The assessment of intracranial translucency at 11-14 weeks in screening for open spina bifida – a case report

Dr Shannon Morris
Fetal Assessment Center
Cape Town
Screening for OSB in the first trimester

- In the last decade there has been wide uptake of the first trimester scan - specifically to screen for T21 by nuchal translucency measurement.
- This has also led to the detection of many other major fetal abnormalities at 11-14 weeks.
- However, the diagnosis of spina bifida before 14 weeks remains elusive.
- Buisson et al (UOG 2002) described the following indirect signs at 12 weeks:
  - Narrowing of the frontal bones at the BPD level, so called “acorn” sign.
  - Parallel peduncles on the BPD level.
  - Displacement of the meta-encephalon on a sagittal view of the fetal head and neck (same midline view required for nuchal translucency measurement).
- These are subtle signs and difficult to recognize.
Intracranial translucency (IT)

ISUOG congress: Hamburg September 2009

Prof Rabi Chaoui presented a “new sign” as possible screen for OSB at 11-14 weeks (absent IT)

• Mid-sagittal view of the fetal face – same view as used for measurement of NT + assessment of nasal bone

• Brain stem and fourth cerebral ventricle are easily visible
  – The fourth ventricle presents as an intracranial translucency (IT)
  – Parallel to the NT and is delineated by two echogenic borders

• Between the fourth ventricle and the occiput there is another thinner translucency generated by the developing cisterna cerebellomedullaris.

Chaoui et al (UOG 2009;34: 249 – 252)
Ultrasound image in the mid-sagittal plane of the fetal face showing the nasal bone, palate, mandible, nuchal translucency (NT), thalamus (T), midbrain (M), brain stem (B) and medulla oblongata (MO). The fourth ventricle presents as an intracranial translucency (IT) between the brain stem and the choroid plexus. UOG 2009; 34: 249-252
Absent Intracranial translucency and OSB

- In some cases of open spina bifida the fourth ventricle is not visible (absent IT)
- This is as a result of leakage of cerebrospinal fluid into the amniotic cavity, causing hypotension in the subarachnoid spaces leading to downward displacement of the brain
  - The “banana” and “lemon” signs seen at 16-24 weeks
- The downward displacement of the fetal brain is visible as absent IT in the first trimester

Chaoui et al (UOG 2009;34: 249 – 252)
Absent intracranial translucency

Ultrasound image in the mid-sagittal plane of the fetal face in a case of open spina bifida demonstrating compression of the fourth ventricle with no visible translucency. B = brain stem; M = midbrain; MO = medulla oblongata; T = thalamus. UOG 2009; 34: 249-252
Clinical Case

- 38 year old primigravida referred for routine first trimester scan
- Ethnic origin: Asian
- Maternal weight: 68kg
- Smoker
- Spontaneous conception
- Rhesus: positive
- Preconception folic acid: No
Clinical Case: US report

**Ultrasound**
- **Operator**: Dr. Shannon Morris
- **US system**: GE Voluson Expert
- **Probe**: 4-8MHz transabdominal
- **View**: good
- **Gestational age**: 13 weeks + 4 days

**First Trimester Ultrasound**
- **Findings**: Alive fetus
- **Fetal heart activity**: present
- **Fetal heart rate**: 137 bpm
- **CRL**: 70.0 mm
- **NT**: 2.9 mm
- **BPD**: 19.0 mm
- **HC**: 72.0 mm
- **AC**: 63.0 mm
- **FL**: 9.0 mm
- **Nasal bone**: present
- **Ductus venosus Doppler**: abnormal (reversed)
- **Skull/brain**: Lemon sign
- **Spine**: Spina Bifida
- **Heart**: VSD
- **Abdominal wall**: appears normal
- **Stomach**: visible
- **Bladder / Kidneys**: Bladder not seen
- **Hands**: both visible
Clinical Case: US report

- Feet: both visible
- Other: Fetal sex: apparently male
- Placenta: anterior high
- Amniotic fluid: normal
- Cord: abnormal
  - single artery
- Cord cysts: 15 x 12 x 8 mm

**Biochemistry**

- Sample taken: 09/11/2009
- Maternal weight: 68.0 kg
- Analysed on: 09/11/2009
- Kit manufacturer: BRAHMS Kryptor
- Free ß-hCG: 11.0 IU/l, 0.199 MoM
- PAPP-A: 0.897 IU/l, 1.079 MoM

Patient counselled and consent given - Maternal age: 38 years

<table>
<thead>
<tr>
<th></th>
<th>Trisomy 21</th>
<th>Trisomy 18</th>
<th>Trisomy 13</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Background risk:</strong></td>
<td>1: 115</td>
<td>1: 271</td>
<td>1: 854</td>
</tr>
<tr>
<td><strong>Adjusted risk:</strong></td>
<td>1: 277</td>
<td>1: 124</td>
<td>1: 1192</td>
</tr>
</tbody>
</table>
Fig 1: Absent intracranial translucency
Fig 2: Increased NT
Fig 3: Single UA
Fig 4: Cord cyst
Fig 5: Parallel peduncles
Fig 6: Lesion on fetal spine
Fig 7: VSD
Clinical Case

- In summary:
  - Low heart rate
  - IUGR
  - Increased nuchal translucency
  - Signs of OSB on scan
    - Absent IT
    - Parallel peduncles
    - Acorn sign
    - Lesion visible on the sacral region of the fetal spine
  - VSD with reversed flow in the ductus venosus
  - Single umbilical artery with cord cyst
  - Fetal bladder not seen
  - Low free BHCG
- This patient screened high risk for an underlying aneuploidy following the 1st trimester scan – specifically T18
Clinical Case

- An uncomplicated CVS with PCR was performed
- Karyotype confirmed Trisomy 18
- The patient terminated the pregnancy
- Advice for the next pregnancy
  - Preconception folic acid for 6 months
  - 11-14 week scan with first trimester biochemistry at 10 weeks
Take home message

• Examination of the mid-sagittal view of the face is performed routinely for assessment of NT and nasal bone

• If the 4th ventricle is not seen – absent IT
  – Consider OSB
  – Look at fetal spine closely

• You will only see what you are looking for……