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

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

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%

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

N. J. WALD, AND A. K. HACKSHAW
However..

- Punching in positive Alpha FTS results into the FMF program have yielded different results
<table>
<thead>
<tr>
<th>Test</th>
<th>Alpha</th>
<th>FMF</th>
</tr>
</thead>
<tbody>
<tr>
<td>bHCG MoMs</td>
<td>2.11</td>
<td>1.981</td>
</tr>
<tr>
<td>Papp-A MoMs</td>
<td>1.06</td>
<td>1.115</td>
</tr>
<tr>
<td>B/ground risk</td>
<td>1:55</td>
<td>1:90</td>
</tr>
<tr>
<td>T21 risk</td>
<td>1:180</td>
<td>1:448</td>
</tr>
</tbody>
</table>

**39yr old/NT 1.2mm/CRL 65.4**
### 35yr old/NT 1.9mm/CRL 55mm

<table>
<thead>
<tr>
<th>Alpha</th>
<th>FMF</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>bHCG MoMs</strong></td>
<td><strong>bHCG MoMs</strong></td>
</tr>
<tr>
<td>1.83</td>
<td>1.637</td>
</tr>
<tr>
<td><strong>Papp-A MoMs</strong></td>
<td><strong>Papp-A MoMs</strong></td>
</tr>
<tr>
<td>0.98</td>
<td>1.229</td>
</tr>
<tr>
<td><strong>B/ground risk</strong></td>
<td><strong>B/ground risk</strong></td>
</tr>
<tr>
<td>1:160</td>
<td>1:226</td>
</tr>
<tr>
<td><strong>T21 risk</strong></td>
<td><strong>T21 risk</strong></td>
</tr>
<tr>
<td>1:190</td>
<td>1:1769</td>
</tr>
</tbody>
</table>
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
Good Plan

Building Blocks

46C → 46XX 46XY → Good Plan

23C → 46C

T21 → 47C → Poor Plan

Heart defects Bowel defects Intellectually challenged

Poor Plan

Intelectually challenged
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