchal Translucency



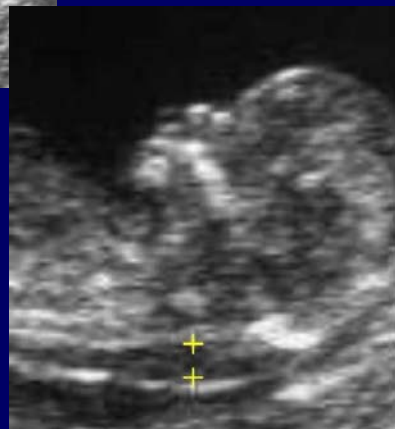
Fetus

PAPPA

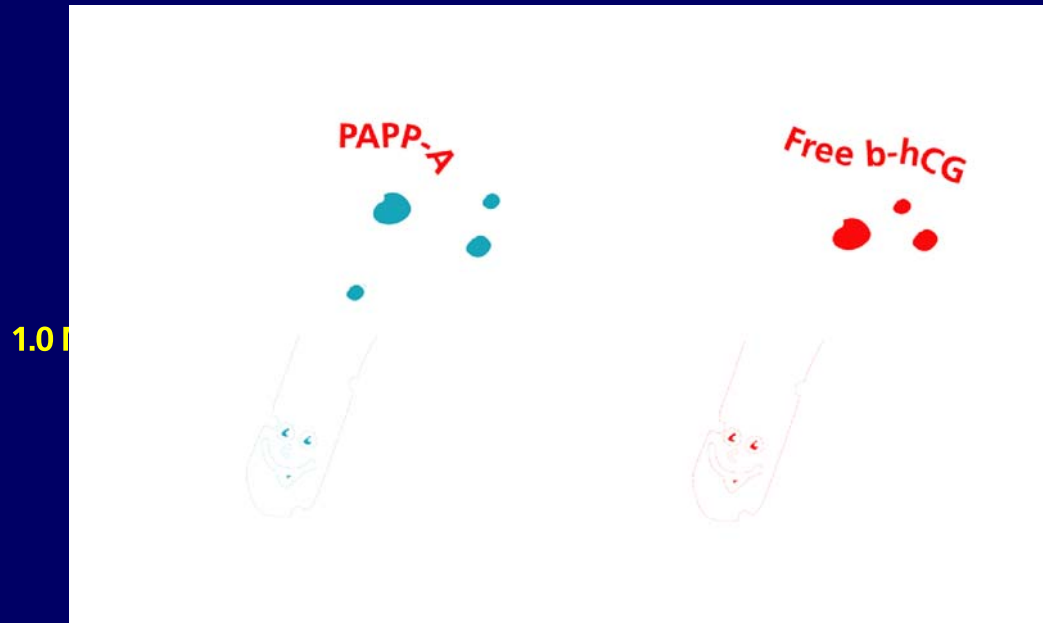
Free β hCG



Chorionic villi

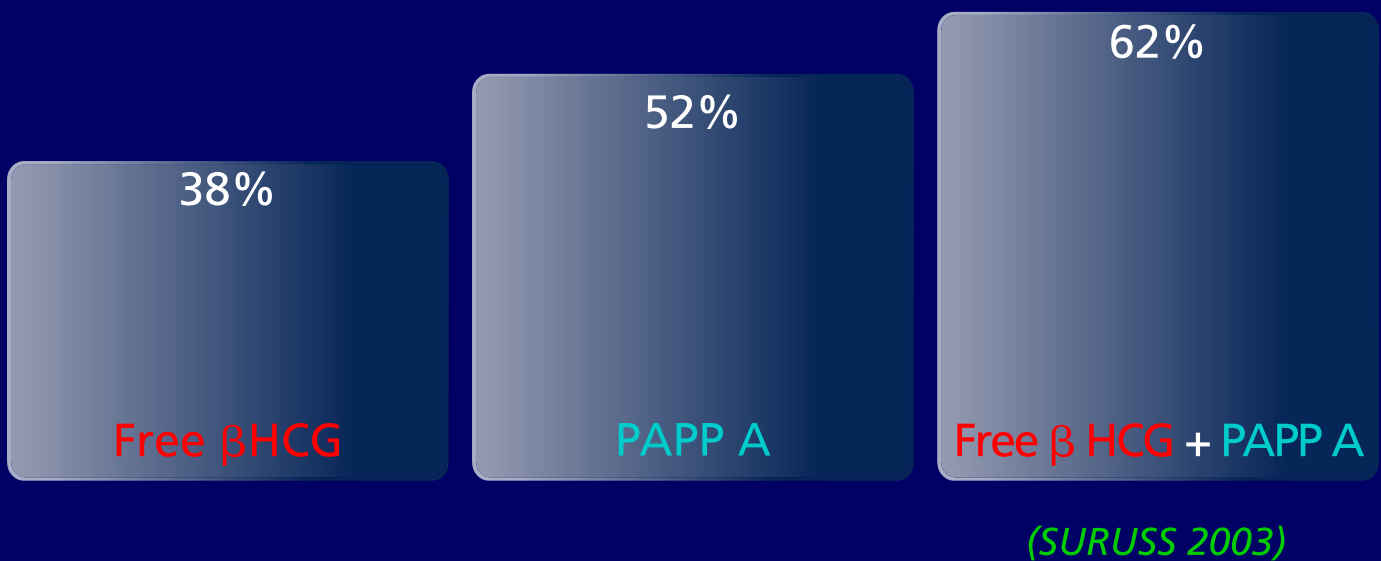


First Trimester Screening Marker Levels in Down's Syndrome Pregnancies



First Trimester Detection Rates

5% False Positive Rate

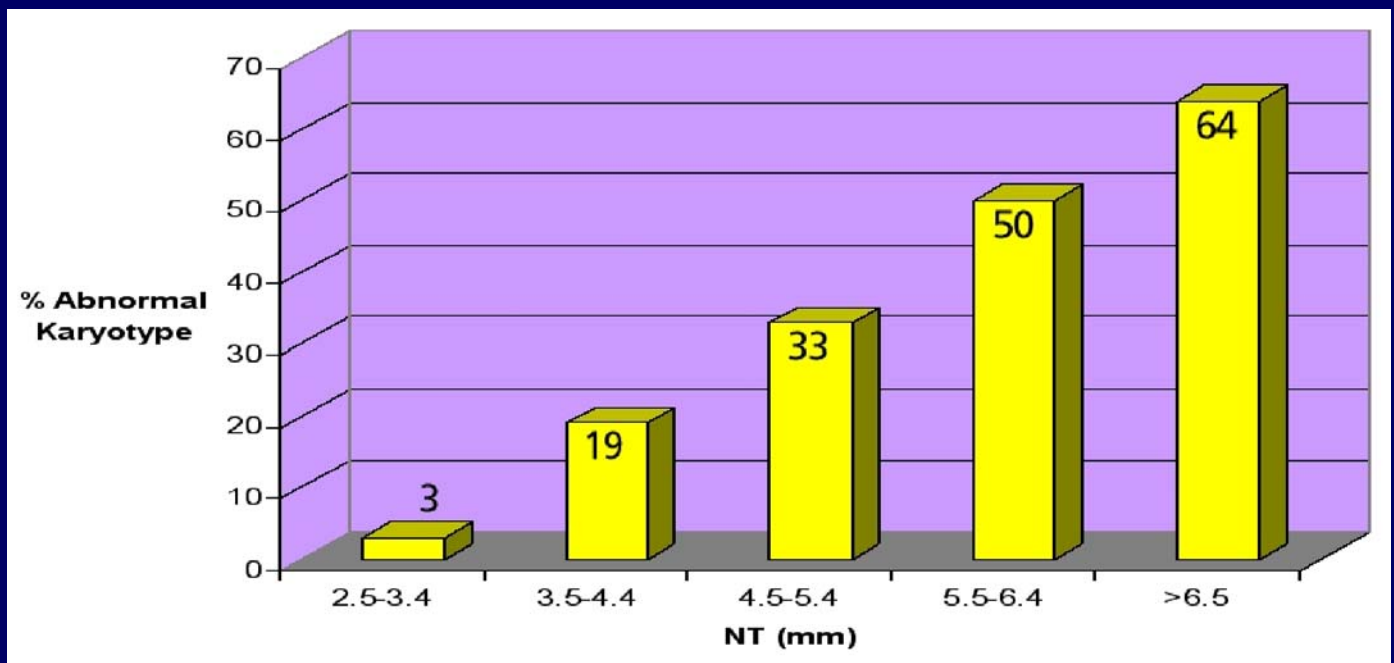


1st Trimester Screening Nuchal Translucency



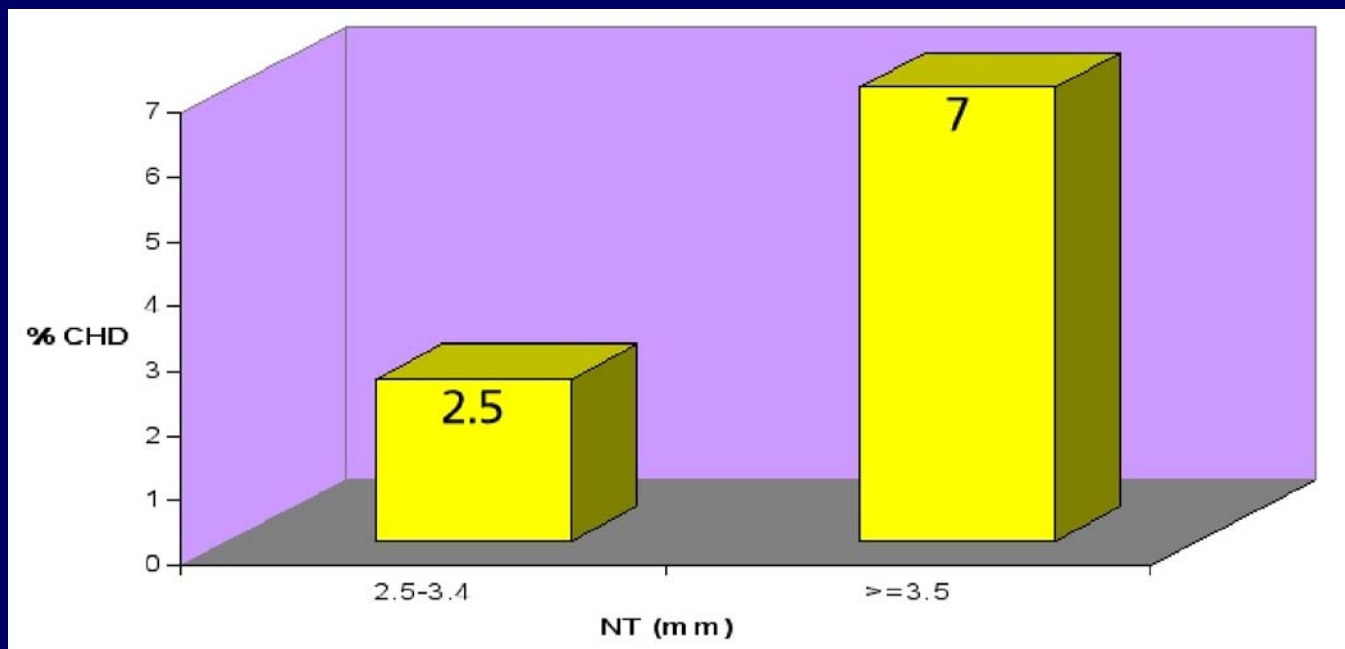
- Measured between 11-13 weeks 6 days
- CRL 45-84 mm

Association Between Increase in NT and Abnormal Fetal Karyotypes



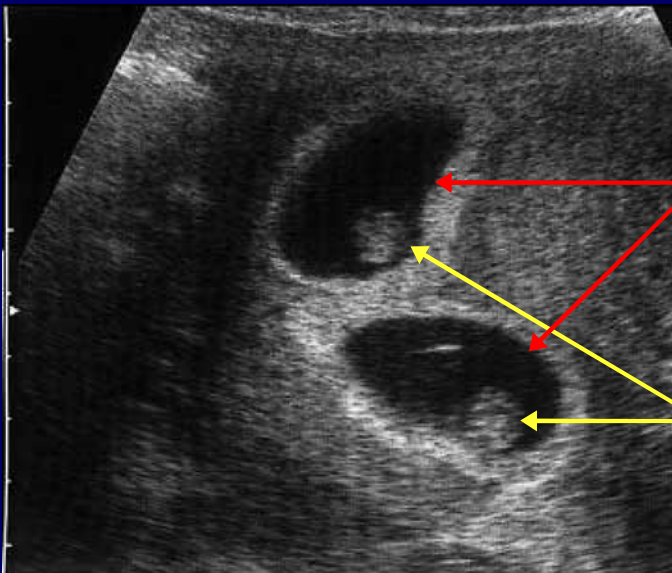
(Snijders et al 1998)

Increased NT is also Associated with Cardiac Defects



(Ghi et al 2001) (n=1321)

Twin Pregnancy



Gestational sacs

Fetal poles

NSC Recommendations

Model of Best Practice (November 2003)

DR of at least 60%, 5% FPR or less by 2004/2005

DR of greater than 75% with a FPR of less than 3% by April 2007