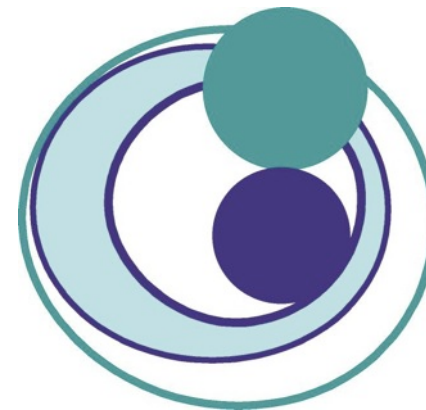


# First Trimester Screening for Aneuploidy

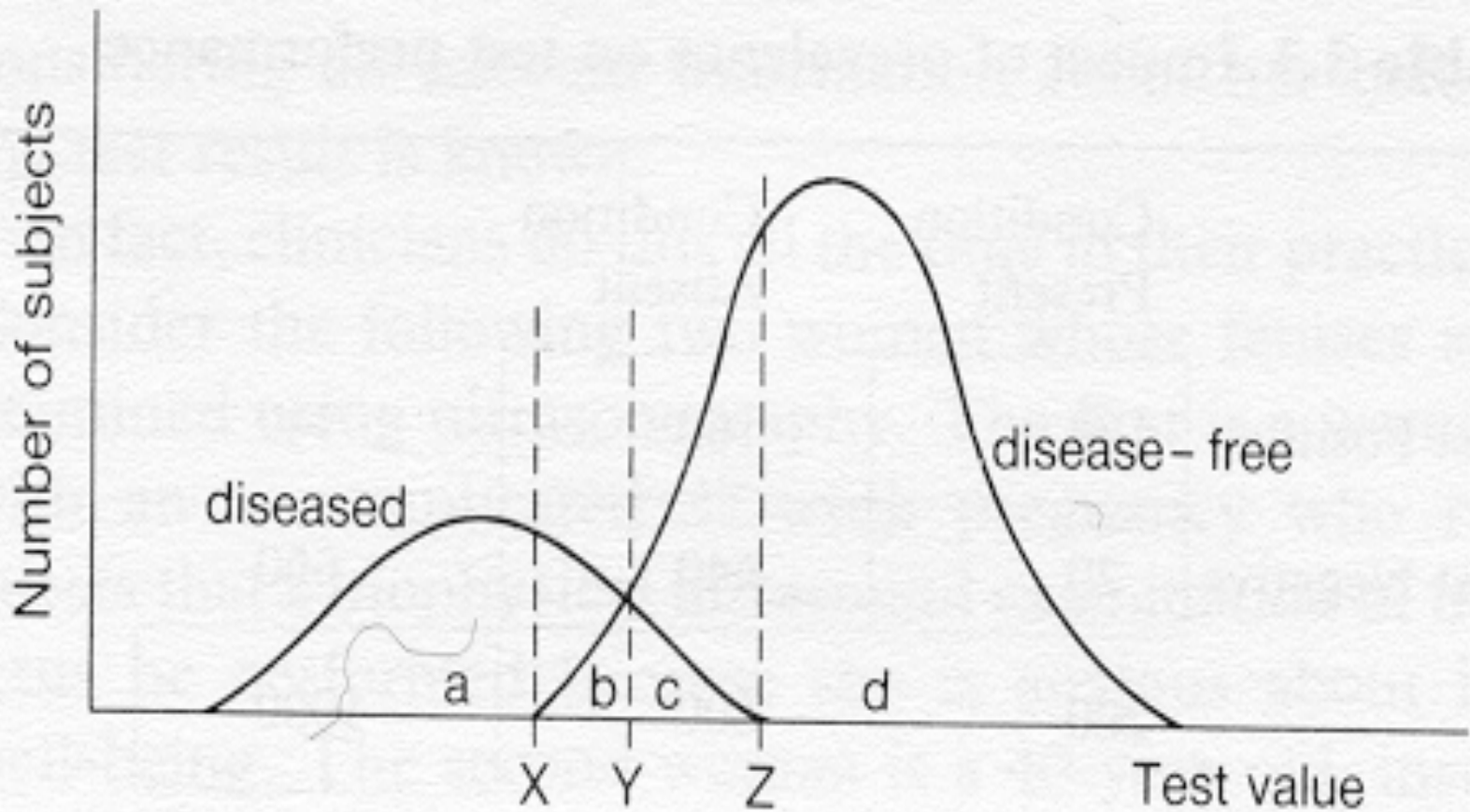
Dr Emma Parry  
Clinical Director Maternal Fetal Medicine  
Auckland District Health Board &  
Sonologist Ascot Radiology



New Zealand  
Maternal  
Fetal  
Medicine  
Network

# What is Screening?

- Screen: an apparatus used in the sifting of grain, coal etc.  
*1573 Shorter Oxford Dictionary*
- A pathway, not a test
- ***Screening is a health service in which members of a defined population...are offered a test to identify those who are more likely to be helped than harmed by further tests... to reduce the risk of a disease or its complications.***  
*( National Health Committee 2003)*



Effect of choice of cut-off on test performance  
x minimises false positives  
z minimises false negatives

# Why screen for aneuploidy?

- Provide information about risk to patients
- Describe choices for invasive testing
- Ensure this information is accurate
- Reassure the majority of women at an early stage
- Include most affected pregnancies in a 'high risk' group

# Antenatal screening for Down syndrome and other conditions

- All pregnant women who are less than 20 weeks pregnant should be advised of the availability of Antenatal screening for Down syndrome and other conditions
- This is optional screening- women make their own choices regarding screening
- This screening provides a single risk estimate for Down syndrome (trisomy21), Edwards syndrome(trisomy18) and Patau syndrome (trisomy13)
- No screening test finds all cases of a condition
- Screening pathway-information she may need to consider
  - Unusual analytes - other conditions
  - Meaning of results- low risk/increased risk
  - Diagnostic testing

# Informed Consent

For consent to be informed it is necessary for health practitioners to have a discussion including:

- the screening and diagnostic processes and results
- the limitations of screening
- the options for women- including not participating in screening or withdrawing from the process at any time.
- additional information as needed
- opportunity to ask questions
- written information
- time to make decisions



# Antenatal screening for Down syndrome and other conditions

Broaching the subject

Would you like to have Antenatal Screening for Down Syndrome and other conditions?

Would you want to know if your baby had special needs or a condition like Down syndrome or if you had problems with your pregnancy?



# Antenatal screening for Down syndrome and other conditions

## Key findings

Six consumer discussion groups pregnant women Maori, Pacific, Chinese, NZ European/mixed ethnicity- 5 groups 32 women and one male group 6 men

- Most women limited understanding of the process and its implications
- Some only discovered they had been screened as part of the consumer group
- Some felt they had been screened without their knowledge
- Did not understand it was both blood test and NT scan
- Confusion about what results meant
- Poor risk literacy amongst practitioners and women
- Most women with a low risk result did not know that they could still have a baby with Down syndrome

Quigley and Watts report for Learning Media and NSU December 2011

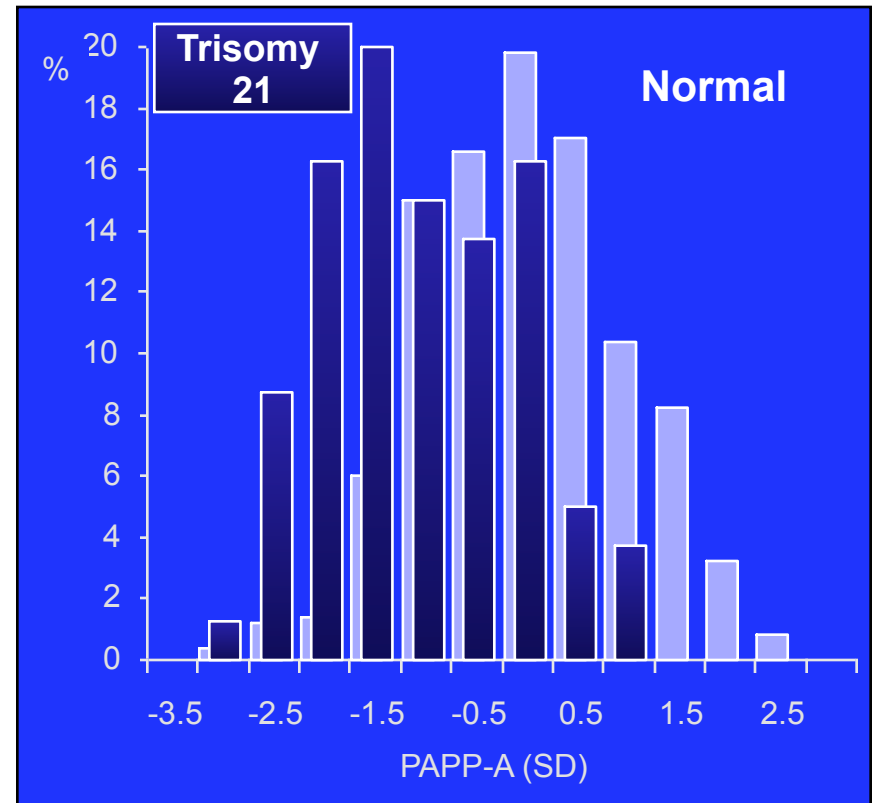
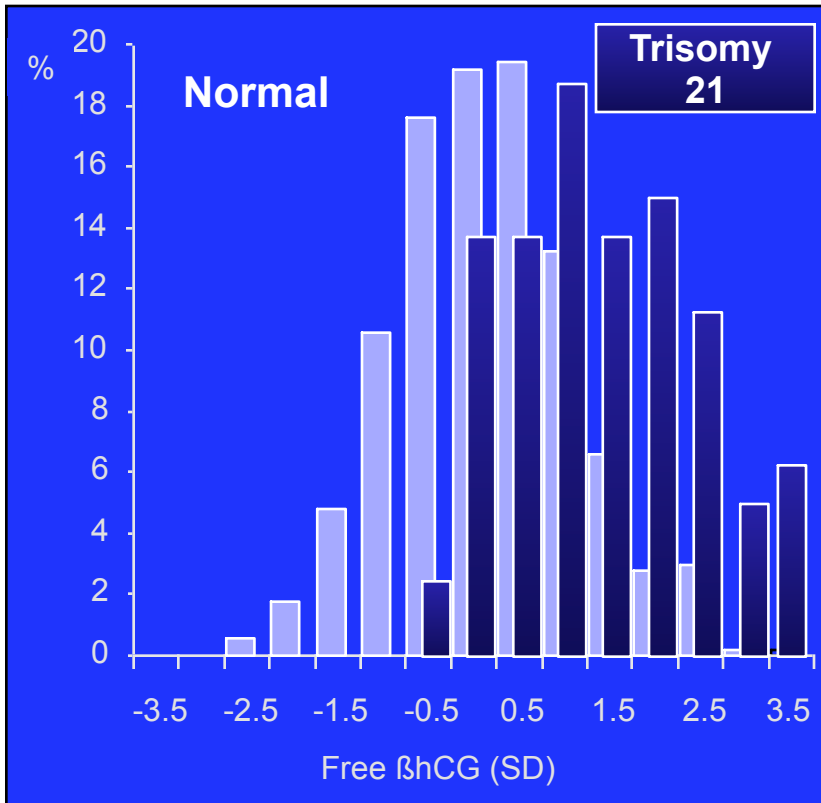


# Maternal Serum analytes

- 1<sup>st</sup> Trimester
  - PAPP-A
  - Free B-HCG
- 2<sup>nd</sup> Trimester
  - Alpha Fetoprotein ) )
  - Oestriol ) Triple Test)
  - Free B-HCG ) ) Quadruple
  - Inhibin-A ) Test

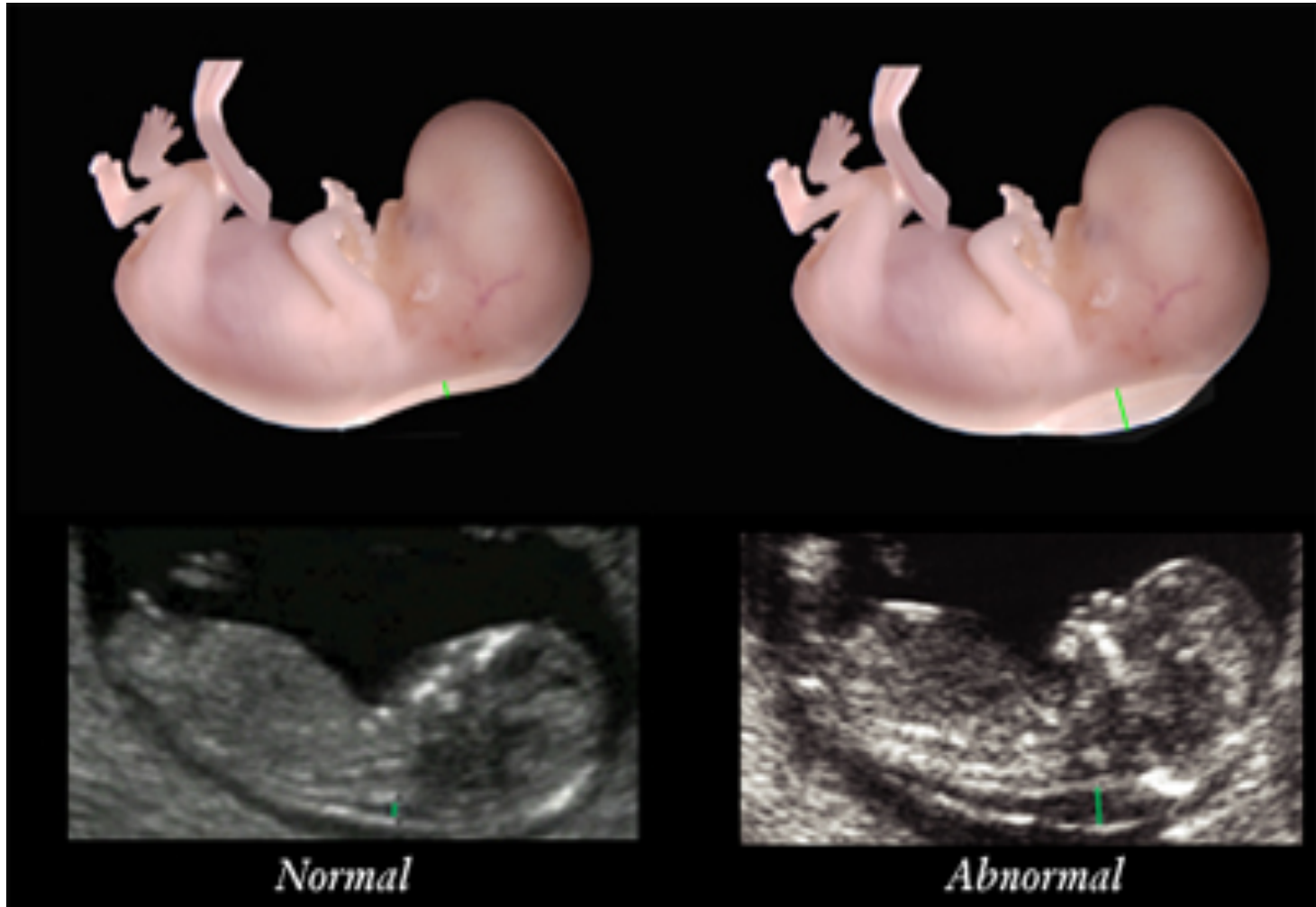
# First trimester screening for trisomy 21

Maternal serum free  $\beta$ -hCG & PAPP-A



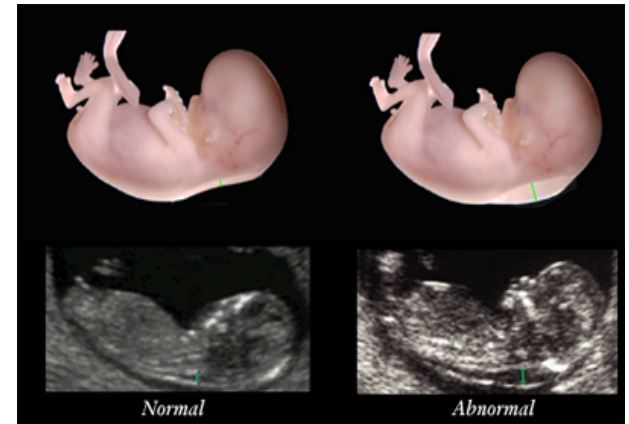
- Detection rates at 12 weeks are similar to those at 16 weeks
- Biochemical changes are independent of fetal NT thickness
- NT, free  $\beta$ -hCG and PAPP-A identifies 90% of cases for FPR of 5%

# Nuchal Translucency



# Nuchal Translucency

- Designed for low risk women (<40 years?)
- USS measurement
  - TA or TV
  - Registered user (FMF)
  - Ongoing audit
- 11+3 to 13+6
- Bayes theory
- Result is a RISK- not a diagnostic test
- Trisomy 13 and 18
- Detection for Trisomy 21 is 85%



# Timing

**Timing is critical and influences the choices available to women:**

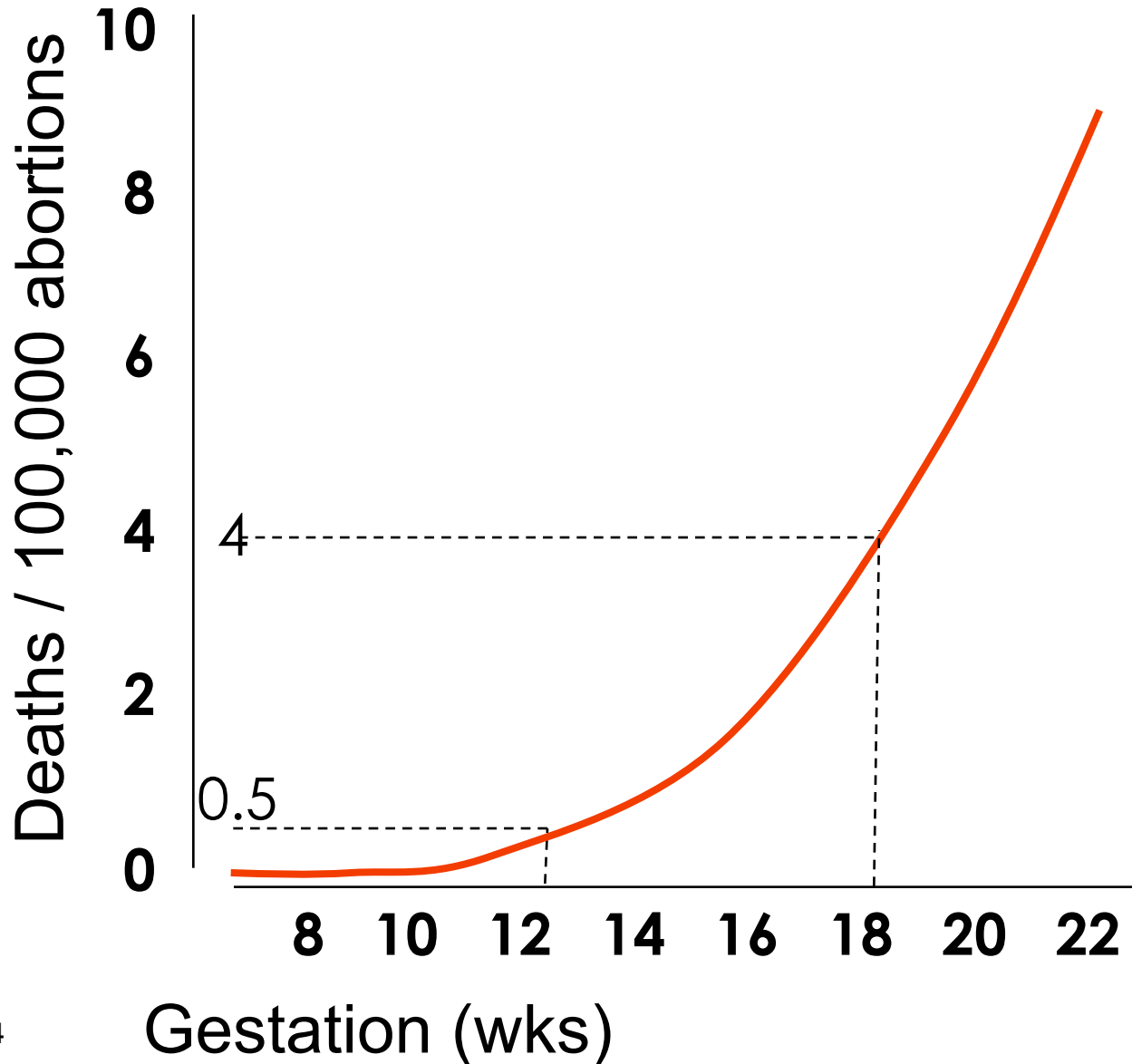
**Combined first trimester screening between 9-13 weeks and 6 days**

- NT Scan between 11 weeks and 2 days and 13 weeks and 6 days - optimum at 12 weeks
- Maternal serum optimum at 9-11 weeks

**Second trimester screening between 14-20 weeks**

- Maternal serum only optimum between 14-18 weeks

# Induced abortion-related maternal mortality: USA 1988-1997



# Results

- Best conveyed to women as **Chance** rather than **Risk**
- Influential moment
- False negative and false positives

*The way you say something can be as powerful as what you say*

# Referral and communication pathways

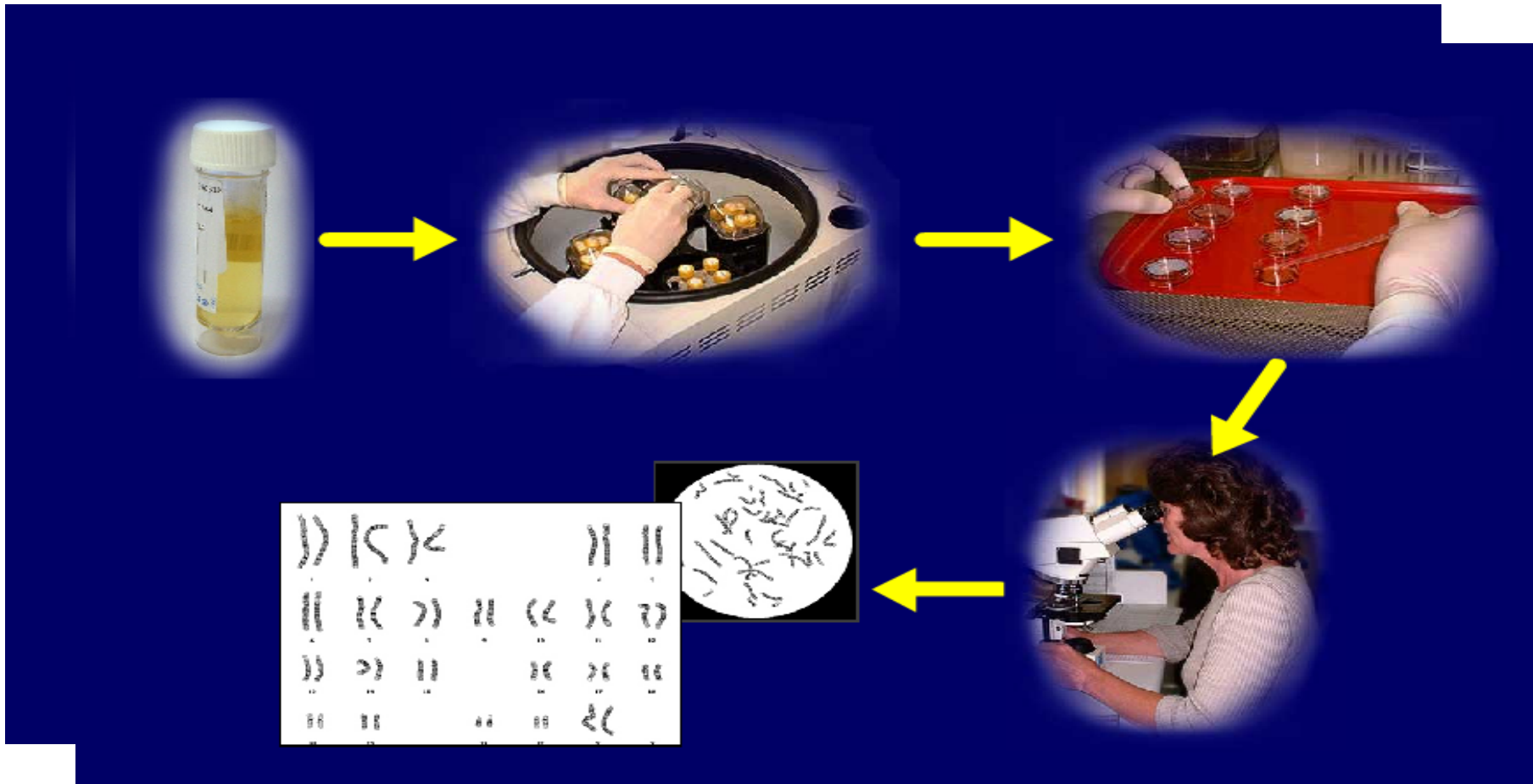
- Maternal anxiety-increased chance result
- Support and information
- Diagnostic testing- timeframes
- Other referral options
- Effective communication
- Referral pathway
- Support groups



# Diagnostic tests

- Chorionic villi sampling (CVS)
- Amniocentesis (amnio)

# Karyotype



# CVS

- Samples tissue for karyotype
- Timing 11-14 weeks
- Provides both advantages and disadvantages
- Carries a number of risks

# Amniocentesis

- Samples amniotic fluid
- Timing 15-20 weeks
- Relatively simple and quick
- Performed in second semester
- Carries a number of risks

# High risk result, low risk for aneuploidy

- High bHCG
- Low Papp-A
  
- Represent a poorly functioning placenta
- Increased risk PTB and PET
- Referral to a general O&G
- Likely increased surveillance in 3<sup>rd</sup> T

# Summary

- Down Syndrome Screening should be offered to all pregnant women
- Counselling should include all possible outcomes and options to ensure screening is right for that individual
- Screening may pick up other problems
- Diagnostic tests have a risk of miscarriage