Over 30 years I was talking, lecturing about & performing 1st trimester fetal anatomy scans
(Sometimes I also included 15 & 16 w)
Few in the world (leave alone my Israeli colleagues) even paid attention.

At the basis of my 1st trimester anatomy scan stood the transvaginal Elscint probe I helped design......

....as well as compiling our experience in the first book on TVS (in1987)
• Hesitant, journal editors
• Disbelief in early anatomy scan
• No USA outcome studies
• Pertinent supporting articles only published in European journals which USA docs seldom read
• No endorsement from AIUM, SPO, ACR, SRU
• No/poor reimbursement

I was still optimistic since at that time the pictures were superior to those created by the transabdominal probes. How could everybody miss this fact?

This is what I wrote then: “I am ... hopeful that those involved will do everything in their power to make the technique available to all pregnant women in the United States and to elevate the image of sonography for fetal malformation screening in the United States”.

My answer was Here in the USA it took us 5 years to evaluate its introduction in clinical practice.

In 2013 a work group was convened to formulate the possible use of US in general as well in the the first trimester and suggest its evidence based use.

• It was sponsored by the Joint Eunice Kennedy Shriver National Institute of Child Health and Human Development and SMFM, AIUM, ACOG, ACR, SPR, and SRU

Uma M. Reddy, MD, MPH, Alfred Z. Abuhamad, MD, Deborah Levine, MD, George R. Saade, MD for the Fetal Imaging Workshop Invited Participants*

The single paragraph dealing with 1st trimester anatomy scan:

“Offering NT screening for aneuploidy assessment at 11 to 13 6/7 weeks’ gestation is part of standard of practice in the U.S.”

“if a late 1st trimester US is performed for dating or NT assessment, evaluation for early detection of severe fetal anomalies such as anencephaly and limb-body wall complex is reasonable. In some experienced centers, detection of other major fetal anomalies in the first trimester is possible.” [15-19]
And finally..... in 2018 I got an email from Bryann Bromley and Alfred Abuhamad to join a wide range of Ob/Gyns representing the pertinent Ob/Gyn/Rads/MFM Societies in an effort to study whether the time is ripe to introduce and offer First Trimester Anatomy Scan in the USA.

After endless live and electronic meetings The Document was conceived, changed, examined and reexamined, rewritten until it became ready to be presented for ratification to the involved Societies.

Ladies and Gentlemen, Sonographers, Obstetricians, MFM Specialists, Geneticists and other interested parties, let me give you a sneak preview of “THE PRODUCT”

This is a “sneak preview” of the future implementation of the First Trimester Anatomy Scan. At the present it is almost at the end of the “pipeline” awaiting FINAL approval of the involved societies.

Disclaimer
- I have no conflicts of interest in this talk
**Introduction**

- Over the past 30 years there has been a trend toward performing the *initial fetal anatomical survey* earlier and earlier during pregnancy.
  - First there was the ‘18-week’ scan
  - Then the ‘16-week’ scan
  - Followed by the ‘14-week’ scan
  - *Now the ‘12-13 6/7 week’ scan*
- Are we ready for this scan?

- To facilitate this move a Task Force Group with representatives of several OB governing groups was convened to decide on the list of the structures.
- Some structures were deemed *‘mandatory’* to be seen in all studies
- Other structures to be assess only *if indicated or suspicious*

**General**

- Images should acquired with appropriate attention to *magnification, depth and focal zone*
- Anatomic structures should be evaluated in *at least one plane* of imaging
- It is recognized that in some imaging situations not all mandatory landmarks will be visualized, and follow-up imaging may be recommended.

**The Scan**

<table>
<thead>
<tr>
<th>Scanning planes/structures</th>
<th>Mandatory</th>
<th>Only if indicated or suspicious</th>
</tr>
</thead>
<tbody>
<tr>
<td>General</td>
<td></td>
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<tr>
<td>Output key standard (ODS)</td>
<td></td>
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</tr>
<tr>
<td>Appropriate ODS Thermal Index Bone ratio ≤ 0.7</td>
<td></td>
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<tr>
<td>Fetal cardiac motion</td>
<td></td>
<td></td>
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<tr>
<td>Number of fetuses and gestational sacs</td>
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<tr>
<td>If multiple gestation</td>
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<td></td>
</tr>
</tbody>
</table>

**DIAGNOSTIC**

<table>
<thead>
<tr>
<th>Ovulation</th>
<th>Mandatory</th>
<th>Only if indicated or suspicious</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fetal head</td>
<td></td>
<td></td>
</tr>
<tr>
<td>N. ANXAL</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Transfontanelle</td>
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<td></td>
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<tr>
<td>Crown boss (bowl-variaans)</td>
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<td></td>
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<tr>
<td>Palpe cerebri</td>
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<td></td>
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<tr>
<td>Vernacles</td>
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</tbody>
</table>

**Anatomy**

<table>
<thead>
<tr>
<th>Ovulation</th>
<th>Mandatory</th>
<th>Only if indicated or suspicious</th>
</tr>
</thead>
<tbody>
<tr>
<td>Crown boss</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Biplane diameter</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Head circumference</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Abdominal circumference</td>
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</tr>
</tbody>
</table>
Fetal thorax continued

Four-chamber view with color Doppler

Transverse aortic arch (may be evaluated as conical shape)

Truncus valve flow

Fetal thorax continued

Ribs with normal shape, length and ossification

Diaphragm examination

Contrast: While the use of Doppler should be limited to the first trimester, color flow imaging is used to exclude the fibroelastic septation, involving the central display window (CDS) in case of...
Mandatory

Only if indicated or suspicious

Notes:
• There were some minor changes suggested by some societies.
• This may be the final version.
• Suggestion: start training the sonographers and physicians involved in the actual scanning so all are ready to implement it when approved.
### Head
- Acrania
- Exencephaly
- Encephalocele: Anterior – Occipital - Parietal
- Craniorachischisis
- Iniencephaly
- Abnormal shape of the head: Brachycephaly - Dolichocephaly
  - Trigonocephaly (strawberry shaped head)
  - Hypomineralised skull

### Brain
- Unclassified severe anomalies of the brain
- Holoprosencephaly
- Amniotic band related brain lesions
- Ventriculomegaly
- Choroid plexus cyst (CPC)
- Striatal cysts
- Spongiform choroid plexus
- Arachnoid cysts
- Midline cysts
- Dandy Walker syndrome
- 4th ventricle cysts
- Blake pouch cysts

### Spine
- Spina bifida:
  - Brain (cranial signs)
  - Meningomyelocele
  - Rachischisis
  - Closed spina bifida
  - Fetal tail
  - Sirenomelia
  - Hemivertebra
  - Diastematomyelia
  - Caudal regression
  - Kyphosis
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12 0/7 weeks

Face
• Midfacial hypoplasia (flat face)
• Anophtalmia
• Microptalmia
• Cyclopia
• Arhinia
• Proboscis
• Facial clefts: Midline cleft - Bilateral cleft - Unilateral cleft
  • Atypical cleft – amniotic band
  • Micrognathia

12 3/7 weeks

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13 3/7 weeks

Neck, skin, effusions and hydrops
• Cervical cysts/jugular lymphatic sacs
• Cystic hygroma
  • Nuchal edema
  • Nuchal cyst
  • Scalp cysts
  • Lymphatic cysts
  • Early fetal hydrops
  • Pleural effusion
  • Pericardial effusion
  • Ascites

12 weeks

Chest
• Congenital diaphragmatic hernia (CDH)
  • Left
  • Right
  • Lung agenesis
  • Small chest
• Congenital High Airway Obstruction Syndrome (CHAOS)

12 2/7 weeks

Heart (congenital heart defects = CHD)
• Transposition of great arteries (TGA)
• Tetralogy of Fallot (TOF)
• Double outlet right ventricle (DORV)
• Hypoplastic left heart syndrome (HLHS)
• Atrioventricular septal defect (AVSD)
• Coarctation of aorta (CoA)
• Absent pulmonary valve
• Bidirectional flow in aorta
• Ventricular septal defect (VSD)
• Hypoplastic aortic arch
• Right and double aortic arch
• Aortic stenosis
• Pulmonary stenosis
• Mitral atresia
• Ebstein anomaly
• Tricuspid dysplasia
• Tricuspid atresia
• Pulmonary atresia with intact ventricular septum (PAIVS)
• Univentricular heart
• Ebstein atroventricular septal defect (AVSD)
• Dextrocardia
• Left atrial isomerism (LAi)
• Right atrial isomerism (RAi)
• Polyacardia
• Congenital heart block
• Dying fetus
• Tachycardia
• Cardiomegaly
The four-chamber view demonstrates a ventricular septal defect (VSD). The 3VVT shows an interrupted aortic arch (IAA) (arrows).

Vessels
• Single umbilical artery (SUA)
• Persistent vitelline artery
• Agenesis of ductus venosus (DV)
  • Intrahepatic
  • Extrahepatic
• Interrupted inferior vena cava (IVC) and azygos continuation
• Persistent right umbilical vein (PRUV)
• Persistent left superior vena cava (PLSVC)

Intrabdominal, renal and bladder
• Bilateral renal agenesis
• Unilateral renal agenesis
• Megacystis
  • Lower urinary tract obstruction (LUTO)
  • Posterior urethral valves (PUV)
  • Urethral atresia
  • Meatal stenosis
  • Renal pelvic dilatation (RPD) and hydronephrosis
  • Hydroureter
  • Echogenic kidney
  • Cystic kidney

• duplex kidney
• Pelvic kidney
• Horseshoe kidney
• Cloacal anomaly
• Patent urachus
• Allantoic cysts
• Intraabdominal cysts
• Anal atresia
• Bowel dilatation

• Megacystis

Abdominal Wall Defect
• Exomphalos (omphalocele):
  • Bowel - Bowel and liver
  • Gastrochisis: Bowel - Bowel and liver
  • Pentolegy of Cantreil
  • Bladder extrophy
  • Cloacal extrophy
  • OEIS complex (omphalocele-extrophy-imperforate anus-spinal)
  • Body stalk anomaly

Lower urinary tract obstruction (LUTO)
Posterior urethral valves (PUV)
Urethral atresia
Meatal stenosis
Abdominal Wall Defect

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  - Body stalk anomaly

Extremities

- Polydactyly: Hands - Feet
- Syndactyly
- Ectrodactyly (Claw hand)
- Clenched hand
- Transverse arm defects
  - Radial aplasia
  - Amniotic band related limb anomalies
  - Focal femoral dysplasia
  - Amelia
  - Short limbs
  - Talipes
  - X deformed legs
  - Fetal akinesia deformation sequence (FADS)
  - Intrauterine fractures

Skeletal Dysplasia

- Thanatophoric dysplasia
- Osteogenesis imperfecta (types II-III)
- Achondrogenesis
- Short rib polydactyly
- Chondrodysplasia punctata
- Diastrophic dysplasia
- Hypophosphatasia
- Jarcho Levin syndrome

Chromosomal syndromes

- Trisomy 21
- Trisomy 18
- Trisomy 13
- Turner’s syndrome
- Triploidy maternal (digynic)
- Triploidy paternal (diandric)
- DiGeorge syndrome (22q11 deletion)

Early growth restriction and a large head; small placenta
**Placenta and amniotic fluid**

- Partial mole
- Placental cysts
- **Chorionic bump**
  - Anhydramnios
  - Oligohydramnios
  - Baby in the envelope
- Amniotic band syndrome
- Intrauterine synechia
- Velamentous cord insertion
- Umbilical cord cysts
- Pregnancy in scar
- Subchorial haematoma

**First Trimester Screening Sensitivity**

- Evaluation of major anomalies in low-risk population
  - Sensitivity 46.10%
  - [36.88-55.46]

- Evaluation of all anomalies in low-risk population
  - Sensitivity 32.35%
  - [22.45-45.12]

- Evaluation of all anomalies in high-risk population
  - Sensitivity 66.29%
  - [43.47-85.69]

**Multiple Pregnancy**

- Twins
  - Dichorionic
  - Monochorionic
  - Monoamniotic
  - Conjoined
- Twin with abnormal co-twin
  - Growth discordancy
  - Triplets
  - High order pregnancies
  - Twin reversed arterial perfusion (TRAP) sequence or acardiac twinning
  - Molar pregnancy with normal co-twin

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**In Conclusion**

- The driven anatomy scan at 12-13 6/7 weeks will soon become routine practice
- It will be offered in indicated and/or high-risk cases.
- This early anatomical scan can detect a significant number of fetal malformations
Thank You
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