

# Counseling for Down syndrome screening in the private sector

Des Sankar

# Offering a test

- Describe the test
- Explain the concept of risk
- Discuss implications of a positive result
- Discuss implications of a negative result
- Determine which diagnostic test is acceptable
- Offer information leaflet

# Before counseling...

- We need to know about the screening test that we are offering and therefore I would like to highlight the important points of the available tests

# What are the available screening programs for Down Syndrome?

- First Trimester Screening

- Fetal Medicine Foundation program

- Alpha program

- Second Trimester Screening

- Prieska program

# Summary of FMF FTS Program

<u>Screening test</u>	<u>DR(%)</u>	<u>FPR(%)</u>
MA+fetal NT	70	5
MA+fetal NT+FHR+Papp-A +beta-HCG @ 12w	84	3
MA+fetal NT+FHR+Papp-A +beta-HCG @ 10w	94	3

In South Africa – 41 registered users

FMF Website Lecture by Professor Kypros Nicolaides

# Summary of Alpha FTS Program

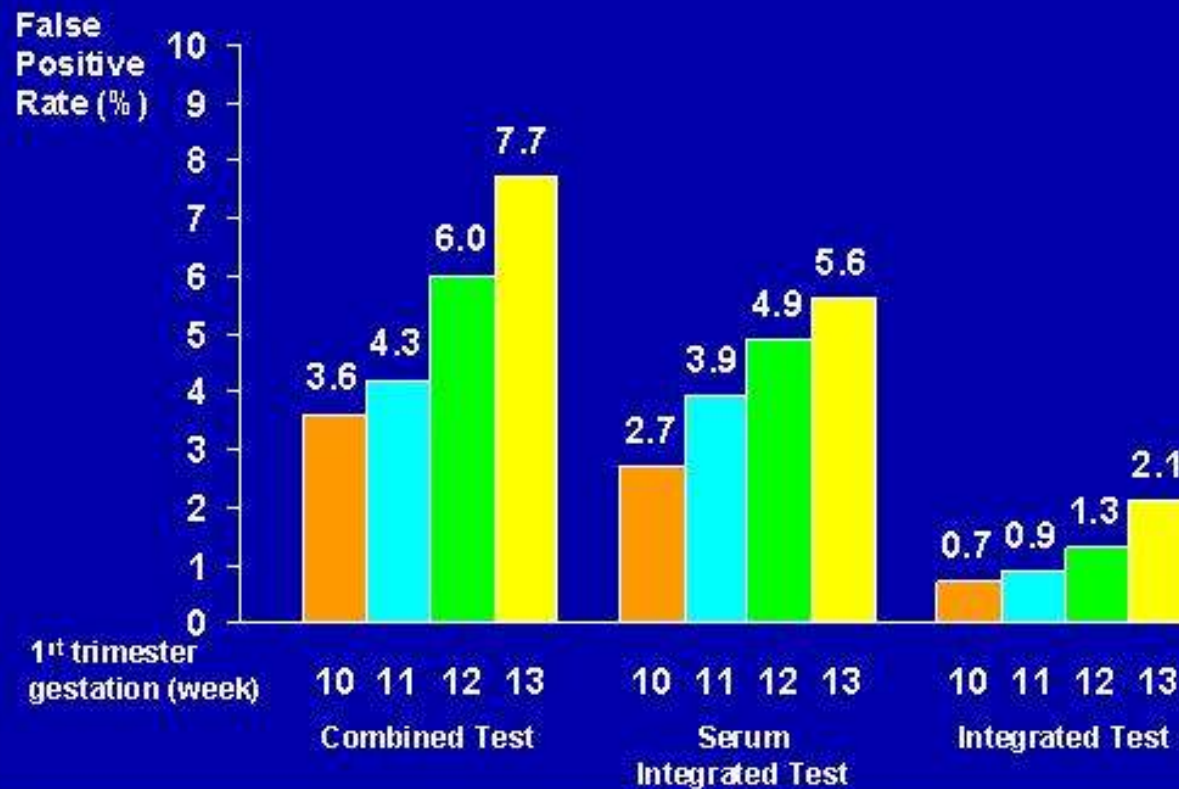
- MA+NT+Papp-A+beta-HCG
- DR 85%
- FPR 5%

PRENATAL DIAGNOSIS, VOL. 17:9: 821-829 (1997)

## COMBINING ULTRASOUND AND BIOCHEMISTRY IN FIRST-TRIMESTER SCREENING FOR DOWN'S SYNDROME

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## False Positive Rate for an 85% Detection Rate



# However..

- Punching in positive Alpha FTS results into the FMF program have yielded different results



# 39yr old/NT 1.2mm/CRL 65.4

## Alpha

■ bHCG MoMs	2.11
■ Papp-A MoMs	1.06
■ B/ground risk	1:55
■ T21 risk	1:180

## FMF

■ bHCG MoMs	1.981
■ Papp-A MoMs	1.115
■ B/ground risk	1:90
■ T21 risk	1:448

# 35yr old/NT 1.9mm/CRL 55mm

## Alpha

- bHCG MoMs 1.83
- Papp-A MoMs 0.98
- B/ground risk 1:160
- T21 risk 1:190

## FMF

- bHCG MoMs 1.637
- Papp-A MoMs 1.229
- B/ground risk 1:226
- T21 risk 1:1769

- Appears to be a higher screen positive rate with the Alpha vs FMF program particularly in the group of patients that are 35 years and older
- Higher rate of invasive procedures
- Higher pregnancy loss rate
- Some obstetricians have reverted back to the use of second trimester screening
- A comparative study needs to be conducted

# Second Trimester Screening

- Triple Test – beta-HCG, AFP & Oestriol
- DR 65-70%
- FPR 5%
- Prieska program
- Lab 1: DR of 50-59%
- Lab 2: DR of 69%

# Laboratory Report

- Both Labs: Risk cutoff : 1:270
- Risk of 1:300 : low risk or screen negative
- Risk of 1:100 : high risk or screen positive
- Some obstetricians have objected to the change in the terminology

- A low risk or negative Down syndrome screen should be combined with a detailed anomaly scan between 18 to 23w
- DR increases to 90%

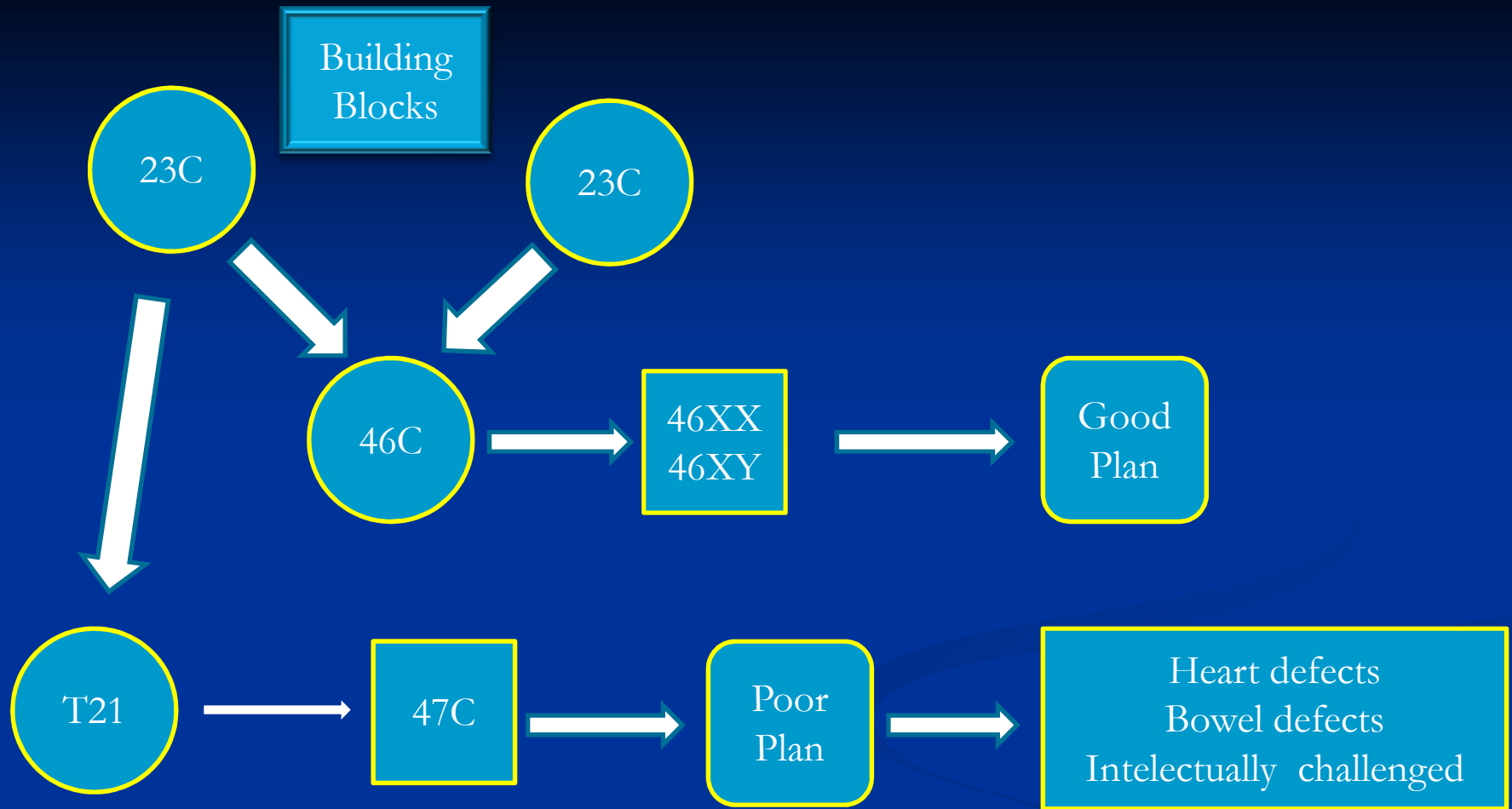
# Newly diagnosed pregnant patient.....

- History
- Examination
- US
- Counseling
  - Exposure to medication
  - Diet/weight gain/sleeping position/clothing
  - Exercise

# Counseling cont....

- Travelling
- Sexuality
- Screening for fetal abnormalities including Down syndrome
- Serological investigations including HIV screening
- Hospitals/costs
- All in 30mins





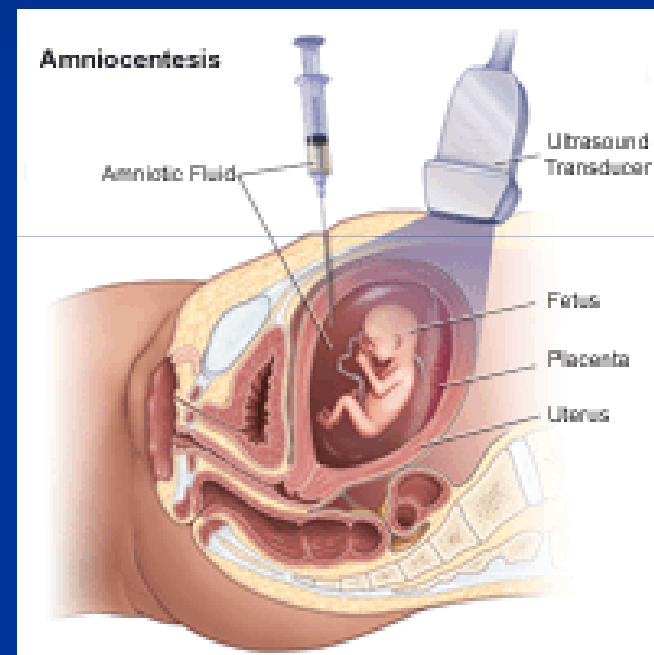
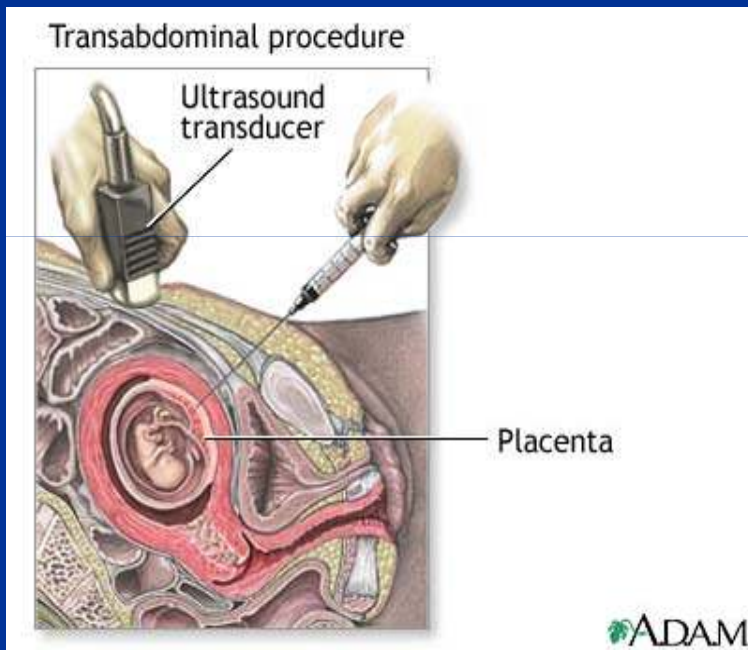
- Now I talk about Down syndrome because this is the commonest chromosomal abnormality in the human being
- Because this is the commonest anomaly, we have developed techniques of detecting Down syndrome

# FTS

- NT alone – DR 70% (miss 30%)
- NT+NB – DR 75% (miss 25%)
- NT+NB+Biochem(9-10w)– DR 94% (miss 6%)
- NT+NB+Biochem(12w)- DR 84% (miss 16%)
- FPR 3%



# Invasive Tests: 1% risk of misc.



# Important questions

- Do you wish to have such screening?
- Will you consider an invasive test if the result was positive?
- What will you do if the baby was proven to have Down syndrome?
- Are you able to cope with a potential loss of a normal pregnancy as a result of the invasive procedure?

# Patients are...

- Requested to consider whether they want screening
- Given the laboratory form with the date on which they should have the test
- Given an appointment for the 12w scan

# 40 years and older...

- Invasive tests should be offered

# The Future

- Cell free fetal DNA in the maternal blood



# Down Syndrome Screening...

- Should be offered to all patients in a non directive manner
- Patients must be given options, particularly those who will not consider pregnancy interruption and who will not accept a 1% risk of pregnancy loss as a result of an invasive test

# Down Syndrome Screening..

- Should never be a 'routine test', as we create undue anxiety when the result is positive or high risk
- We should respect our patient's decision

**Thank you for your attention**

Thanks to Namitha Chabilal for her assistance