

Gottesfeld-Hohler Memorial Ultrasound Conference 2019



**FIRST TRIMESTER ANATOMY SCAN**  
Position of heart

**First trimester diagnosis- the time has come!**

Ana Monteagudo, MD  
Ilan Timor-Tritsch, MD



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First a personal historical prospective of the First Trimester Anatomy Scan

2

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

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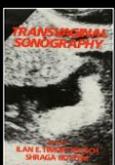
From the time I started to use my first transvaginal probe and saw the resolution it rendered gynecologic US images, I thought about using it to scan the fetus in the first trimester.

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



....as well as compiling our experience in the first book on TVS (in1987)



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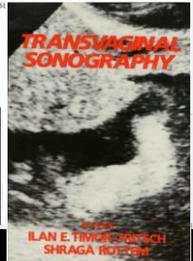
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2. The First Edition of the Book *Transvaginal Ultrasound* by Ilan E. Timor-Tritsch and Shraga Rottem (1987).

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- 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

**Transvaginal Sonographic Evaluation of Fetal Anatomy at 14 to 16 Weeks**

Why Is This Technique Not Attractive in the United States?

**My answer was**

Jan E. Tenor-Reddy, MD  
 Department of Obstetrics and Gynecology  
 Division of Ultrasound  
 New York University School of Medicine  
 New York, New York

J Ultrasound Med 20:705-711, 2001

I was still optimistic.....since at that time the

“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”.

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© Elsevier. Ultrasound Clin 2013, 41, 102-113  
 Published online in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/ulq.12342

**isuog.org** **GUIDELINES**

ISUOG Practice Guidelines: performance of first-trimester fetal ultrasound scan.

**ISUOG published in 2013 its guidelines for the 1<sup>st</sup> trimester US scan**

ISUOG practice guidelines: performance of first-trimester fetal ultrasound scan.

Assessment Obstet Gynecol. 2013 Jun;41(1):102-13. doi: 10.1002/ulq.12342.  
 Salomon LJ, Alfirevic Z, Blaineau OJ, Chalouhi GE, Cho T, Karam AO, Lau TK, Pappasopoulos AT, Paine-Farnham AJ, Shrimpratt J, Suneh S, Taniguchi H, Toza T, et al.

Here in the USA it took us 5 years to evaluate its introduction in clinical practice

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**AIUM Practice Guideline for the Performance of Obstetric Ultrasound Examinations**

**ACOG PRACTICE BULLETIN**

**AIUM has a guideline for anatomy scan. However it was intended for the second trimester**

**Essential Elements of Standard Examination of Fetal Anatomy**

Face and head

- Skull
- Midline
- Frontal plane
- Profile image
- Profile image of cerebral ventricles
- Eye balls
- Forebrain
- Cervical spine
- Cervical spine

Abdomen

- Stomach (presence, size, and situs)
- Gallbladder
- Bladder
- Umbilical cord insertion site into the fetal abdomen
- Umbilical cord vessel number
- Spine-Cervical, thoracic, lumbar, and sacral spine
- Extremities-Legs and arms (presence or absence)
- Sex-Medically indicated in low-risk pregnancies only for the evaluation of multiple gestations

Measurement of the nuchal fold may be helpful during a specific age window to suggest an increased risk of aneuploidy.

American College of Radiology. ACR practice guideline for the performance of obstetric ultrasound. In: ACR practice guideline and technical manual, 2012. Reston (VA): ACR, 2007. p. 1023-1032.

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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\*

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**Due to its attributed clinical importance it was published in the three most important Ob/Gyn Journals**

Am J Obstet Gynecol. 2014 May;210(5):387-97.  
 Obstet Gynecol. 2014 May;123(5):1070-82.  
 J Ultrasound Med. 2014 May;33(5):745-57. .

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**Here is the “Mouse”**

The single paragraph dealing with 1<sup>st</sup> 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** 1<sup>st</sup> 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**.”<sup>15-19</sup>

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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.

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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.

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

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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.

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## Disclaimer

- I have no conflicts of interest in this talk

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### 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<sup>6/7</sup> week' scan**
- Are we ready for this scan?

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### Introduction

- 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*

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### General

- Images should be 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.

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### The Scan

Scanning planes/structures	Mandatory	Only if indicated or suspicious
<b>GENERAL</b> Output <input type="radio"/> Play standard (ODS)		
Appropriate ODS Thermal Index Bone ratio ≤ 0.7		
Fetal cardiac motion		
Number of fetuses and gestational sacs if multiple gestation		

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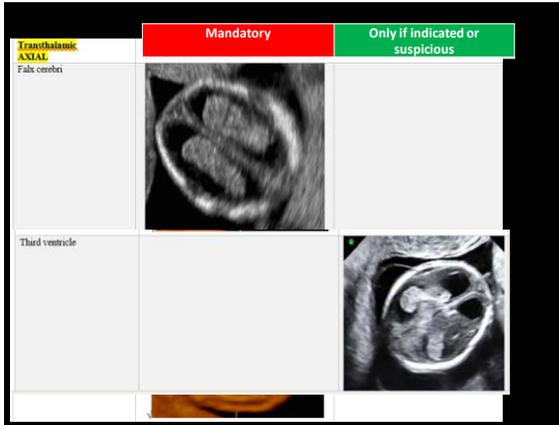
BIOMETRY	Mandatory	Only if indicated or suspicious
Crown-rump length		
Biparietal diameter		
Head circumference		
Abdominal circumference		

Report the mean of 3 acceptable measurements.

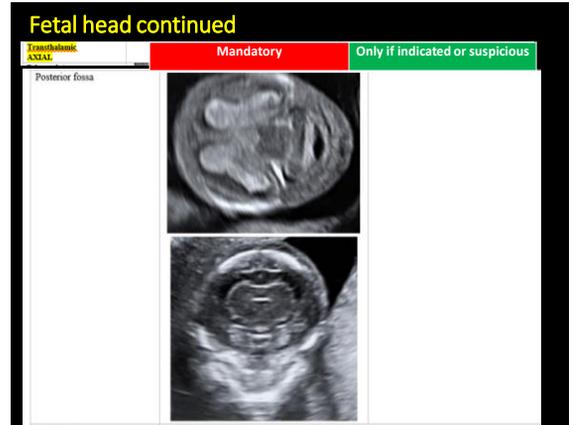
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FETAL HEAD	Mandatory	Only if indicated or suspicious
<b>A. AXIAL</b> Transventricular Cranial bones (calvarium)		
Falx cerebri		
Ventricles		

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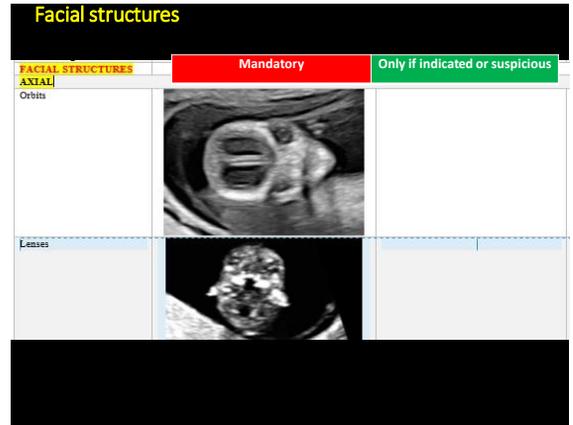
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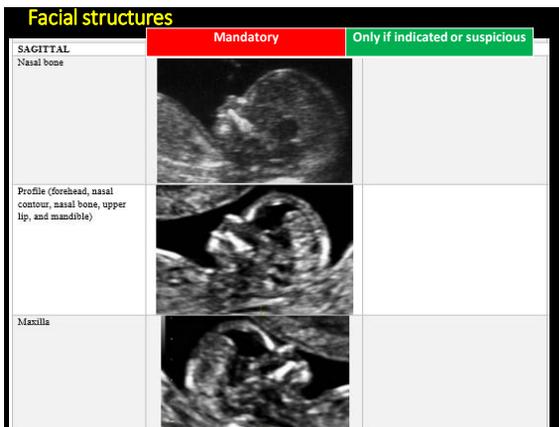
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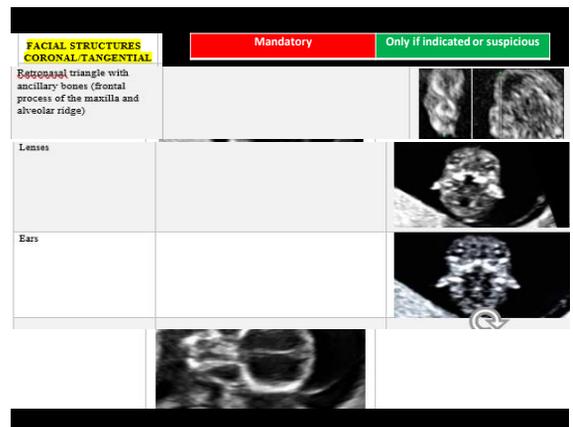
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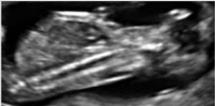
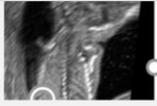
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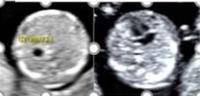
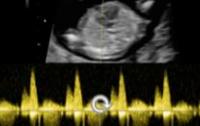
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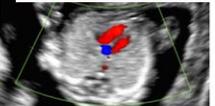
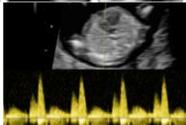
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		Mandatory	Only if indicated or suspicious
<b>NECK</b>			
<b>A. AXIAL, SAGITTAL, CORONAL</b>			
Evaluation for cystic hygroma, dilated jugular lymphatic sacs, other abnormal fluid collections, or masses			
<b>B. SAGITTAL</b>			
Neckal transmissivity measurement	Precise measurement of the NT if clinically requested as part of screening protocol or subjectively abnormal, criteria for measurement and credentialing via <a href="#">Perinatal Quality Foundation</a> or <a href="#">Fetal Medicine Foundation</a>		

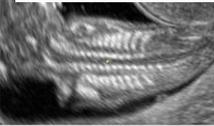
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		Mandatory	Only if indicated or suspicious
<b>FETAL THORAX</b>			
<b>A. AXIAL</b>			
Position of heart			
Heart rate			
Cardiac axis (angle measurement)			

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		Mandatory	Only if indicated or suspicious
<b>Fetal thorax continued</b>			
Four chamber view with color			
Symmetrical lungs (may be evaluated in coronal plane)			
Tricuspid valve flow			

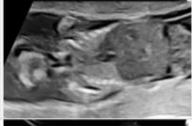
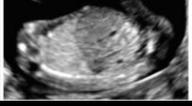
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		Mandatory	Only if indicated or suspicious
<b>Fetal thorax continued</b>			
Ribs with normal shape, length and ossification			
Diaphragm demarcation			
Comment:	While the use of Doppler should be limited in the first trimester, color flow imaging is useful to evaluate the fetal circulation; monitoring the output display standard (ODS) to keep $TIb \leq 0.7$		

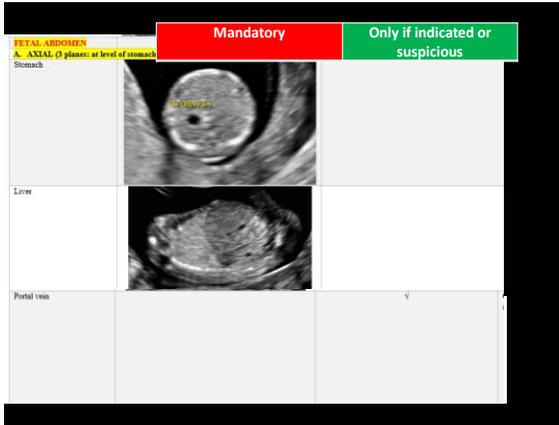
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		Mandatory	Only if indicated or suspicious
<b>Fetal thorax continued</b>			
<b>B. SAGITTAL</b>			
Aortic arch with color			
Ductal arch with color			

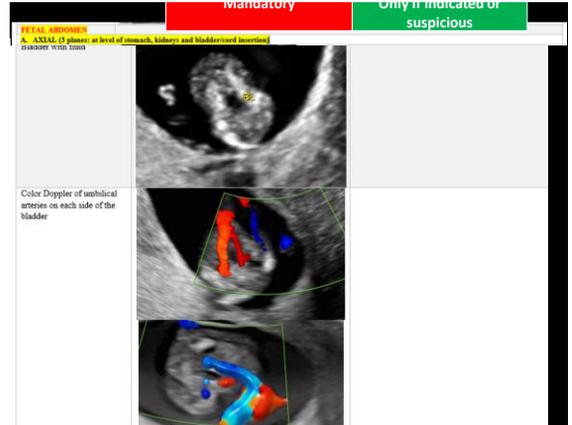
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		Mandatory	Only if indicated or suspicious
<b>Fetal thorax continued</b>			
<b>C. CORONAL</b>			
Lungs			
			
Diaphragm demarcation			

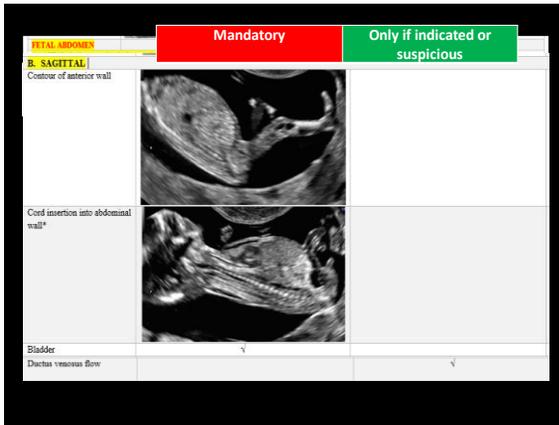
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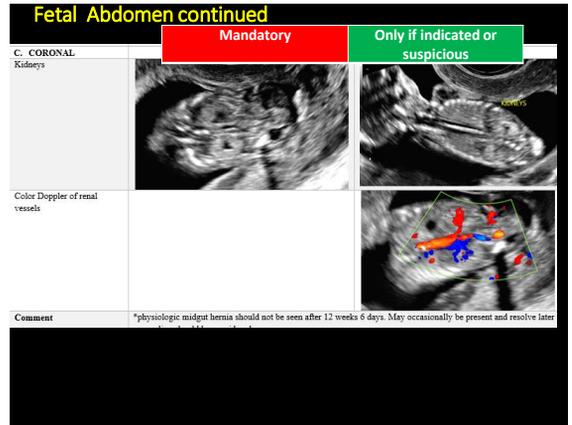
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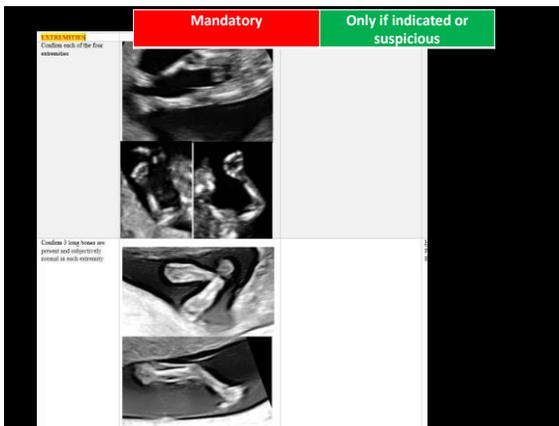
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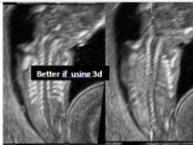
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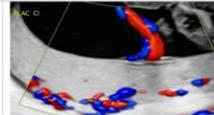
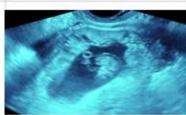
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	Mandatory	Only if indicated or suspicious
3-D assessment of extremities		

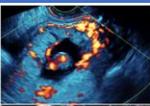
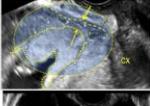
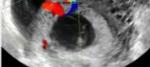
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	Mandatory	Only if indicated or suspicious
SPINE A. AXIAL, SAGITTAL, CORONAL Vertebral elements alignment		
Skin edge		
Scapula		

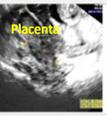
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	Mandatory	Only if indicated or suspicious
PLACENTA A. AXIAL AND SAGITTAL Location of gestational sac and position of placenta including relation to lower uterine segment prior hysterectomy (if applicable), and cervix.		
Umbilical cord insertion into placenta		
Echotexture of placenta		

45

	Mandatory	Only if indicated or suspicious
Placenta Color Doppler evaluation		
Myometrial thinning independent of retroplacental clear zone		
Bladder wall structure		
Comment:	In cases of suspected PAS, imaging should be done with a partially deflated bladder and trans-axial scanning should be performed. Color Doppler using a low-velocity scale, low filters and high gain to maximize detection of flow may be helpful.	
Uterine vessel vascularity		

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	Mandatory	Only if indicated or suspicious
UTERUS, ADNEXA and CUL-DE-SAC Myometrial masses (leiomyomas)	<input checked="" type="checkbox"/>	
Müllerian duct anomalies intrauterine linear structures	<input checked="" type="checkbox"/>	
Cul-de-sac Document right and left ovaries/adnexa		
CERVIX??		

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## The end of the list of required structures

- 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.

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List of Anomalies	
The list of anomalies, which are currently detectable or potentially detectable at 11-13 weeks.	
+ Head	+ Intraabdominal, renal and bladder
+ Brain	+ Abdominal wall defects
+ Spine	+ Extremities
+ Face	+ Skeletal dysplasias
+ Neck, skin, effusions and hydrops	+ Chromosomal syndromes
+ Chest	+ Syndromes and associations
+ Heart (congenital heart defects = CHD)	+ Placental and amniotic fluid
+ Vessels	

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### Head

- **Acrania**
- **Exencephaly**
- Encephalocele : Anterior – Occipital - Parietal
- Craniorachischisis
- Iniencephaly
- Abnormal shape of the head: Brachycephaly - Dolichocephaly
  - Trigenocephaly (strawberry shaped head)
  - Hypomineralised skull



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### Head

- Acrania
- Exencephaly
- **Encephalocele** : Anterior Occipital - Parietal
- Craniorachischisis
- Iniencephaly
- Abnormal shape of the head: Brachycephaly - Dolichocephaly
  - Trigenocephaly (strawberry shaped head)
  - Hypomineralised skull



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### 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



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### Brain

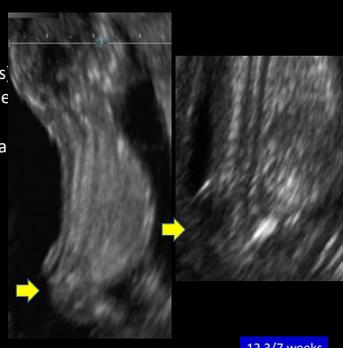
- Unclassified severe anomalies of the brain
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- Arachnoid cysts
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### Spine

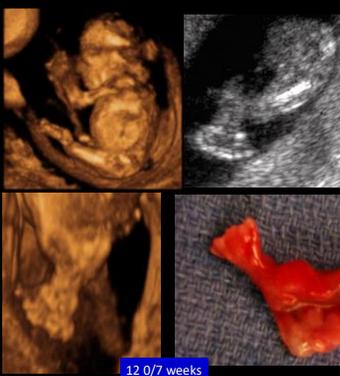
- **Spina bifida:**
  - Brain (cranial signs)
  - Meningomyelocele
  - Rachischisis
  - Closed spina bifida
- Fetal tail
- Sirenomelia
- Hemivertebra
- Diastematomyelia
- Caudal regression
- Kyphosis



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### Spine

- Spina bifida:
  - Brain (cranial signs)
  - Meningomyelocele
  - Rachischisis
  - Closed spina bifida
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- **Sirenomelia**
- Hemivertebra
- Diastematomyelia
- Caudal regression
- Kyphosis

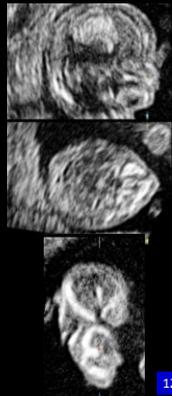


12 0/7 weeks

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### Face

- Midfacial hypoplasia (flat face)
- Anophthalmia
- Microphthalmia
- Cyclopia
- Arhinia
- Proboscis
- **Facial clefts:** Midline cleft - Bilateral cleft - Unilateral cleft
  - Atypical cleft – amniotic band
- Micrognathia

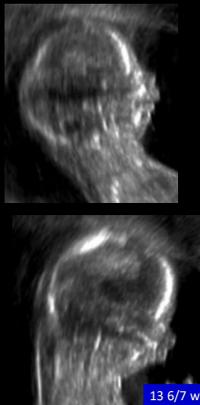


12 1/7 weeks

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### Face

- Midfacial hypoplasia (flat face)
- Anophthalmia
- Microphthalmia
- Cyclopia
- Arhinia
- Proboscis
- Facial clefts: Midline cleft - Bilateral cleft - Unilateral cleft
  - Atypical cleft – amniotic band
- **Micrognathia**



13 6/7 weeks

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### 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

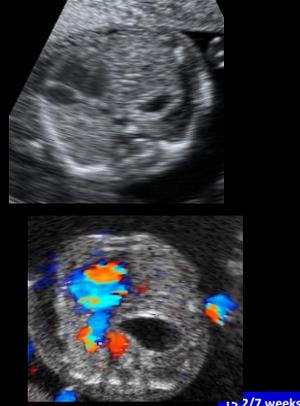


12 weeks

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### Chest

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



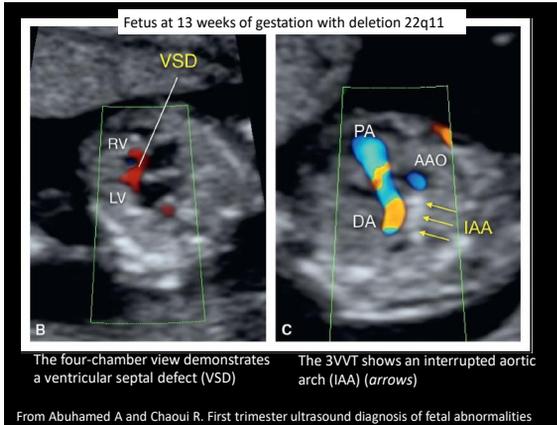
12 2/7 weeks

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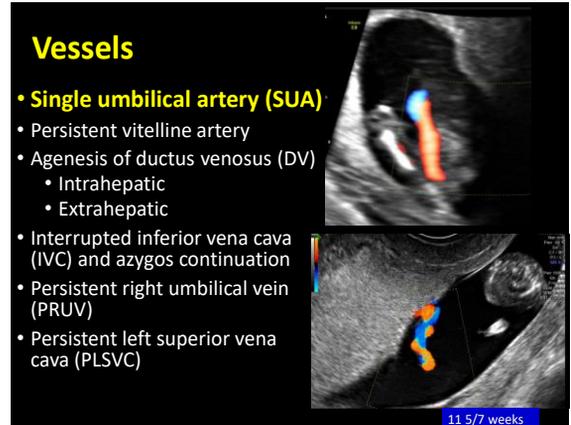
### Heart (congenital heart defects = CHD)

- 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)
- **Interrupted 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
- Ectopia cordis
- Dextrocardia
- Left atrial isomerism (LAI)
- Right atrial isomerism (RAI)
- Bradycardia
  - Congenital heart block
  - Dying fetus
- Tachycardia
- Cardiomegaly

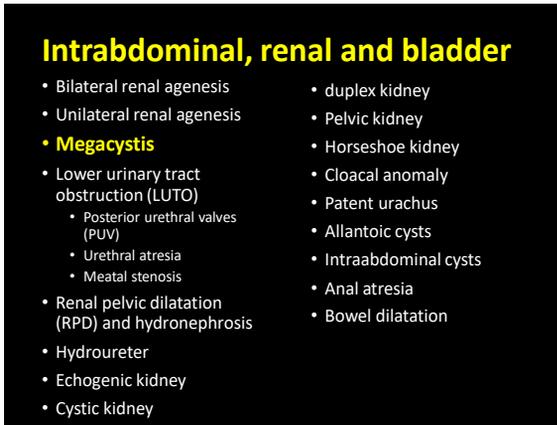
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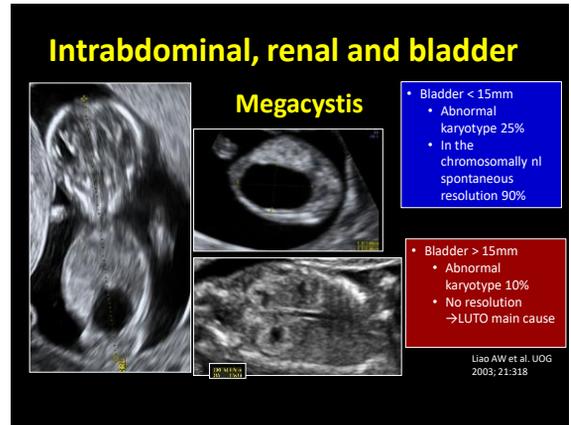
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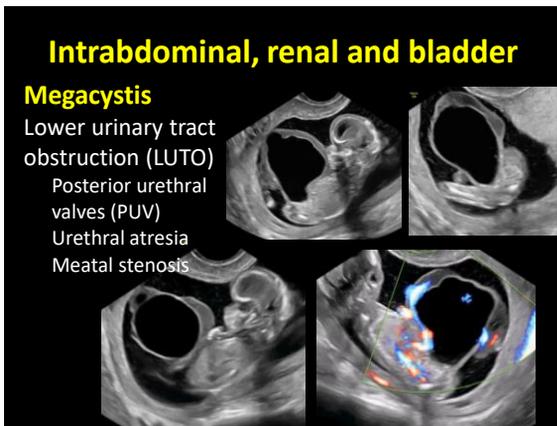
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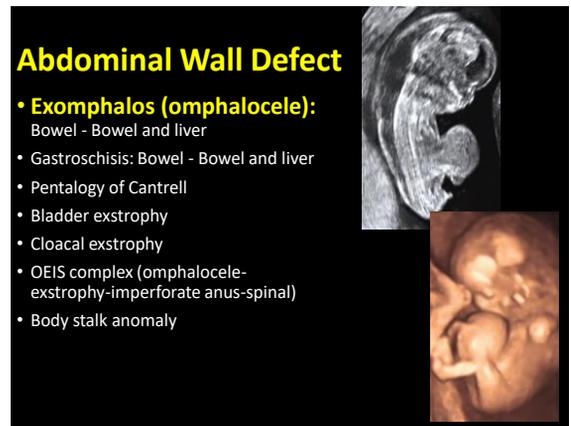
63



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### Abdominal Wall Defect

- Exomphalos (omphalocele): Bowel - Bowel and liver
- Gastroschisis: Bowel - Bowel and liver
- Pentalogy of Cantrell
- **Bladder exstrophy**
- Cloacal exstrophy
- OEIS complex (omphalocele-exstrophy-imperforate anus-spinal)
- Body stalk anomaly

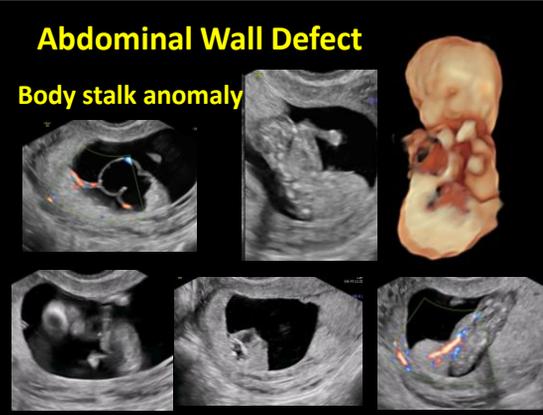


11 5/7 weeks

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### Abdominal Wall Defect

#### Body stalk anomaly



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### 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



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11 0/7 weeks

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### Skeletal Dysplasia

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



"hitchhiker" thumb  
12 weeks

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### Chromosomal syndromes

- Trisomy 21
- Trisomy 18
- Trisomy 13
- Turners syndrome
- **Triploidy maternal (digynic)**
- Triploidy paternal (diandric)
- DiGeorge syndrome (22q11 deletion)

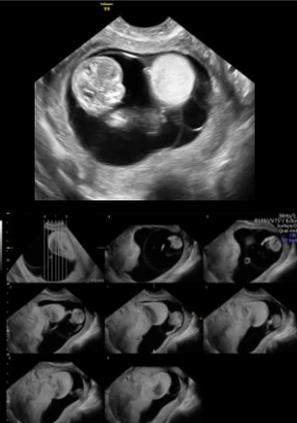


Early growth restriction and a large head; small placenta

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### 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



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12 <sup>3/7</sup> weeks

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### Multiple Pregnancy

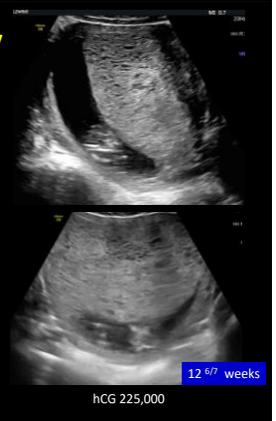
- Twins
  - Dichorionic
  - Monochorionic
  - Monoamniotic
  - Conjoined
- **Twin with abnormal co-twin**
- Growth discordancy
- Triplets
- High order pregnancies
- Twin reversed arterial perfusion (TRAP) sequence or acardiac twinning
- Molar pregnancy with normal co-twin



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hCG 225,000  
12 <sup>5/7</sup> weeks

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### First Trimester Screening Sensitivity

Evaluation of <i>major</i> anomalies in low-risk population	Sensitivity 46.10% [36.88-55.46]
Evaluation of <i>all</i> anomalies in low-risk population	Sensitivity 32.35 % [22.45-45.12]
Evaluation of <i>all</i> anomalies in high-risk population	Sensitivity 66.29% [43.47-85.69]

Karim et al 2017

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### .... In Conclusion

- The driven anatomy scan at 12-13 <sup>6/7</sup> 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



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**Thank You  
:)**

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