Isolated ultrasound marker in the first trimester - is invasive testing justified?
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Introduction
- First-trimester screening is a highly effective method to detect aneuploidies
- Routinely used parameters
  - Nuchal translucency
  - Maternal serum biochemistry
- Inclusion of additional markers
  - Nasal bone (NB)
  - Tricuspid blood flow
  - Ductus venosus waveform
  Detection rates 80-90% for FPR of 5%
  Detection rate 92-96% for FPR of 3%

Aim
To assess the strength of an isolated ultrasound marker in the first trimester in predicting aneuploidy in an otherwise structurally normal fetus

Materials and Methods

Study Period
Jan 2005 - Dec 2015
Mean maternal age 30 years

No of fetuses analyzed
(No Structural defects & known outcomes) = 9342

Ultrasound markers assessed
- NT > 95th centile
- Absent/ Hypoplastic NB
- Tricuspid regurgitation
- Abnormal DV waveform

Results

No of fetuses analyzed
(No Structural defects & known KT) = 9342

Significant karyotypic abnormality
33/9342 (0.3%) 1 in 333

<table>
<thead>
<tr>
<th>Abnormal KT</th>
<th>US markers present</th>
<th>US markers absent</th>
<th>Abnormal KT</th>
<th>Abnormal KT 11/429 (2.6%)</th>
<th>1 in 39</th>
</tr>
</thead>
<tbody>
<tr>
<td>24/460 (5.2%)</td>
<td>460/9342 (4.9%)</td>
<td>8882 (95.1%)</td>
<td>9/3882 (0.14%)</td>
<td>1 in 714</td>
<td></td>
</tr>
<tr>
<td>T21</td>
<td>13</td>
<td>T18</td>
<td>47 XXY</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>T18</td>
<td>5</td>
<td>T5</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>45 XO</td>
<td>2</td>
<td>Triploidy</td>
<td>1</td>
<td></td>
<td></td>
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<tr>
<td>Chr 46</td>
<td>1</td>
<td>Unbalanced</td>
<td>1</td>
<td></td>
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<tr>
<td>Mosaic</td>
<td>1</td>
<td>T21</td>
<td>3</td>
<td></td>
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</tr>
<tr>
<td>T8 and M22</td>
<td>1</td>
<td>45XO mosaic</td>
<td>1</td>
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</tbody>
</table>

Conclusions
This is the largest first trimester series in Indian population to provide likelihood ratios for isolated 1st trimester ultrasound markers in "structurally normal fetuses" for detection of fetal aneuploidies

- 4.9% fetuses had ultrasound markers in the IT
- 93% of these fetuses had "Isolated ultrasound marker"
- 11/24 (45.8%) fetuses with an isolated marker had an isolated marker
- Presence of an isolated marker cannot be ignored as 1 in 39 such fetuses which are structurally normal can still be chromosomally abnormal
- Isolated increased NT followed by hypoplastic/ absent NB had a strong association with aneuploidies
- Absence of any marker significantly reduced the risk of aneuploidies, as only 1:714 fetuses had a chromosomal anomaly

Likelihood Ratios: Isolated Increased NT = 18.9
Absent/ hypoplastic NB = 10.2

References

All Scans were performed by FMF certified operators at Bangalore Fetal Medicine Centre
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