11-14 week ultrasound examination

Nimrah Abbasi MD
Maternal-Fetal Medicine, Mount Sinai Hospital

Sinai Health System

Ontario Fetal Centre

Obstetrics & Gynaecology
UNIVERSITY OF TORONTO

Objectives

- Role of T1 US in detection of anomalies
- Setting the standards for first trimester assessment: Review of ISUOG guidelines
- Review of always detectable vs. undetectable anomalies in T1 (cases)

Editorial

What will be the role of first-trimester ultrasound if cell-free DNA screening for aneuploidy becomes routine?

J. D. SONEK*† and H. S. CUCKLE‡

- **Complimentary**
  - **Accurate dating**
  - **Determine chorionicity**
  - **Evaluation of uterus and adnexa**
  - **Clarify cfDNA results**
    - Failure of cfDNA in 1-8%
    - Unexpected findings at 11- to 14-week scan in 3.5% of patients at risk based on age/medical history with neg cfDNA[1]
  - **Detect fetal structural abnormalities**
  - **Preeclampsia screening**

1Reiff E. et al. Prenat Diagn 2016, 36, 260–265)

Ultrasound Obstet Gynecol 2017; 50: 429–441
Published online 7 September 2017 in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/uog.17246

Systematic review of first-trimester ultrasound screening for detection of fetal structural anomalies and factors that affect screening performance

J. N. KARIM[1], N. W. ROBERTS[2], L. J. SALOMON[3] and A. T. PAPAGEORGHIOU[1,4]

Conclusions *Detection rates of first-trimester fetal anomalies ranged from 32% in low-risk groups to more than 60% in high-risk groups, demonstrating that first-trimester ultrasound has the potential to identify a large proportion of fetuses affected with structural anomalies. The use of a standardized anatomical protocol improves the sensitivity of first-trimester ultrasound screening for all anomalies and major anomalies in populations of varying risk. The development and introduction of international protocols with standard anatomical views should be undertaken in order to optimize first-trimester anomaly detection.*

Detection rate vs. GA

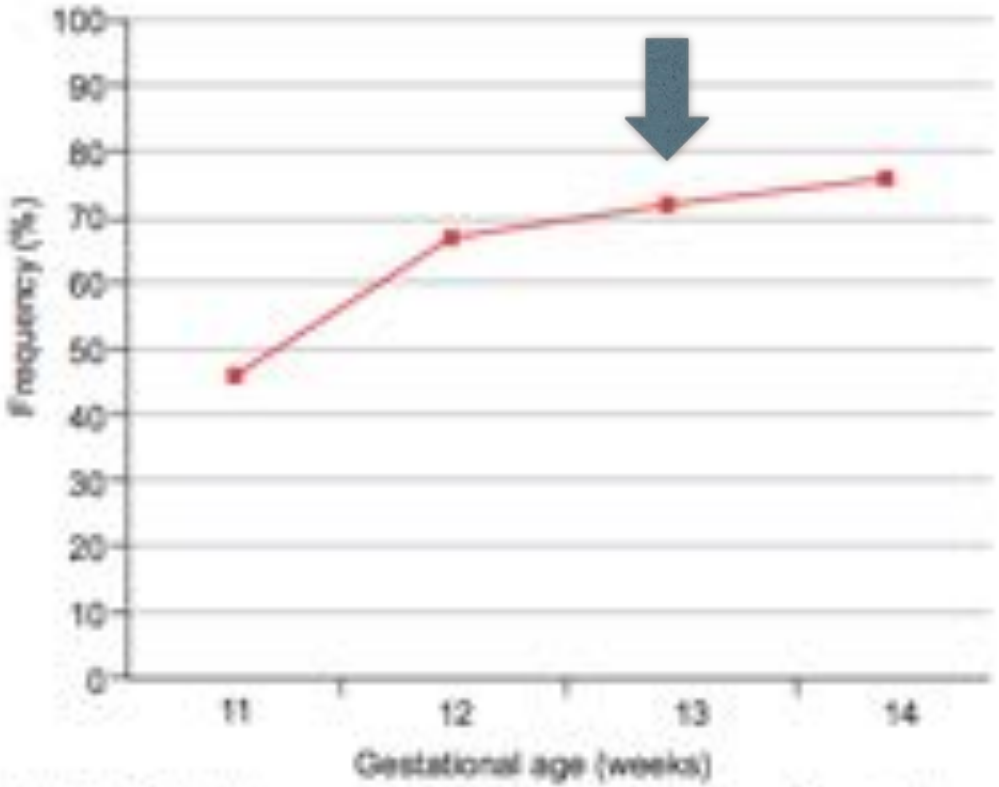


Fig. 3. Detection rates from 11–14 weeks of gestation.
Rossi. First-Trimester Ultrasonography. Obstet Gynecol 2013.

Ultrasound Obstet Gynecol 2013; **41**: 102–113
Published online in Wiley Online Library (wileyonlinelibrary.com). **DOI:** 10.1002/uog.12342

isuog.org

GUIDELINES

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

- **11- 13+6 weeks GA**
- **Viability**
- **Dating**
- **Number of fetuses**
- **Chorionicity & amnionicity**
- **Aneuploidy screening (NT)**
- **Fetal anatomy assessment**
  - 18-22 weeks = standard of care

Head	Present
	Cranial bones
	Midline falx
	Choroid-plexus-filled ventricles
Neck	Normal appearance
	Nuchal translucency thickness (if accepted after informed consent and trained/certified operator available)*
Face	Eyes with lens*
	Nasal bone*
	Normal profile/mandible*
	Intact lips*
Spine	Vertebrae (longitudinal and axial)*
	Intact overlying skin*
Chest	Symmetrical lung fields
	No effusions or masses
Heart	Cardiac regular activity
	Four symmetrical chambers*
Abdomen	Stomach present in left upper quadrant
	Bladder*
	Kidneys*
Abdominal wall	Normal cord insertion
	No umbilical defects
Extremities	Four limbs each with three segments
	Hands and feet with normal orientation*
Placenta	Site and texture
Cord	Three-vessel cord*

Dating

- 10-13+6 wks GA
- **CRL most precise parameter ≤ 84.0mm**
  - Predicts date of conception **+/- 5 days** (95% of cases)
- **Use HC (better than BPD) when CRL > 84 mm**

isuog.org

No. 303, February 2014

Determination of Gestational Age by Ultrasound

Summary Statements

1. When performed with quality and precision, ultrasound alone is more accurate than a "certain" menstrual date for determining gestational age in the first and second trimesters (≤ 23 weeks) in spontaneous conceptions, and it is the best method for estimating the delivery date. (II)

Recommendations

1. First-trimester crown-rump length is the best parameter for determining gestational age and should be used whenever appropriate. (I-A)
2. If there is more than one first-trimester scan with a mean sac diameter or crown-rump length measurement, the earliest ultrasound with a crown-rump length equivalent to at least 7 weeks (or 10 mm) should be used to determine the gestational age. (III-B)

Number of fetuses

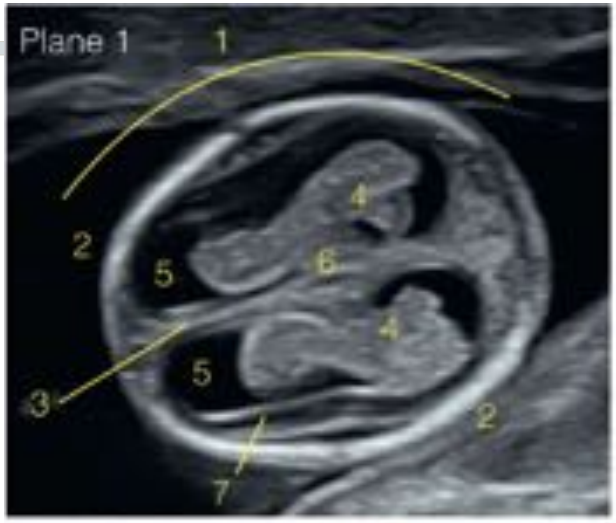
Chorionicity & Amnionicity

Empty lambda = MC

- **11- 13 + 6 weeks GA**
- Membrane thickness at site of insertion of amniotic membrane into placenta **(T or lambda sign)**
- **Number of placental masses**
- **Store images!!**

isuog.org

Head & Neck



First Trimester Ultrasound Diagnosis of Fetal Abnormalities. Abuhamad-Chaoui. September 2017

- Cranial bone ossification
- No skull defect
- Lateral ventricles large and filled with echogenic choroid plexuses in posterior two thirds
- Symmetrical
- Interhemispheric fissure and falx
- Brain mantle is thin (anterior)
- Evaluate neck for NT, jugular sacs

isuog.org

Spine

- Progressive ossification of spine between 11-13 weeks
- Normal vertebral alignment and integrity
- Intact overlying skin

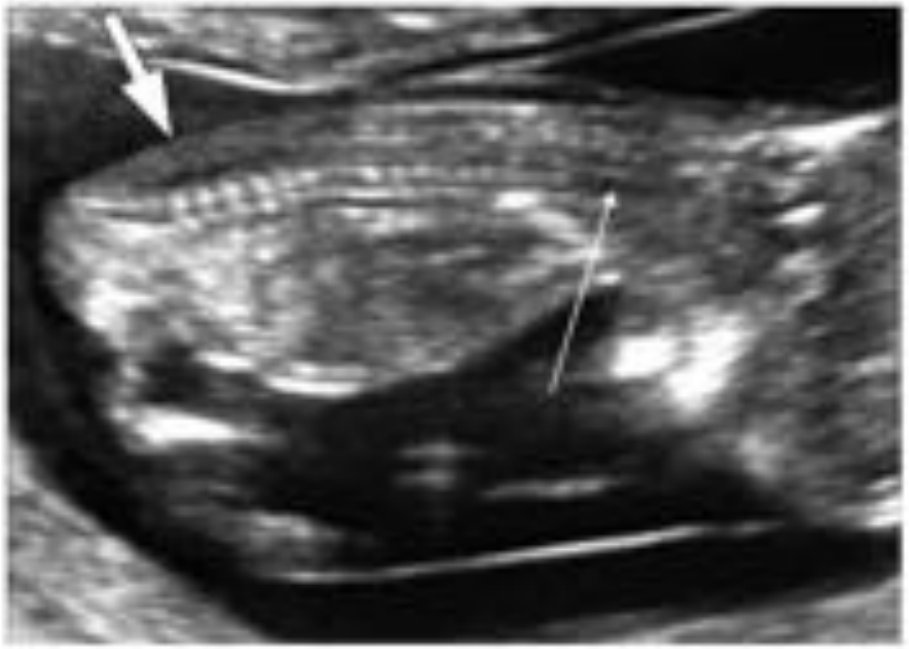


Figure 4 Fetal spine. Intact skin (short thick arrow) is visible posterior to the vertebrae from neck to sacrum in a true median view. Note vertebral bodies show ossification, but neural arches, which are still cartilaginous, are isoechoic or hypoechoic. In cervical region (long arrow) the vertebral bodies have not yet ossified and the cartilaginous anlage is hypoechoic; this is normal.

From nuchal translucency to intracranial translucency: towards the early detection of spina bifida

Ultrasound Obstet Gynecol 2010; 35: 133–138

R. CHAOUI†*and K. H. NICOLAIDES‡

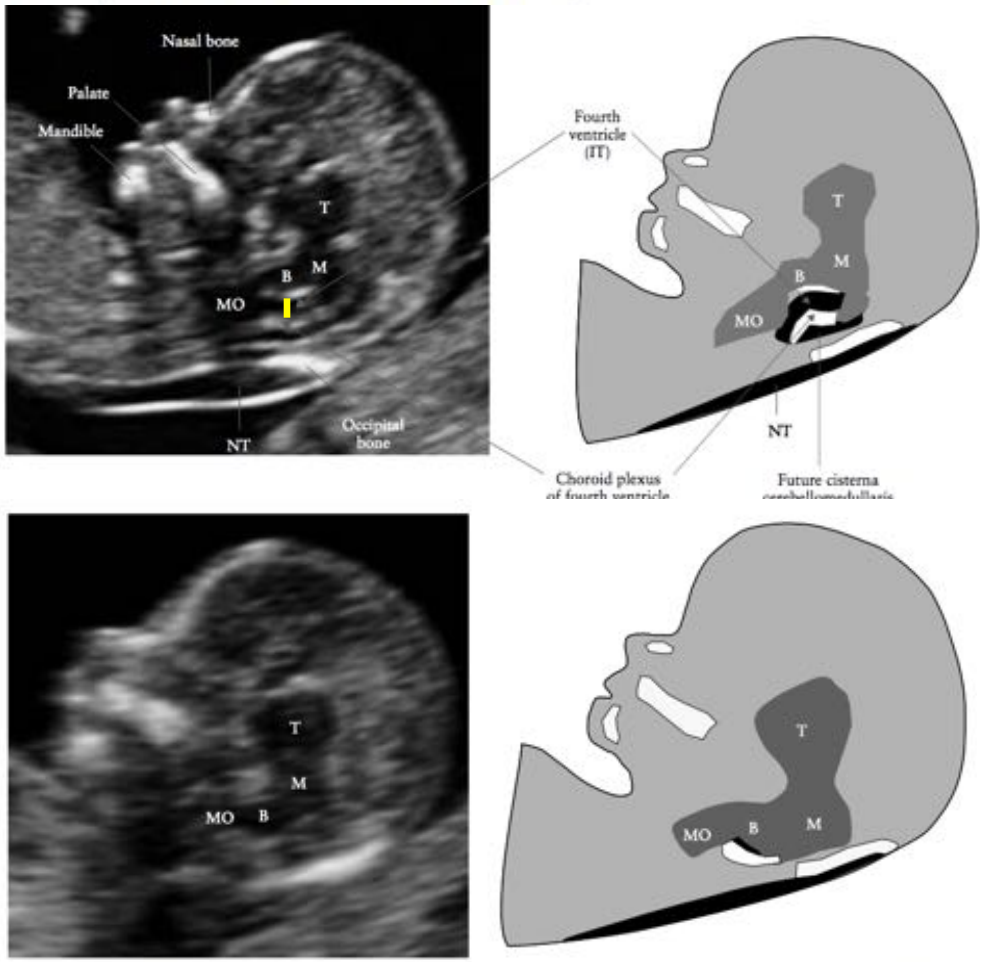
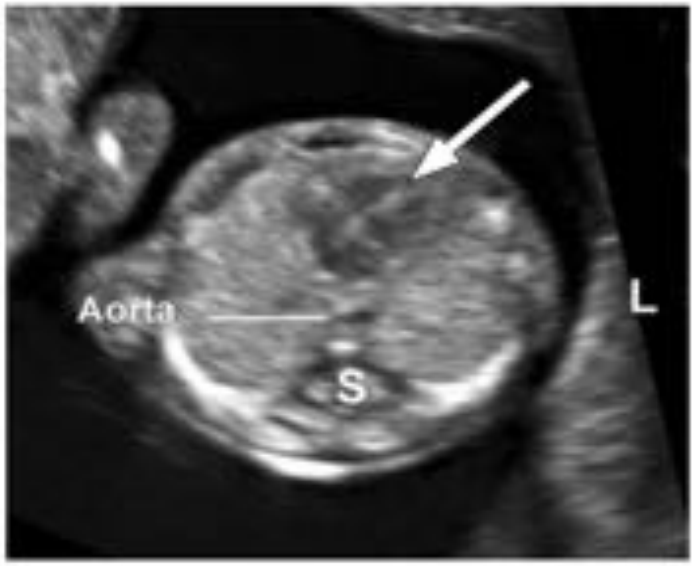


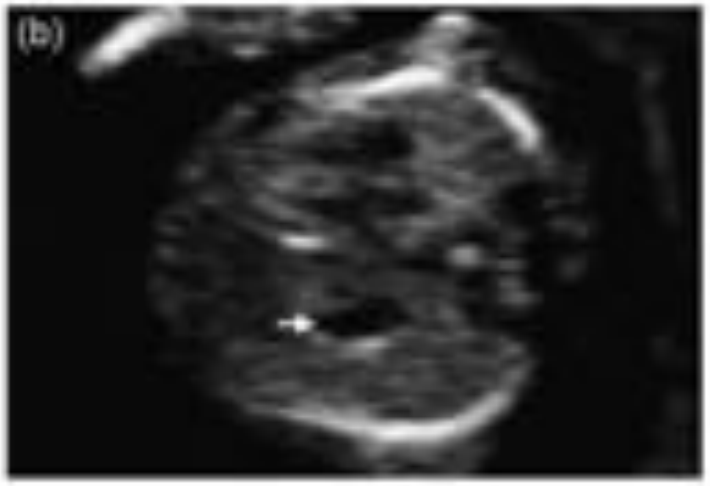
Figure 3 Ultrasound image in the mid-sagittal plane of the fetal face in a case of open spina bifida demonstrating compression of the fourth ventricle with no visible translucency. B, brain stem; M, midbrain; MO, medulla oblongata; T, thalamus.

- In T2, manifestations of Arnold–Chiari malformation are the lemon and banana signs
- In T1, caudal displacement of the brain results in compression of the fourth ventricle and loss of the normal IT

Chest

- Lungs homogeneous echogenicity
- No pleural effusions
- No cystic or solid masses
- Levocardia (axis 45° +/- 15°)
- Symmetrical atria and ventricles
- Aorta left of spine
- Diaphragmatic continuity
- Stomach and liver intrabdominal





Prenat Diagn 2011; **31**: 90–102.

isuog.org

Fetal Cardiac Axis and Congenital Heart Defects in Early Gestation

Elena S. Sinkovskaya, MD, PhD, Rabih Chaoui, MD, Katrin Karl, MD, Elena Andreeva, MD, PhD, Ludmila Zhuchenko, MD, PhD, and Alfred Z. Abuhamad

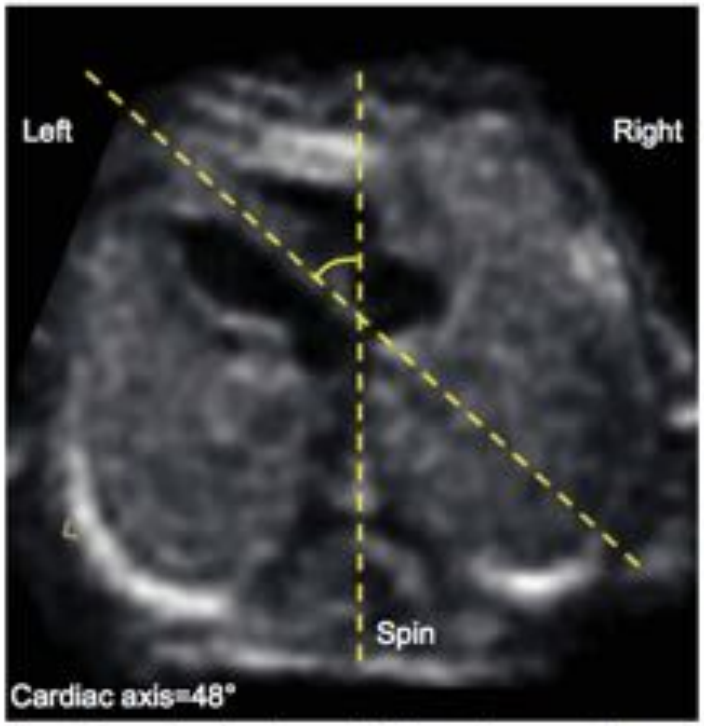
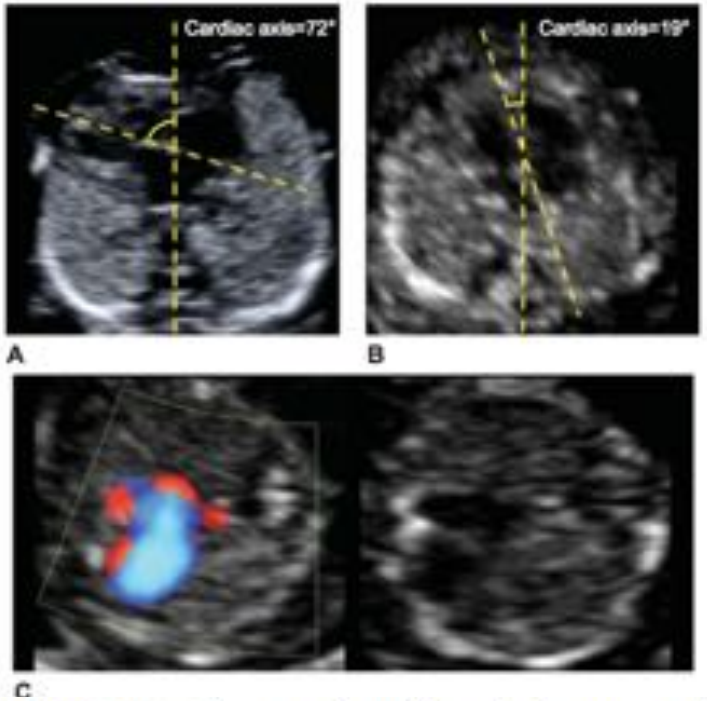


Fig. 1. Four-chamber view of the heart of a normal fetus at 12 6/7 weeks of gestation. Measurement of the cardiac axis is demonstrated.

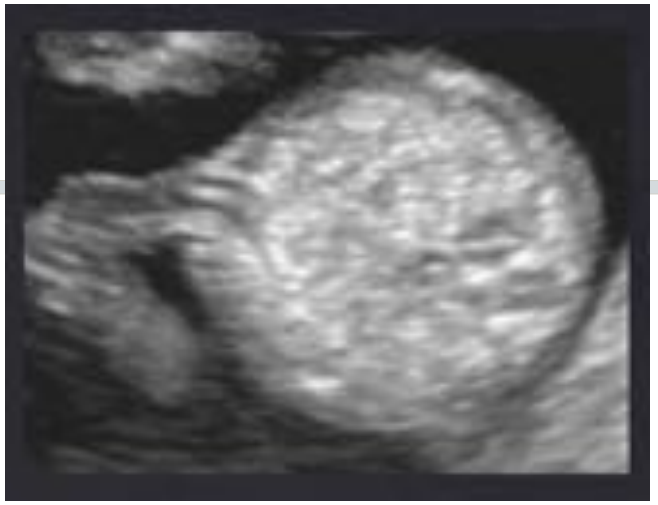


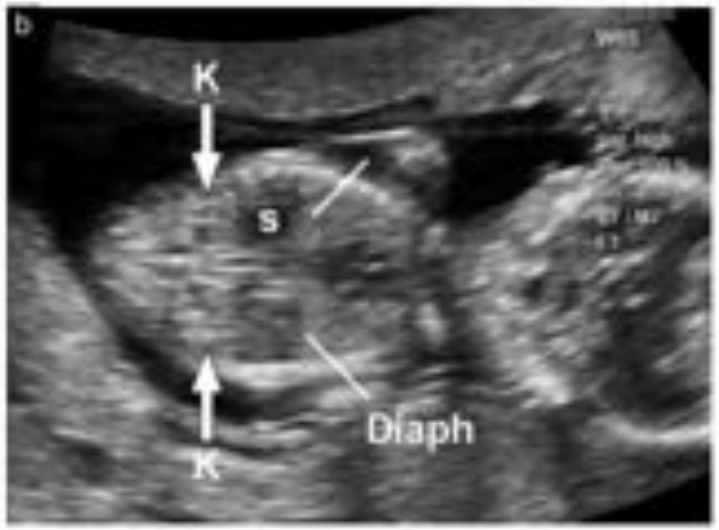
CONCLUSION: Abnormal cardiac axis is present in two-thirds of fetuses with congenital heart defect in early gestation. Adding cardiac axis assessment to the nuchal translucency measurement is helpful in defining a population at risk for fetal congenital heart defect.

Normal cardiac axis defined as ≥ 30° but < 60°.

Abdomen/GU

- Stomach on left side + levocardia (situs visceralis)
- Umbilical cord vessels, cord insertion (>12 wks/ CRL >45mm), cysts
- Kidneys slightly echogenic
- Bladder > 12 wks (<7mm)

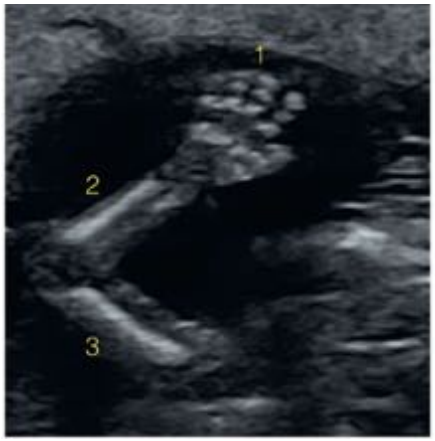


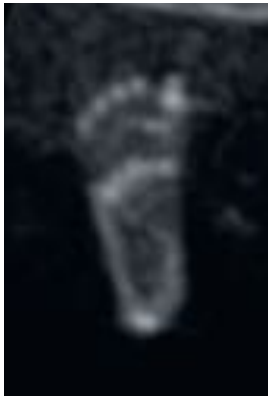


CRL 34.0mm physiological herniation (bowel)

Limbs

- Presence of each bony segment of the upper and lower limbs
- Presence and normal orientation of two hands and feet



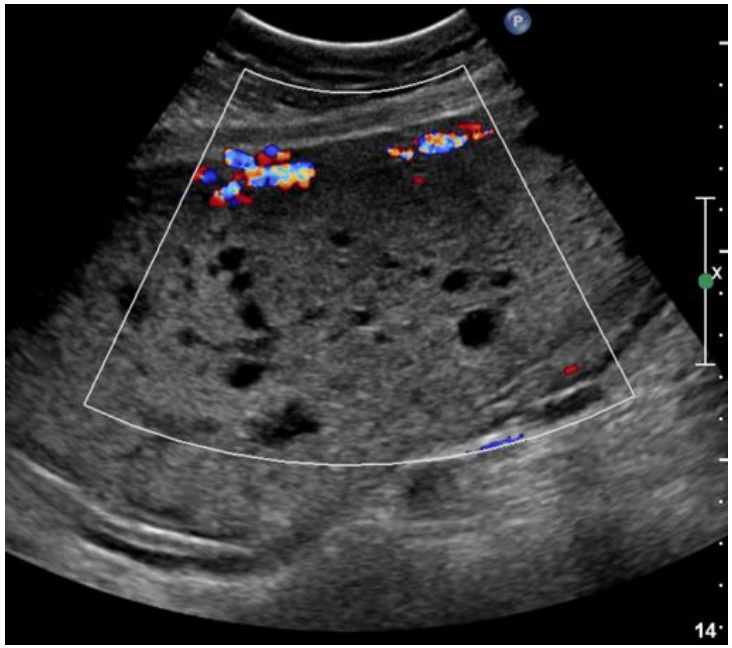


First Trimester Ultrasound Diagnosis of Fetal Abnormalities. Abuhamad-Chaoui. September 2017

isuog.org

Placenta, uterus & adnexa

- Placenta
  - Echostructure, masses, cystic spaces, subchorionic fluid collection (> 5 cm)
  - If prior C/S:
    - Area between bladder and uterine isthmus at site of CS scar (scar pregnancy, AIP)
- Abnormalities of uterus (septa, bicornuate, fibroids) and adnexa (masses, cysts)



Use a checklist ✔

APPENDIX: ROUTINE ULTRASOUND WORKSHEET (EXAMPLE)

isuog

Patient: ID number:

Date of birth (DD/MM/YYYY):
Referring physician:

Date of exam (DD/MM/YYYY):
Sonographer / Supervisor:

Indication for scan and relevant clinical information:

LMP

Technical conditions: *Good / Limited by:*
Singleton / Multiple (use 1 sheet/fetus)
=> Chorionicity:

ADNEXA:
Appearance ☐ Normal ☐ Abnormal*
Anomaly:

MEASUREMENTS	mm	Centile (Reference range)
Crown-rump length		
Nuchal translucency (optional)		
Biparietal diameter		
Head circumference		
Abdominal circumference		
Femoral diaphysis length		

*Abnormal findings (please detail):

Ultrasound-based estimate of GA:

.....weeks + ...days

CONCLUSION:
☐ Normal and complete examination.
☐ Normal but incomplete examination.
☐ Abnormal examination*
☐ Plans: ☐ No further ultrasound scans required
☐ Follow-up planned in ... weeks

SONOGRAPHIC APPEARANCE OF FETAL ANATOMY: (N = Normal; Ab = Abnormal*; NV = Not visualized; Gray = optional)	N	Ab*	NV
Head			
Shape			
Cranial ossification			
Midline falx			
Choroid plexus			
Face			
Orbits			
Profile			
Neck			
Thorax			
Pulm. area			
Diaphragm			
Heart			
Heart activity			
Size			
Cardiac axis			
Four-chamber view			
Abdomen			
Stomach			
Bowel			
Kidneys			
Urinary bladder			
Cord insertion / abdominal wall			
Cord vessels			
Spine			
Limbs			
Right arm (incl. hand)			
Right leg (incl. foot)			
Left arm (incl. hand)			
Left leg (incl. foot)			
Gender (optional): ♂ ♀			
Other:			

isuog.org

Mount Sinai Hospital

Early Anatomy / Echo Worksheet

Fetal Centre – Special Pregnancy Program

(Place patient label)

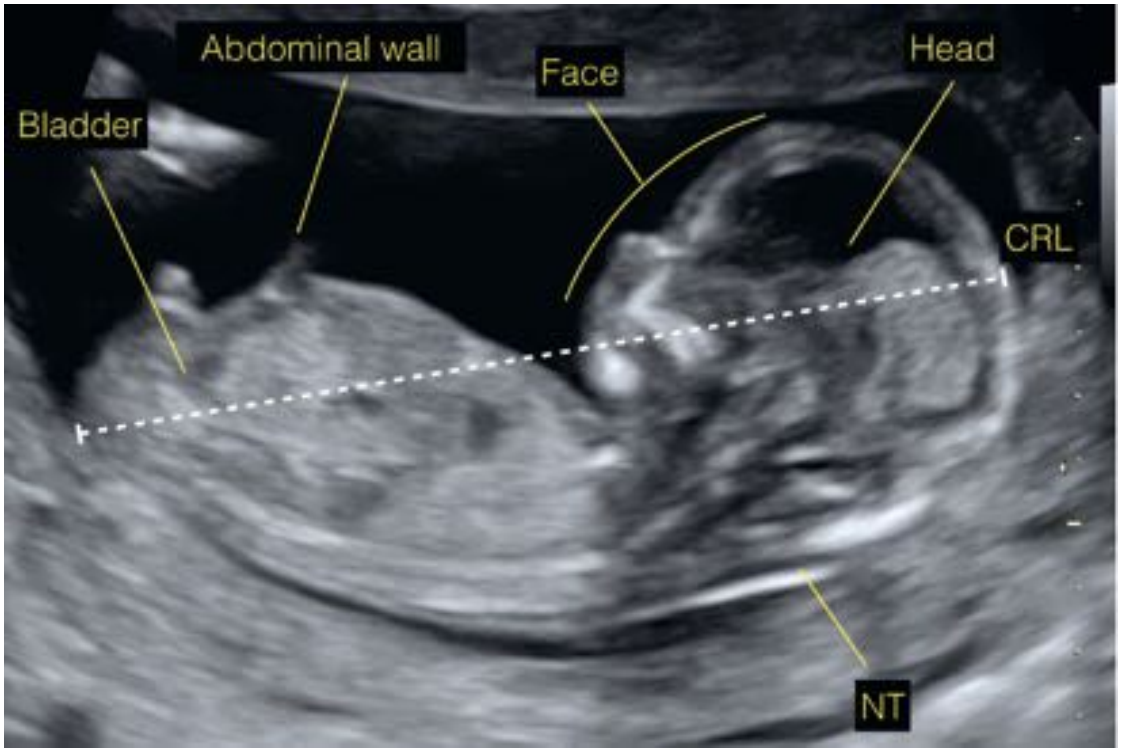
Indication

Measurements	date	Centile (Reference range)
Crown-rump length		
Nuchal translucency (optional)		
Biparietal Diameter		
Head circumference		
Abdominal circumference		
Femoral diaphysis length		

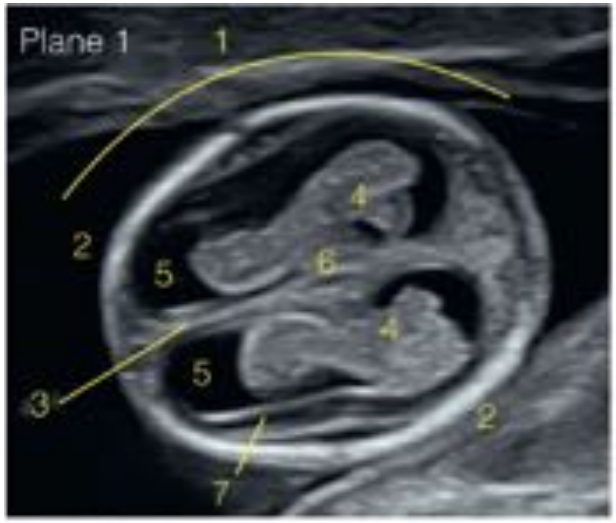
abnormal findings (please detail):

Sonographic Appearance of Fetal Anatomy: (N=Normal; AB*=Abnormal; NV=Not visualized)	N	AB*	NV
Head			
Shape			
Cranial ossification			
Midline falx			
Choroid plexus			
3rd			
Face			
Orbits			
Profile			
Nasal bone			
Retronasal			
Neck			
Thorax			
Diaphragm			
Heart			
Heart Activity			
Fill			
Cardiac axis & situs			
Four chamber view			
LVOT			
RVOT			
AVV			
TV			
Abdomen			
Stomach			
Bowel			
Kidneys			
Urinary bladder			
Cord insertion/abdominal wall			
Cord vessels			
Spine			

Always detectable abnormalities



- CRL, NT, CI, bladder
- Extremities (3 segments)



- Choroids, ventricles, midline/falx, cranial ossification
- Cord insertion
- Extremities (3 segments)

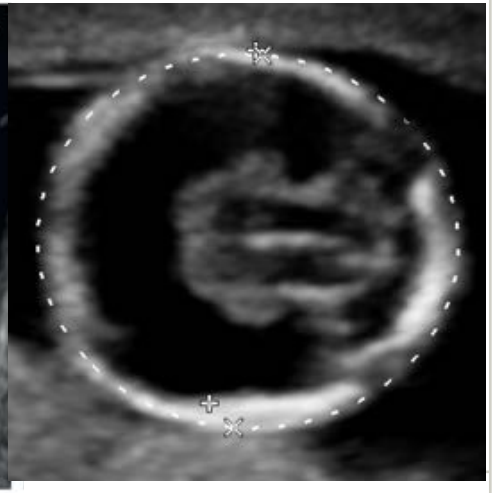
First Trimester Ultrasound Diagnosis of Fetal Abnormalities. Abuhamad-Chaoui. September 2017

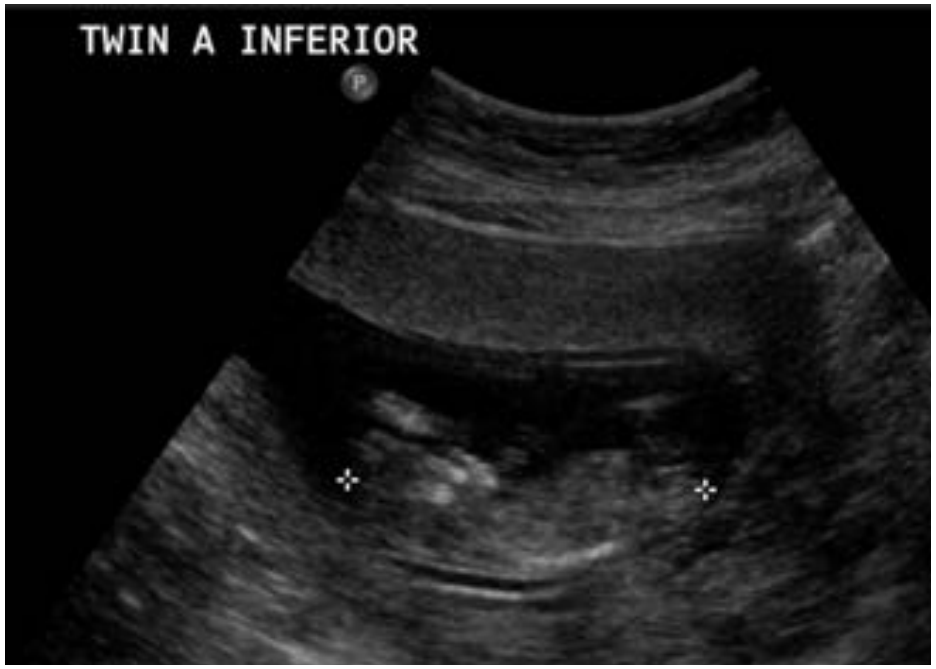

R 60Hz
2.0
M5
X
HEAD
JPEG
*** bpm

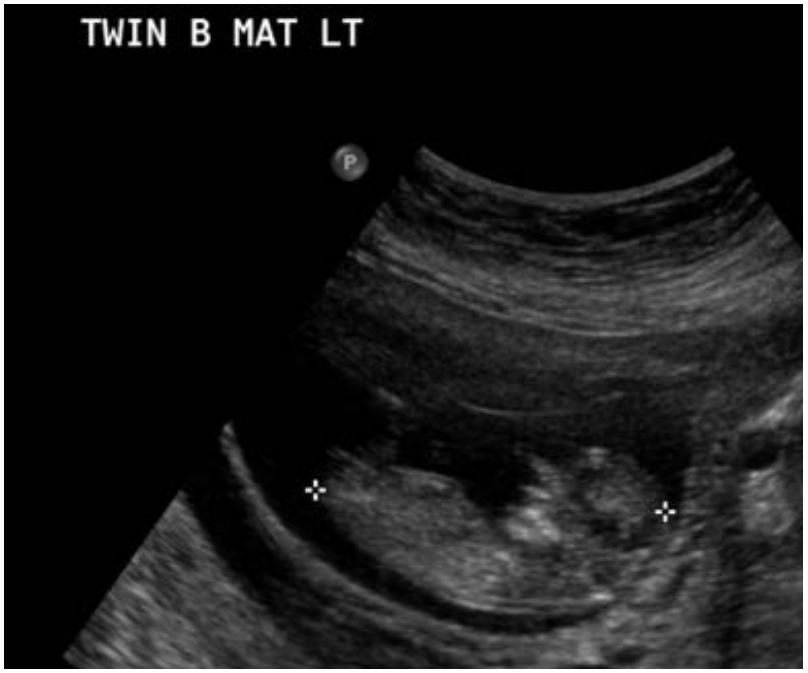
Alobar holoprosencephaly (HPE)

- Incomplete cleavage of forebrain with varying degree of fusion of cerebral hemispheres
- Alobar HPE
  - **Crescent shaped single ventricle**
  - **Fused thalami**
  - **No falx cerebri** (no “butterfly”)
  - Cyclopia, proboscis

- ~80% abnormal karyotype
  - (T13 (62%) , T18 (17%) , triploidy (17%), AD nonsyndromic HPE (Syngelaki et al. UOG 2016)



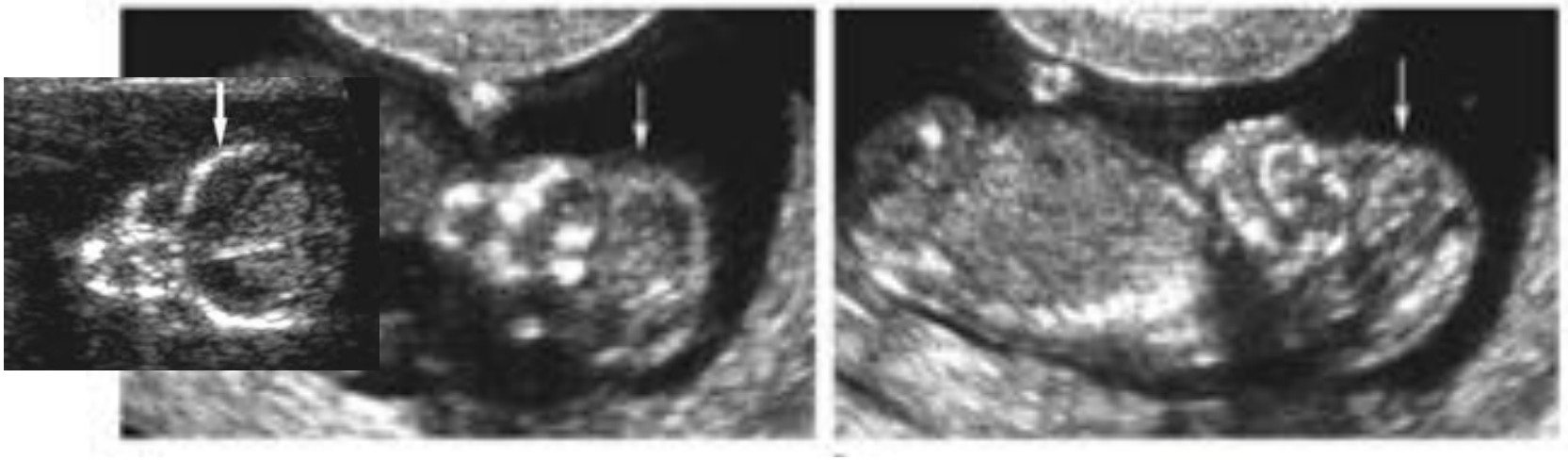

TWIN A INFERIOR


TWIN B MAT LT


FR 23Hz
RS
TWIN A
M3
2D
58%
C 60
P Off
HGen
JPEG
6.0
*** bpm

Acrania, Exencephaly/Anencephaly

- Failure of closure of rostral portion of neural tube
- **Acrania:** Absence of cranial vault above orbits
- **Exencephaly**: Acrania + abnormal mass/bulge from skull base & membrane
- **Anencephaly**: Acrania, no cerebral hemispheres & mid brain (18-22 wks)
- **Echogenic AF, amniotic bands**
- **At 11 – 13 weeks, the pathognomonic feature is acrania with brain appearing either normal or at varying degrees of distortion and disruption**



Fong KW et al. RadioGraphics 2004; 24:157–174

“Mickey Mouse” sign
Amorphous brain tissue bulging from base of skull (exencephaly)

“Frog eyes” sign
Little/ no brain tissue above orbits on coronal view of face (anencephaly)

Importance of image audits

Ultrasound Obstet. Gynecol. 9 (1997) 14–16

Ultrasound screening for anencephaly at 10–14 weeks of gestation

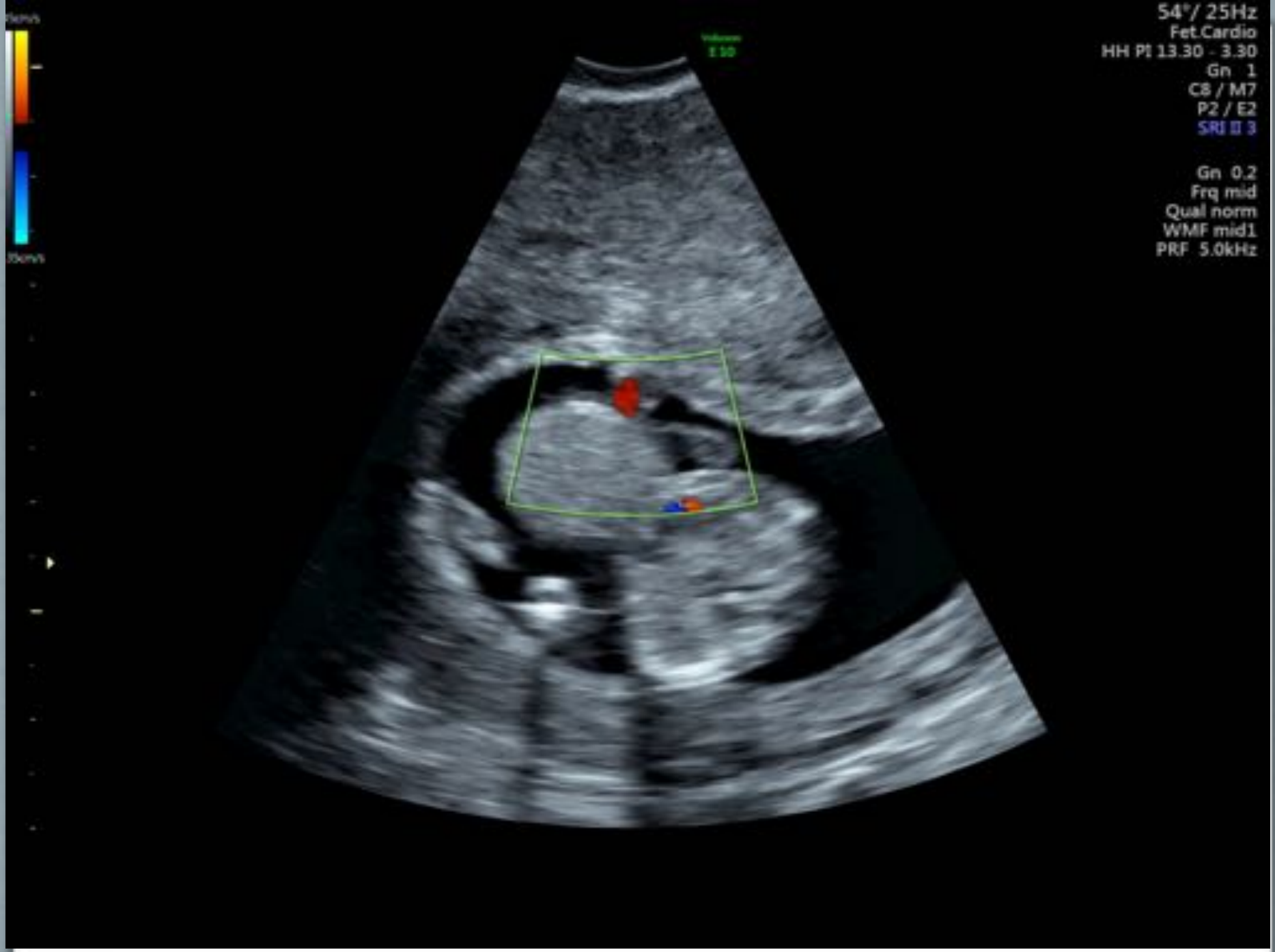
S. P. Johnson, N. J. Sebire, R. J. M. Snijders, S. Tunkel and K. H. Nicolaides

Lethal abnormality with no cranial vault and absent cerebral hemispheres

	Pre-audit n=31	**Post-audit* n=16**
Detection 10-14 weeks GA	23 (74%)	16 (100%)
Detection at 18-22 weeks GA	8 (26%)	0

*p= 0.03

- Determine detection rate
- Image review→ different appearance of anencephaly in T1 (acrania only) vs. T2
- Sonographer retraining
- Re-audit & demonstrate improvement


54°/ 25Hz
Fet.Cardio
HH PI 13.30 - 3.30
Gn 1
C8 / M7
P2 / E2
SRI II 3
Gn 0.2
Frq mid
Qual norm
WMF mid1
PRF 5.0kHz

Abdominal wall defects

Table 12.1 • First Trimester Ultrasound and Ventral Wall Defects

Physiologic midgut herniation	Herniation of small bowel in a small midline sac, measuring less than 7 mm and physiologically seen until the 12th week of gestation
Omphalocele	Midline defect with viscera covered by a membrane. Umbilical cord arises from the dome of the sac. Content can be small with bowel, but can also be large including bowel, liver, stomach, and other organs
Gastroschisis	Paraumbilical defect typically to the right of the umbilical cord insertion with evisceration of bowel. No covering membrane
Pentalogy of Cantrell	Five features: Abdominal defect similar to omphalocele but higher on abdomen (1), anterior defect of diaphragm (2), distal sternal defect (3), pericardial defect (4), cardiac abnormalities with partial or complete ectopia cordis (5)
Ectopia cordis	Sternal defect with the heart partly or completely exteriorized, with or without cardiac abnormalities
Body stalk anomaly (limb-body wall complex)	Complex large anterior wall defect with the fetus fixed to the placenta because of a short or absent umbilical cord. Deformities of body, spine, and limbs. Body stalk anomaly can also result from an amniotic band syndrome with a normal umbilical cord. See also in OEIS
Bladder exstrophy	Defect in the abdominal wall below the attachment of the umbilical cord. The insertion of the cord is low and below it bladder tissue is exteriorized. Urinary bladder is not visible. Female and male genitalia malformed. Can be part of cloacal exstrophy
Cloacal exstrophy, (OEIS complex)	In addition to bladder exstrophy, a low omphalocele is present in association with rectal and anorectal malformations and distal spine anomaly. Anomaly of genitalia is part of complex. OEIS complex is rare and stands for omphalocele, exstrophy of bladder, imperforate anus, and spinal defect. A body stalk anomaly of the lower body can present as OEIS complex, usually legs are completely or partly absent.

Physiologic herniation at 6-11 weeks GA with CRL < 45mm

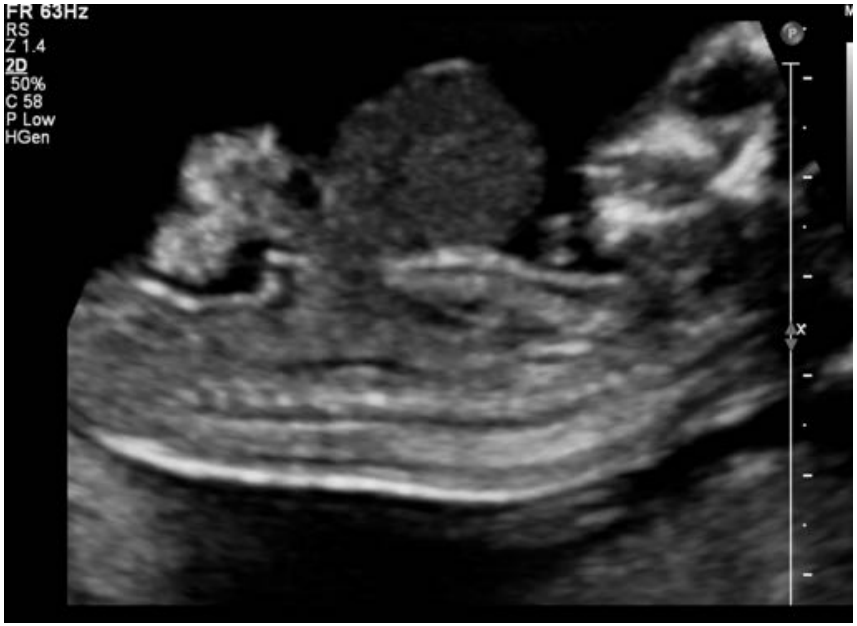
Unlikely to resolve if contains liver

Cannot reliably diagnose <12 weeks GA

First Trimester Ultrasound Diagnosis of Fetal Abnormalities. Abuhamad-Chaoui. September 2017

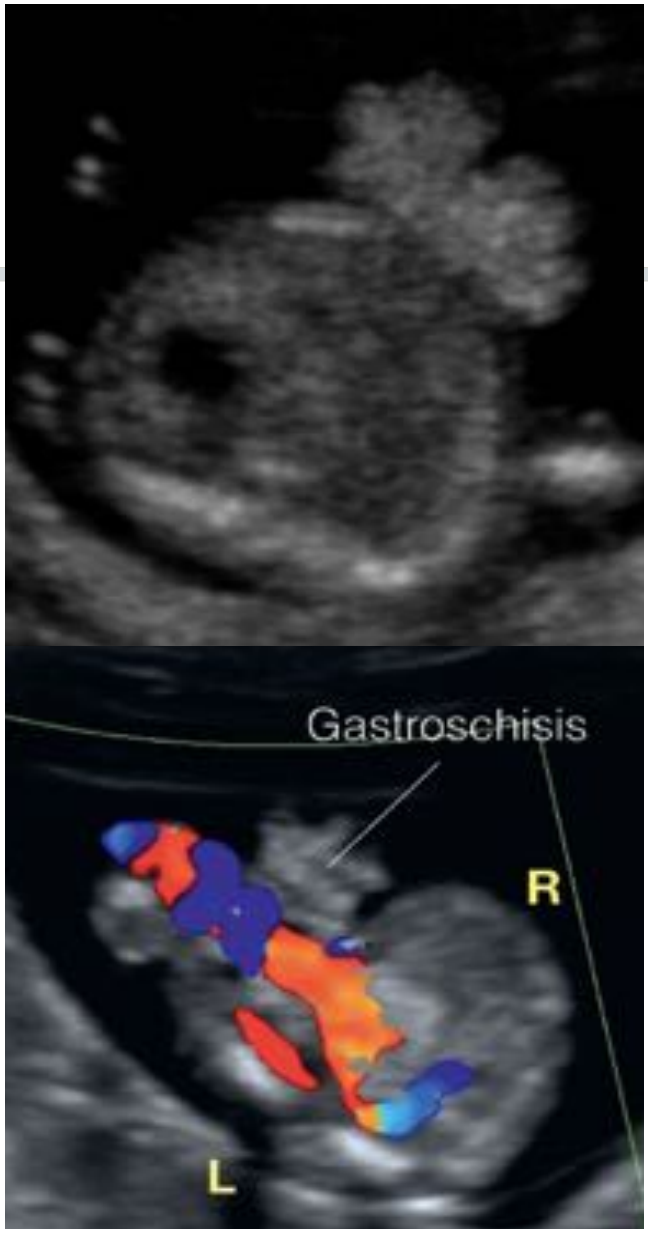
Omphalocele

- **Midline, covered by membrane**
- Bowel +/- liver, stomach, GB
- Umbilical cord arises from dome of herniated sac (color Doppler)
- Size inversely correlates with chromosomal abnormalities
  - 20-40% (T18)
  - BWS
  - Associated anomalies (cardiac)



Gastroschisis

- Para-umbilical defect, **right side of UC insertion** (color Doppler)
- **Herniated free floating bowel loops** without membrane
- Rarely associated with chromosomal or structural abnormalities



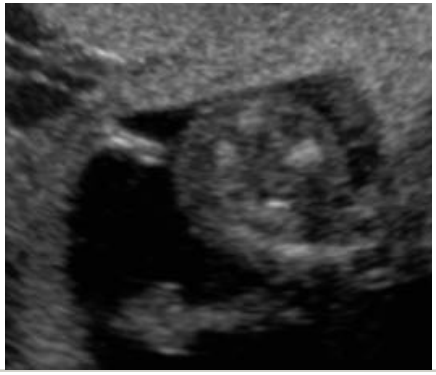
First Trimester Ultrasound Diagnosis of Fetal Abnormalities. Abuhamad-Chaoui. September 2017


FR 24Hz
RS
2D
52%
C 60
P Med
Res
M3
JPEG
7.1
*** bpm

Limb body wall/ Body stalk anomaly

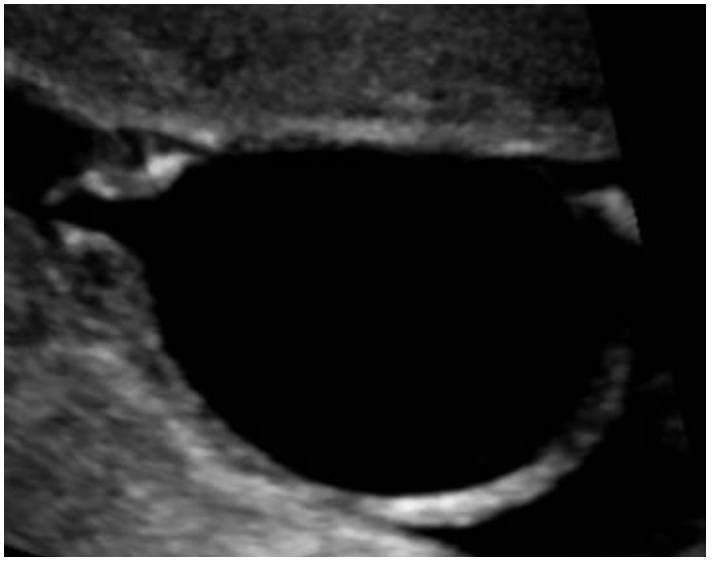
- Defective development of germinal disc (vascular insult) → amnion rupture with amniotic band-type defects
- Multiple malformations
  - **Thoracoabdominal wall defect**
  - **Craniofacial**
  - **Spine & extremities** (kyphoscoliosis→ no mid sagittal plane)
  - **Short/absent umbilical cord** (fetus "stuck" to placenta)
  - **+/- amniotic bands**

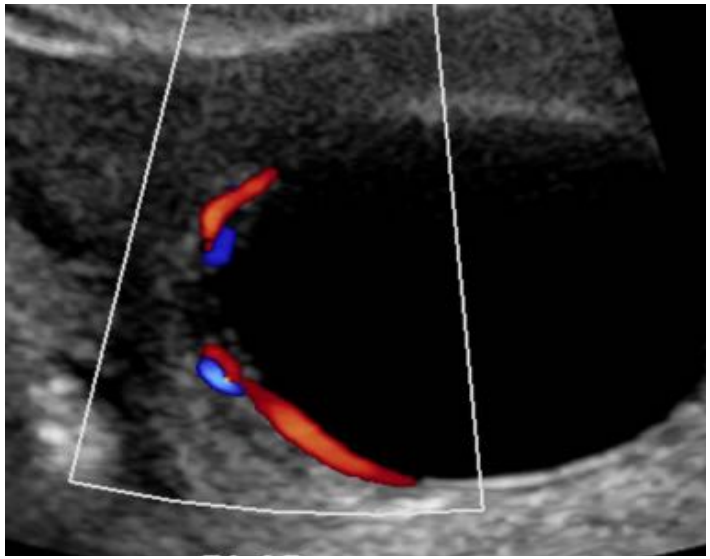
First Trimester Ultrasound Diagnosis of Fetal Abnormalities. Abuhamad-Chaoui. September 2017



Ectopia cordis

- Isolated or associated with pentalogy of Cantrell body stalk anomaly, amniotic band syndrome
- Omphalocele (large, high with liver) and ectopia cordis (midsagittal in T1)
- **Pentalogy of Cantrell**
  - Midline supra-umbilical AWD (vs. omphalocele at UC insertion)
  - Defect of lower sternum
  - Defect in diaphragmatic pericardium
  - Deficiency of anterior diaphragm
  - Intracardiac abnormalitie s(partial/ complete ectopia cordis)





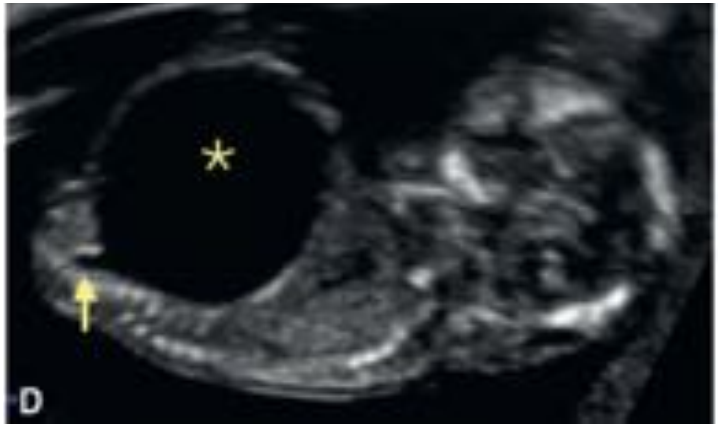
Megacystis at 10–14 weeks of gestation: chromosomal defects and outcome according to bladder length

A. W. LIAO, N. J. SEBIRE, L. GEERTS, S. CICERO and K. H. NICOLAIDES

- Bladder length ≥7mm
- Thickened/echogenic bladder wall
- Keyhole sign (dilated proximal urethra)
- Normal AF < 16 weeks!!

- **Bladder diameter 7-15mm**
  - Chromosomal abn. ~25% (T13/18)
  - If normal, 90% resolve
- **Bladder diameter >15mm**
  - Chromosomal abn. ~ 10% (T13/18)
  - **If normal, nearly all associated with progressive obstructive uropathy**

- Transient, LUTO (PUV, urethral atresia, megacystis-microcolon intestinal hypoperstalsis, cloacal malformations)

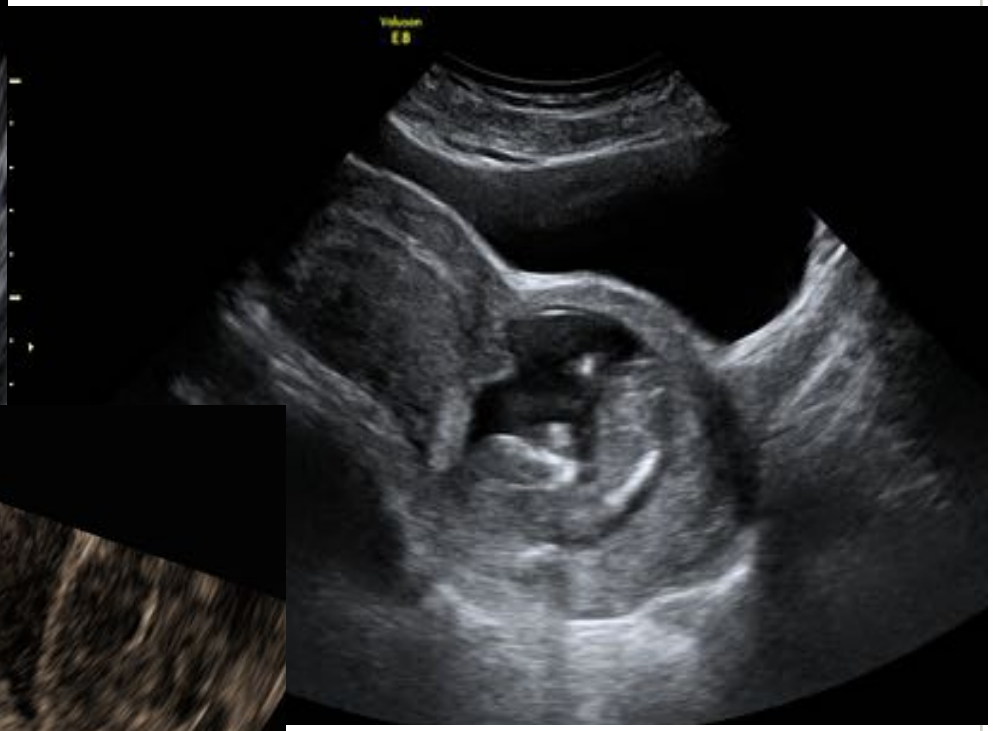


First Trimester Ultrasound Diagnosis of Fetal Abnormalities. Abuhamad-Chaoui. September 2017

6 weeks

16 weeks

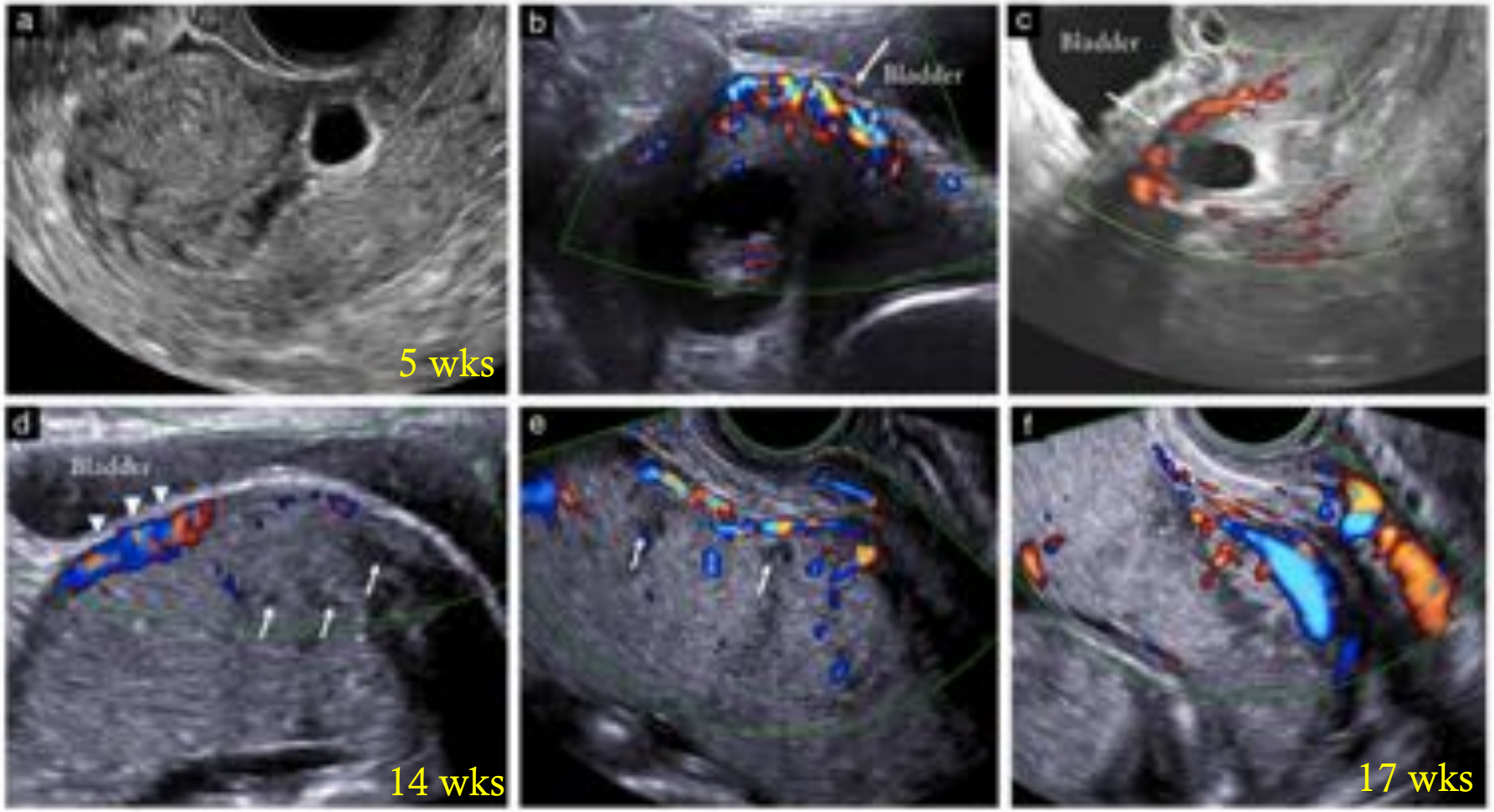
Cesarean scar pregnancy



First-trimester detection of abnormally invasive placenta in high-risk women: systematic review and meta-analysis

F. D'ANTONIO[1], I. E. TIMOR-TRITSCH[2], J. PALACIOS-JARAQUEMADA[3], A. MONTEAGUDO[2], D. BUCA[4], F. FORLANI[5], G. MINNECI[5], F. FOTI[5], L. MANZOLI[6], M. LIBERATI[4], G. ACHARYA[7] and G. CALÌ[5]

Ultrasound Obstet Gynecol 2018; **51**: 176–183



Undetectable at 11-13 wk

- **Embryology (CNS)**
- **Natural history**
- **Sequelae of fetal infection/hemorrhage**
  - Agenesis of corpus callosum (14-18wks GA), semilobar/ lobar HPE, microcephaly, craniosynostosis, cerebellar/ vermian hypoplasia ("open vermis" <18 wks GA)
  - Echogenic lung lesions (CCAM, BPS)
  - Duodenal atresia, bowel obstruction
  - Renal/GU abnormalities, ovarian cysts
  - Fetal tumors

Syngelaki A. et al. *Prenat Diagn* 2011; **31**: 90–102

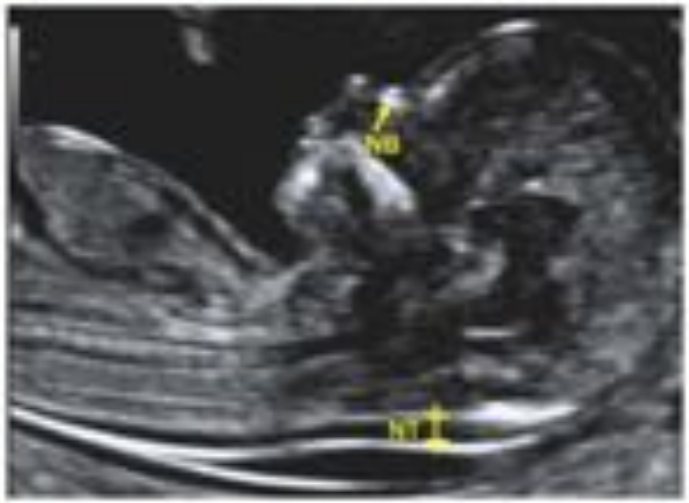
Summary

- Be systematic (use a checklist)
- Dating, # of fetuses/ chorionicity, CRL, BPD & NT
- Always detectable anomalies[1]
  - Body stalk anomaly
  - Acrania/ exencephaly-anencephaly
  - Alobar holoprosencephaly
  - Omphalocele
  - Ectopia cordis
  - Gastroschisis
  - Megacystis
  - Molar pregnancy
- * Midsagittal and axial view will exclude most major anomalies

[1]Syngelaki A. et al. *Prenat Diagn* 2011; **31**: 90–102

Certificates of competence

Nuchal translucency scan



Requirements for certification

The requirements for obtaining the FMF certificate of competence in the nuchal translucency (NT) scan are:

1. Attendance of the internet based course on the 11-13 weeks scan.
2. Submission of a logbook of 3 images demonstrating the measurement of NT.

11- 13 + 6 wks GA
CRL 45- 84 mm
Neutral position

Protocol for measurement

- The gestational period must be 11 to 13 weeks and six days.
- The fetal crown-rump length should be between 45 and 84mm.
- The magnification of the image should be such that the fetal head and thorax occupy the whole screen.
- A mid-sagittal view of the face should be obtained. This is defined by the presence of the echogenic tip of the nose and rectangular shape of the palate anteriorly, the translucent diencephalon in the centre and the nuchal membrane posteriorly. Minor deviations from the exact midline plane would cause non-visualization of the tip of the nose and visibility of the zygomatic process of the maxilla.
- The fetus should be in a neutral position, with the head in line with the spine. When the fetal neck is hyperextended the measurement can be falsely increased and when the neck is flexed, the measurement can be falsely decreased.
- Care must be taken to distinguish between fetal skin and amnion.
- The widest part of translucency must always be measured.
- Measurements should be taken with the inner border of the horizontal line of the callipers placed ON the line that defines the nuchal translucency thickness - the crossbar of the calliper should be such that it is hardly visible as it merges with the white line of the border, not in the nuchal fluid.
- In magnifying the image (pre or post freeze zoom) it is important to turn the gain down. This avoids the mistake of placing the calliper on the fuzzy edge of the line which causes an underestimate of the nuchal measurement.
- During the scan more than one measurement must be taken and the maximum one that meets all the above criteria should be recorded in the database.

Helpful References

- http://www.isuog.org/NR/rdonlyres/9225E408-C904-4A7F-84AE-812E456FBDDD/0/ISUOG1stTguidelines2013.pdf
- https://fetalmedicine.org/education/the-11-13-weeks-scan
- Fong KW et al. Detection of Fetal Structural Abnormalities with US during Early Pregnancy RadioGraphics 2004; 24:157–174