Conclusion

We suggest that NFM has a negative association with Trisomy 21 with high NPV and may be helpful in counselling. Furthermore UAC seems to be only associated with Trisomy 21 and no other chromosomal abnormality in this population. We suggest further prospective study of this phenomenon. Abnormalities of cell adhesion molecules (encoded on C21) are well described in Down’s (DSCAM – Down’s Cell Adhesion Molecule) and this suggests a possible aetiology.

Abstract PF.42 Table

<table>
<thead>
<tr>
<th></th>
<th>Down’s present</th>
<th>Down’s absent</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>UAC</td>
<td>10</td>
<td>11</td>
<td>21</td>
</tr>
<tr>
<td>NFM</td>
<td>23</td>
<td>381</td>
<td>404</td>
</tr>
<tr>
<td></td>
<td>33</td>
<td>392</td>
<td>425</td>
</tr>
</tbody>
</table>

Sensitivity of UAC = 10/33 = 30.3%
Specificity of UAC = 381/392 = 97.2%
PPV = 10/21 = 47.6%
NPV = 381/404 = 94.3%

Conclusion

In our screening population, median PAPP-A MoM was 1.074 MoM. Within this group of pregnancy cases detected antenatally, gestational age at diagnosis varying size were analysed. For those cases detected antenatally, gestational age at diagnosis was recorded, and the booking hospital anonymised. Eight trusts of varying size were analysed. Results

Although the vast majority of FASP cases were identified before delivery, only the anencephaly target was met by all eight, while Spina Bifida and Trisomy 18 targets were missed by five. One trust reached only four of nine targets, missing three of the others by a single case. However, none of the FASP targets was achieved by 20 + 6 weeks.

Conclusions

Most trusts met expected antenatal detection rates specified by FASP, but not by ≤20 + 6 weeks. Considerable variability exists both between trusts and anomalies. Data produced here should enable the precise training needs of each trust to be identified more accurately.