FIRST TRIMESTER FETAL ANATOMIC ASSESSMENT

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Disclosures

- Reem S. Abu-Rustum has no disclosures.

Lecture Objectives

By the end of this lecture the viewer should:

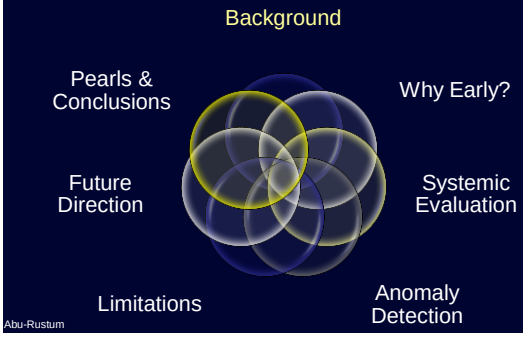
- Understand the reasons behind an 11-14 week anatomic assessment
- Understand how to carry out a systematic anatomic assessment at 11-14 weeks
- Understand the various type of anomalies amenable to detection at 11-14 weeks: what should be detected, what may be detected, and what cannot be detected
- Understand the limitations of fetal assessment at 11-14 weeks
- Understand the safety concerns at 11-14 weeks
- Understand future direction of fetal evaluation in the first trimester
- Take away key tips and pearls

Lecture Outline

First trimester fetal anatomic assessment at 11-14 weeks

- Background information
- Why early?
- How to carry out a systematic anatomic assessment at 11-14 weeks
- Detection of anomalies: what should be detected, what may be detected, and what cannot be detected
- Limitations and safety in the first trimester
- Future direction
- Tips and pearls

Objectives



Congenital Malformations

- Affect 3-5% of all pregnancies
- Most common cause of infant mortality
- Suboptimal detection 16-77%

RS Abu-Rustum. A Practical Guide to 3D Ultrasound. CRC Press 2015.

Rosano et al. J Epidimiol Community Health 2000; 54:660
Ewigman et al. NEJM 1993; 329: 821-827; Chitty et al. Prenat Gaid 1995; 15:1241

But…

Majority have no risk factors

MUST SCREEN
THE ENTIRE POPULATION

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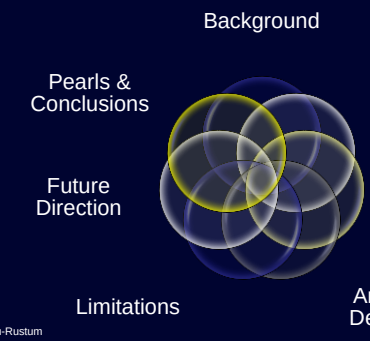
Objectives



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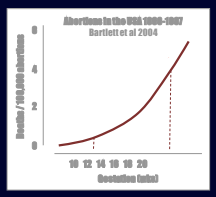
Why Detect Early? Main Considerations

- Workup
- Options
- TOP Limitations
- Safety
- Explain sudden IUFD
- Natural progression
- Psychological
- Obstetrical care
- Early Reassurance



Courtesy of Prof. Nicolaides

Barlett et al. Obstet Gynecol 2004; 103:729
Maiz et al. Prenatal Diagnosis 2016; ePub ahead of print.

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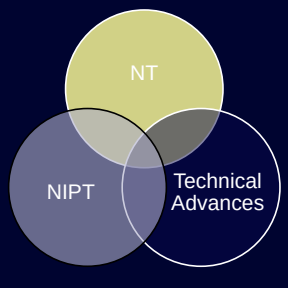
Global Implementation as a Result of…



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- Today we have access to more than 70% of pregnancies who are undergoing NT assessment at 11-14 weeks

Salveson et al. UOG 2011; 37:625

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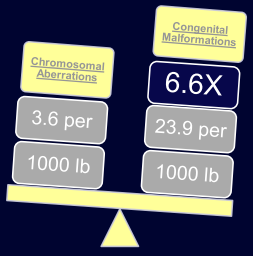
Keep In Mind…



Reiff et al. Prenatal Diagnosis 2016; 36:260

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… A Shift in the Role of NT with NIPT…

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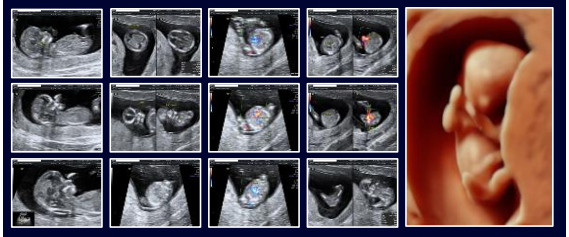
Full Anatomic Survey



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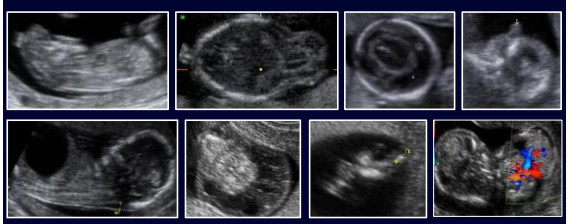
It is Now Possible to Rule out



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First Trimester Detection of Structural Anomalies

In the most ideal situations, FTS can detect

- Up to 82% of anomalies and
- 2/3 of cardiac lesions

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Oral Communication ISUOG 2007

Review

Accuracy of Ultrasonography at 11–14 Weeks of Gestation for Detection of Fetal Structural Anomalies

A Systematic Review

A. Cristina Rossi, MD, and Federico Prefumo, MD, PhD

Box 1. Detection Rate of Fetal Malformations in the First Trimester

100% detection rate
Acania, anencephaly, ectopia cordis, encephalocele

50–99% detection rate
Cystic hygroma, double-outlet right ventricular flow, Fallot, gastroschisis, omphalocele, holoprosencephaly, hypoplastic left heart syndrome, limb reduction, megacystis, polydactyly, septal defects, transposition of great vessels, valvular disease

1–49% detection rate
Spina bifida, hydrocephalus, skeletal dysplasia, facial cleft, Dandy-Walker, aortic coarctation, arthrogryposis

0% detection rate
Corpus callosum agenesia, bladder exstrophy, congenital cyst adenomatoid malformation, cerebellar hypoplasia, duodenal atresia, hydronephrosis, renal agenesia, duplex kidneys, bowel obstruction, extralobar sequestration

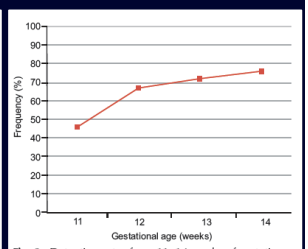

Fig. 3. Detection rates from 11–14 weeks of gestation. *Rossi. First-Trimester Ultrasonography. Obstet Gynecol 2013.*

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Rossi et al. AJOG 2013; 122:1160

Objectives

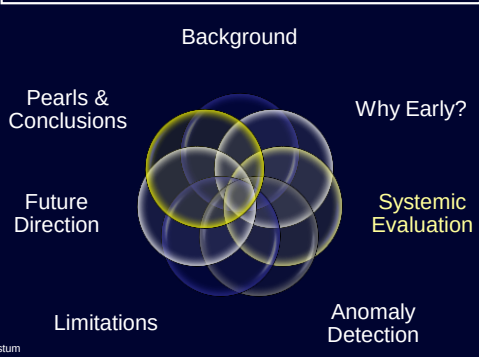

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Table 2 Suggested anatomical assessment at time of 11 to 13+6-week scan

Organ/anatomical area	Present and/or normal?
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 Cord	Size and texture Three-vessel cord*

*Optional structures. Modified from Fong *et al.*[28], McAuliffe *et al.*[87], Taipale *et al.*[60] and von Kaisenberg *et al.*[88].

isuog GUIDELINES

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

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Must Keep in Mind Timing…

- Limits are 11w0d-13w6d
- Ideally 12-13 weeks
- Skull ossifies at 10.5-11 weeks
- Fetuses have exompholos at 9-10 weeks that resolves by 12 weeks

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Systemic Approach

- Just as in the second trimester
- Fetal position and dexterity
- Placental localization
- Full anatomic evaluation

RS Abu-Rustum. A Practical Guide to 3D Ultrasound CRC Press 2015

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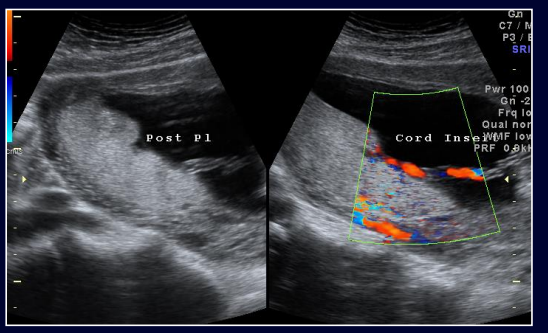
Placenta



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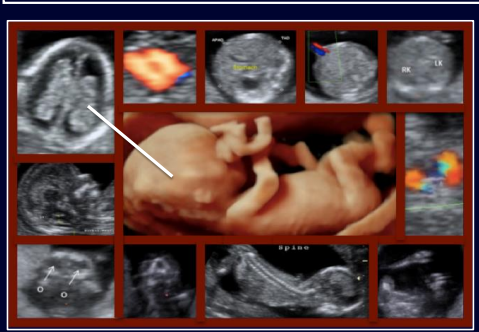
Systemic Approach



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Butterfly Sign

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Sepulveda et al. J Ultrasound Med 2004; 23:761

Posterior Fossa

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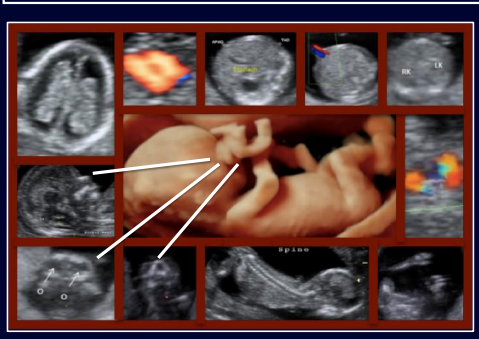
Systemic Approach



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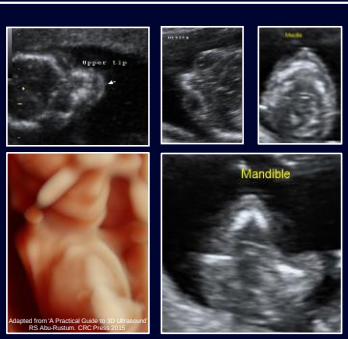
Face



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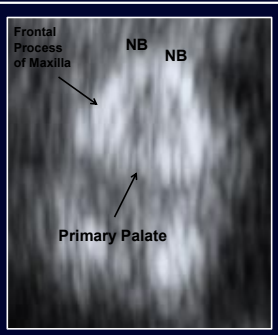
Retronasal Triangle



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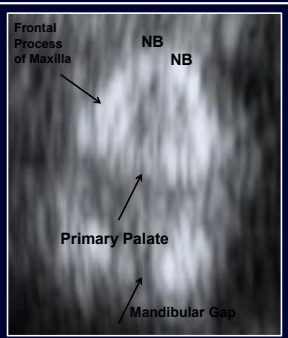
Sepulveda et al. UOG 2010, 35:7
J Ultrasound Med 2010, 29:1555

Mandibular Gap



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Sepulveda et al. UOG 2012; 39:152

Maxillary gap at 11–13 weeks' gestation: marker of cleft lip and palate

R. CHAOUI*, G. OROSZ†, K. S. HELING*, A. SARUT-LOPEZ* and K. H. NICOLAIDES†

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Chaoui et al. UOG 2015; 46:665

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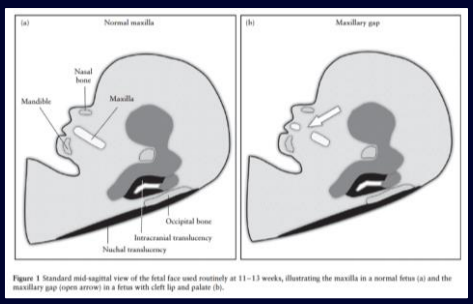

Figure 1 Standard mid-sagittal view of the fetal face used routinely at 11–13 weeks, illustrating the maxilla in a normal fetus (a) and the maxillary gap (open arrow) in a fetus with cleft lip and palate (b).

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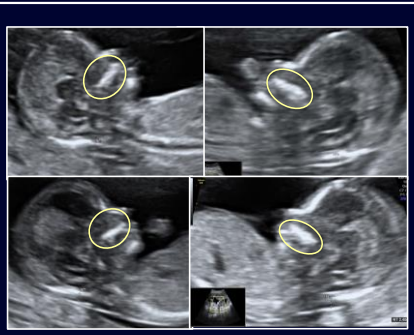
Chaoui et al. UOG 2015; 46:665

The Mid-Sagittal View



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Maxillary gap at 11–13 weeks' gestation: marker of cleft lip and palate

R. CHAOUI*, G. OROSZ†, K. S. HELING*, A. SARUT-LOPEZ* and K. H. NICOLAIDES†

MG Characteristic	Normal Controls (n=86)	Facial Clefts	
		Isolated (n=37)	Other Defects (n=49)
No Gap	**80 (93%)**	13 (35.1%)	2 (4.1%)
Partial Gap	6 (7%)	24 (64.9%)	34 (69.4%)
Complete Gap	-	-	13 (26.5%)
Gap < 1.5 mm	**6 (100%)**	11/24 (45.8%)	13/47 (27.7%)

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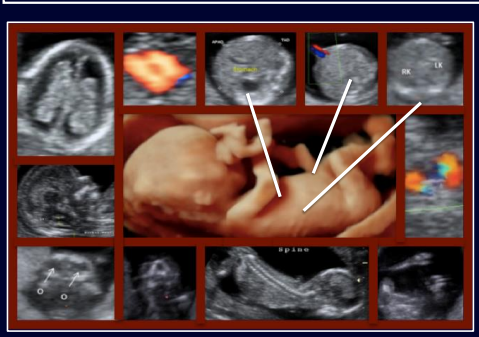
Chaoui et al. UOG 2015; 46:665

Systemic Approach



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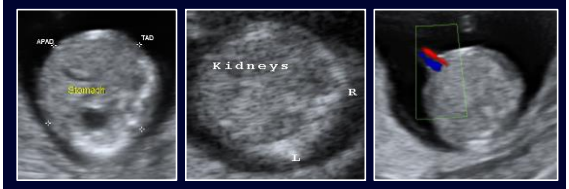
Abdomen



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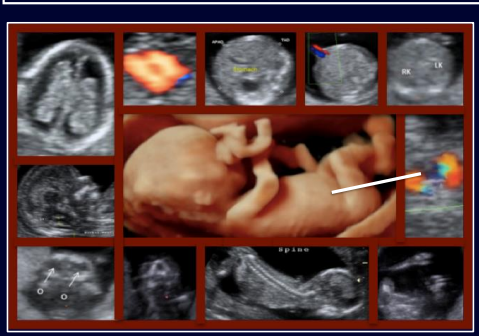
Systemic Approach



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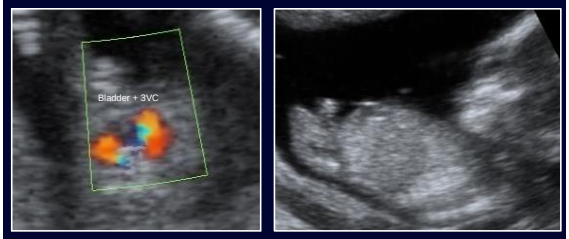
Pelvis



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Evanascent Pelvic Lucency in Anal Atresia

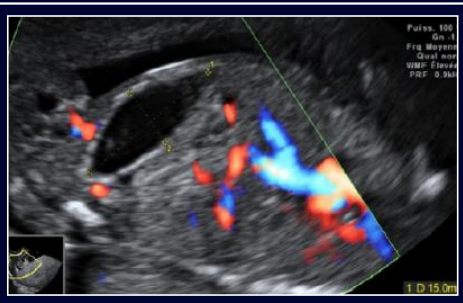

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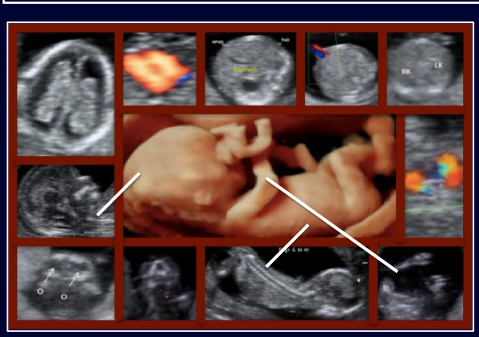
Image adapted from Bault et al. UOG 2010; 36:11

Systemic Approach



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Intracerebral Tanslucency

Chaoui et al. UOG 2009; 34: 249-252 and UOG 2010; 35:133

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Ratio of BPD to TAD in NTD

- BPD/TAD ~ 1

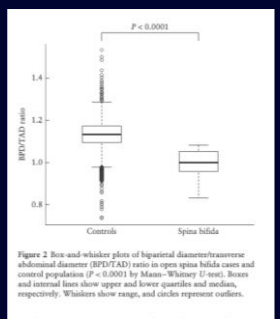

Figure 2 Box-and-whisker plots of biparietal diameter/transverse abdominal diameter (BPD/TAD) ratio in open spina bifida cases and control population ($P < 0.0001$ by Mann–Whitney U-test). Boxes and internal lines show upper and lower quartiles and median, respectively. Whiskers show range, and circles represent outliers.

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Simon et al. UOG 2015; 45:267

Spine

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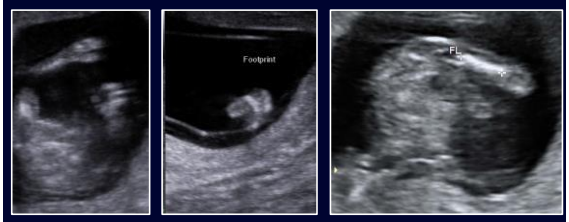
Extremities



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The First Trimester Sweep

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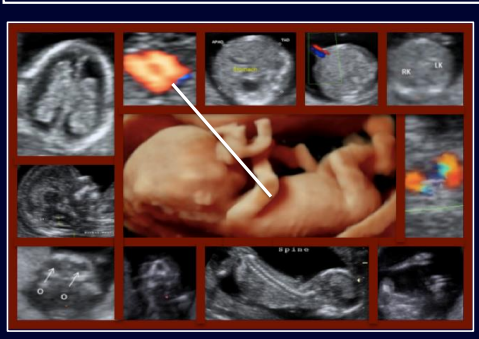
TV at 11w3d Using RIC 6-12

Systemic Approach



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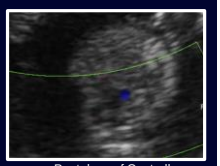
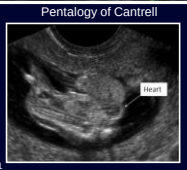
Congenital Heart Disease

- Most common major abnormality
- Incidence: 8.8/1000 live births
- 30% with associated defects
- Contributes to >50% of congenital anomaly-related deaths in childhood



Pentalogy of Cantrell



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Hoffman et al. Am J Cardio 1978; 42:641
Abuhamad & Chaoui. Practical Guide to Fetal Echocardiography: Normal and Abnormal **Hearts. 2nd Edition**

How Good Are We?

- Non-selected population in Norway
- 30149 fetuses

Detection Rate at

57%

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Tegnander et al. UOG 2006; 27:252

How Good Are We?

- Prospective 1 year study
- Northern California
- Fetuses and infants with CHD < 6 months
- 98/309 diagnosed (36%)

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Friedberg et al. J. Pediatr. 2009; 155:26

Can We Improve Our Detection?

Prenatal recognition of CHD rose from 17% in 1994 to 30% in 1995 and 36% in 1996.

Conclusions—A simple training program for obstetric ultrasonographers increased their ability to detect serious congenital heart disease at a routine 18–20 week anomaly scan.

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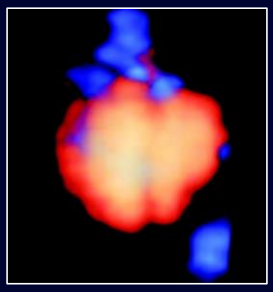
Hunter et al. Heart 2000; 84:294

The First Trimester Heart



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Keeping in Mind…

Rice Grain Coin: 1 Euro

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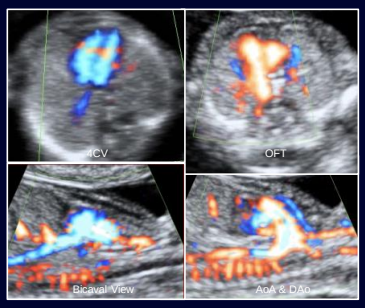
Allan, Cook & Huggon. Fetal Echocardiography: A Practical Guide. 2009

In Order to Obtain These Views



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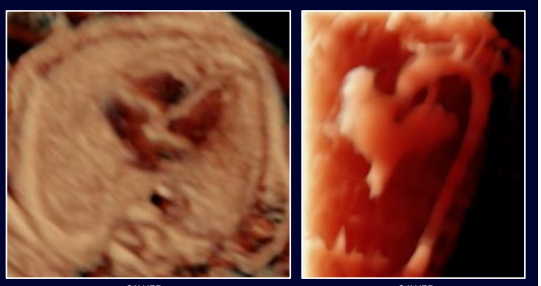
Must Acquire Skill in the Second Trimester…



21W5D 24W5D

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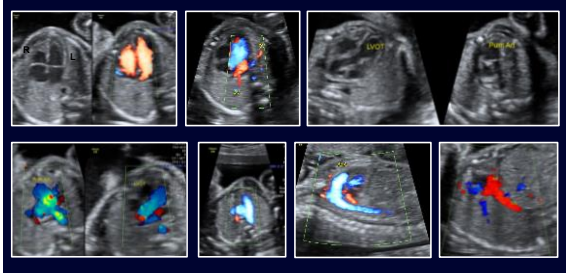
RS Abu-Rustum. A Practical Guide to 3D Ultrasound. CRC Press 2015

Systematic Approach 18-22 Weeks



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Nuchal Translucency

- Cardiac Abnormalities 5/1000 (0.5%)
- Diabetic Mom 10-15/1000 (1-1.5%)
- Previous Affected Child 20/1000 (2%)
- NT > 3.5 mm 50-70/1000 (5-7%)

Abu-Rustum Practical Guide to Fetal Echocardiography: Normal and Abnormal Hearts: Abuhamad and Chaoui 2009

Ultrasound Obstet Gynecol 2013; 42: 383–389
Published online 2 September 2013 in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/uog.12488

Nuchal translucency and major congenital heart defects in fetuses with normal karyotype: a meta-analysis

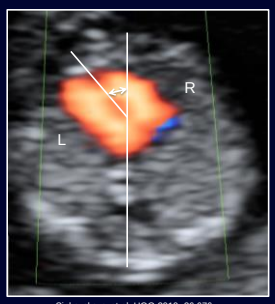
A. SOTIRIADIS*, S. PAPATHEODOROU†, M. ELEFTHERIADES‡ and G. MAKRYDIMAS§

*Second Department of Obstetrics and Gynecology, Ippokration General Hospital, Aristotle University of Thessaloniki, Thessaloniki, Greece; †Cyprus International Institute for Environmental and Public Health, Cyprus University of Technology, Limassol, Cyprus; ‡Embryocare, Prenatal Diagnostic Center, Athens, Greece; §Department of Obstetrics and Gynecology, University of Ioannina, Ioannina, Greece

- Pooled Sensitivity NT >95th Centile is **46%**
- Pooled Sensitivity NT >99th Centile is **21%**

Abu-Rustum Sotiriadis et al. UOG 2013; 42:383

Cardiac Axis 30-60º



Sinkovskaya et al. UOG 2010; 36:676
Sinkovskaya et al. UOG 2014; 44:10

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Original Research

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

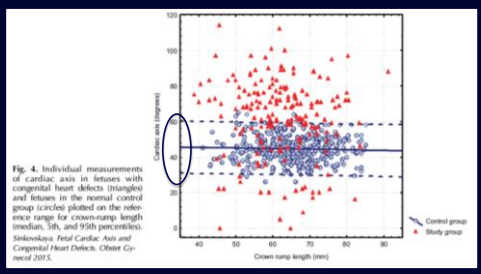


Fig. 4. Individual measurements of cardiac axis in fetuses with congenital heart defects (*triangles*) and fetuses in the normal control group (*circles*) plotted on the reference range for crown-rump length (median, 5th, and 95th percentiles). Sinkovskaya. Fetal Cardiac Axis and Congenital Heart Defects. Obstet Gynecol 2015.

Abu-Rustum Sinkovskaya et al Obstet Gynecol 2015; 125:453

Original Research

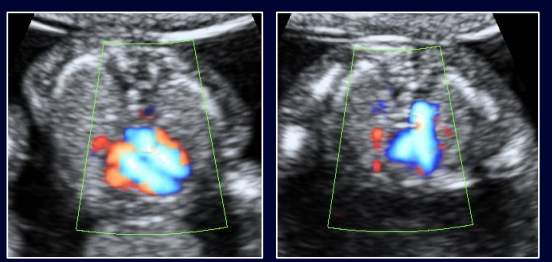
Fetal Cardiac Axis and Congenital Heart Defects in Early Gestation

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

Type of CHD	NT > 95th Centile	CAx Abnormal
Conotruncal	30.6%	81.6%
Univentricular Hearts	37.9%	96.6%
Combined CHD	57.9%	94.7%
Total	**51.7%**	**74.1%**

Abu-Rustum Sinkovskaya et al Obstet Gynecol 2015; 125:453

What Can We See?



Abu-Rustum

- Haak et al

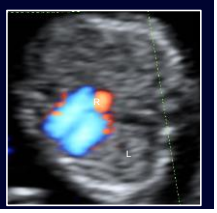
UOG 2002; 20:9

Transvaginal 92%

- Huggon et al

UOG 2002; 20:22

Transabdominally 84%



Abu-Rustum

Cardiac Imaging at 11-14 Weeks

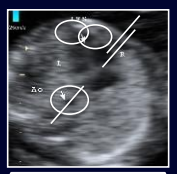
Key Points
- Heart Develops GA 5-8 Weeks
- Chest AP diameter is about 2.5 cm at 12-13 weeks

Can Assess
- Position
- Connections
- Symmetry of 4 Chambers
- 2 AV valves/Septum (Doppler)
- Septoaortic Continuity
- 2 Semilunar Valves (Doppler)
- Normal Cross Over of Arteries

Abu-Rustum

Anatomic Landmarks

- Right ventricle is the most anterior, below the sternum
- Left atrium is closest to the spine most central structure in the chest
- Aorta is just anterior to the left of the spine
- Tricuspid valve is more apical than mitral valve
- Flap of the foramen ovale in the left atrium
- Moderator band is in the right ventricle
- Apex formed by the left ventricle



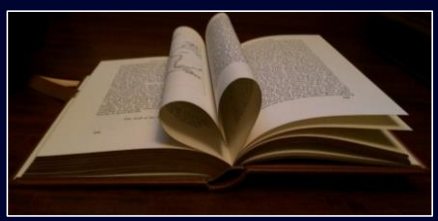
Abu-Rustum

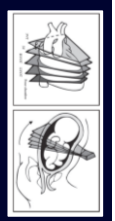
Anatomic Landmarks

- Left atrium and aorta occupy the center of the chest
- Aorta points to the right shoulder as it exits then heads posteriorly towards the spine
- Pulmonary artery (PA) points to the left shoulder as it exits
- Outflow tracts cross over, with the PA being more anterior than the left ventricular outflow tract
- Post bifurcation of the PA, the aorta and PA are almost parallel

Abu-Rustum

Systemic Evaluation Transverse Views





Abu-Rustum

Systemic Evaluation Sagittal Views

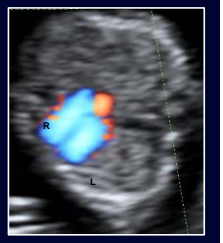
Abu-Rustum

Systemic Evaluation Transverse Views

Abu-Rustum

Diagram Courtesy of L. Dacu, MD

Establishing Situs



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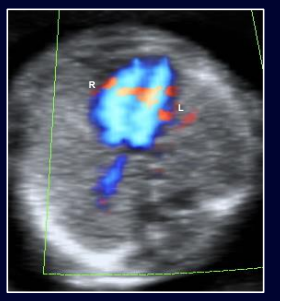
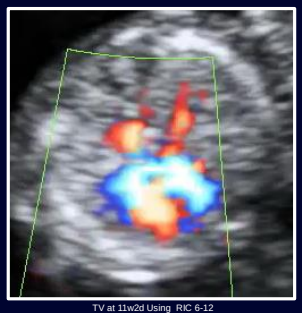
4 Chamber View



Abu-Rustum

4 Chamber View



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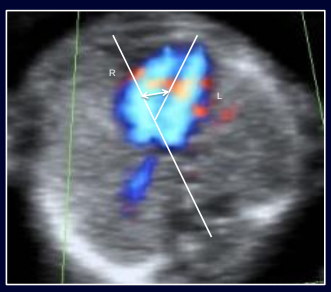
TV at 11w2d Using RIC 6-12

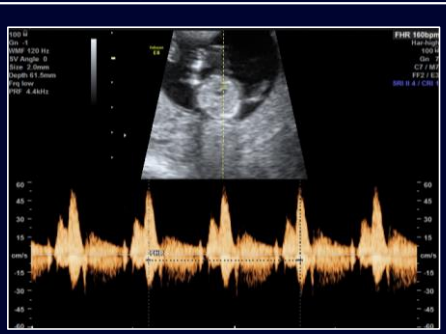
Cardiac Axis 30-60º



Sinkovskaya et al. UOG 2010; 36:676
Sinkovskaya et al. UOG 2014; 44:10

Abu-Rustum

Tricuspid Regurgitation



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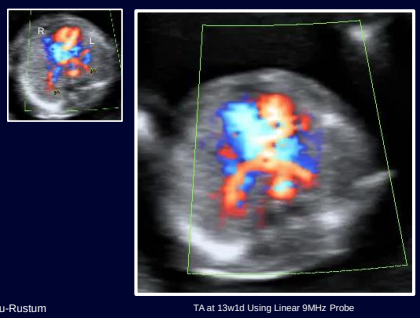
Pulmonary Veins



Abu-Rustum

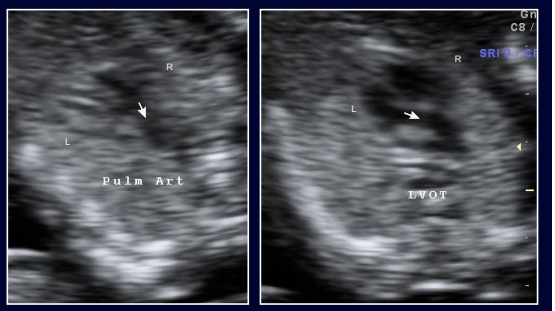
TA at 13w1d Using Linear 9MHz Probe

Systemic Evaluation Transverse Views

Abu-Rustum

Diagram Courtesy of L. Daou, MD

Outflow Tracts



Abu-Rustum

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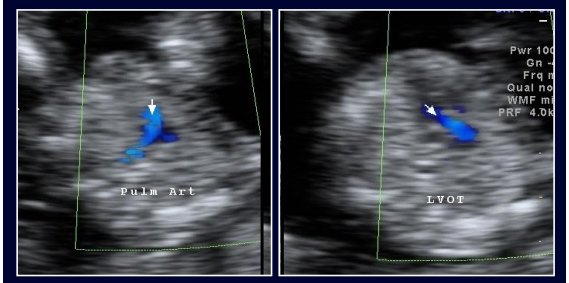

Abu-Rustum

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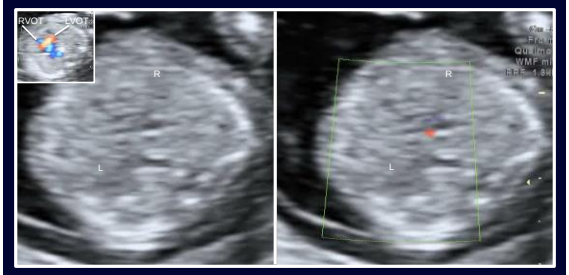

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TA at 13w5d Using RMC/OB

Cross Over

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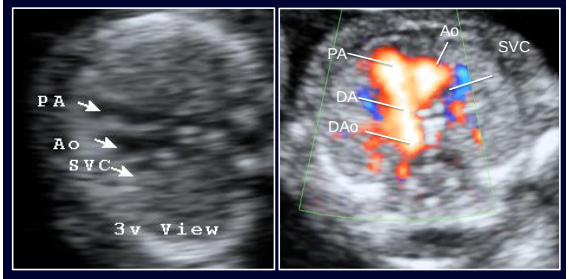
Systemic Evaluation Transverse Views

Abu-Rustum

Diagram Courtesy of L. Daou, MD

3 Vessel View



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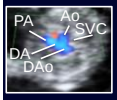
3 Vessel View

Abu-Rustum

TV at 13w1d Using RIC 6-12

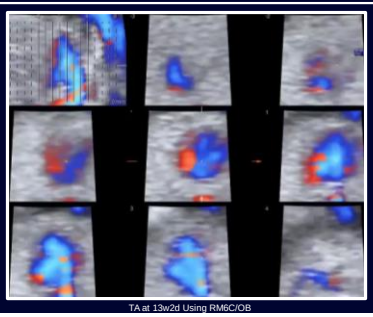
3 Vessel View



Abu-Rustum

TV at 9w5d Using RIC6-12

Systemic Evaluation Transverse Views



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TA at 13w2d Using RM6C/OB

Systemic Evaluation Sagittal Views

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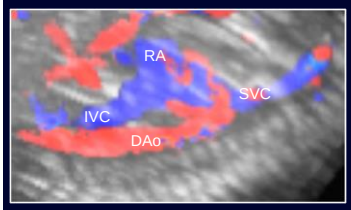
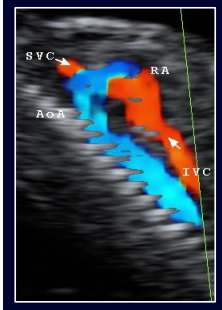
Right Atrial Inflow



Abu-Rustum

Right Atrial Inflow



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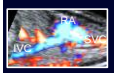
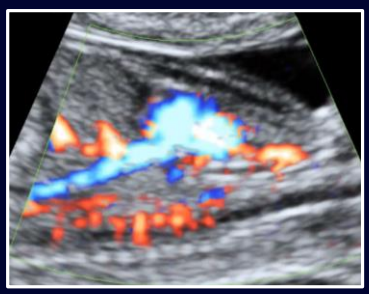
TA at 12w6d Using RM6C/OB

Right Atrial Inflow

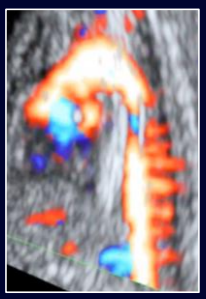




Abu-Rustum

TV at 13w1d Using RIC 6-12

Aortic Arch & Descending Aorta



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Aortic Arch & Descending Aorta

Abu-Rustum

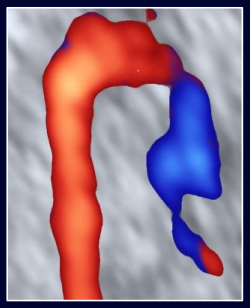
TA at 13w2d Using RM6C/OB

Aortic Arch & Descending Aorta

Abu-Rustum

TV at 13w1d Using RIC 6-12

Ductal Arch



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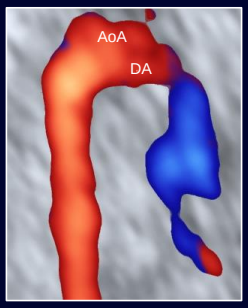
Ductal Arch



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STIC at 11-14 Weeks

Ultrasound Obstet Gynecol 2013; **42**: 669–678
Published online in Wiley Online Library (wileyonlinelibrary.com). **DOI:** 10.1002/uog.12548

Use of spatiotemporal image correlation at 11–14 weeks' gestation

C. VOTINO*, T. COS*, R. ABU-RUSTUM†, S. DAHMAN SAIDI*, V. GALLO*, O. DOBRESCU*, H. DESSY‡ and J. JANI*

*Departments of Obstetrics and Gynaecology, University Hospital Brugmann, Brussels, Belgium; †Center For Advanced Fetal Care, Tripoli, Lebanon; ‡Department of Pediatric Cardiology, Hôpital Universitaire des Enfants Reine Fabiola, Brussels, Belgium

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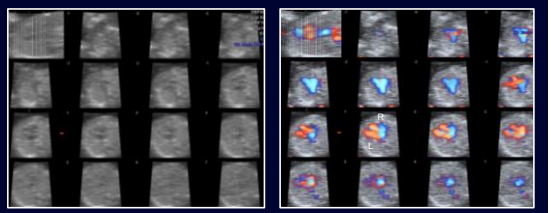
STIC Volume with HF Flow



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TA at 13w0d Using RM6C/OB

This work was presented as an oral poster at the 20th World Congress of the International Society of Ultrasound in Obstetrics and Gynecology; 2010; Prague, Czech Republic. It received the best Free Communication Award in its category.

TECHNICAL INNOVATION

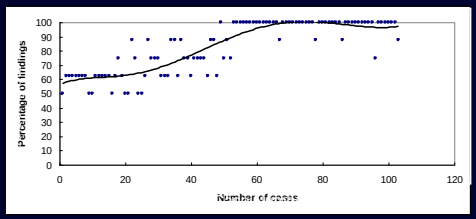
Learning Curve and Factors Influencing the Feasibility of Performing Fetal Echocardiography at the Time of the First-Trimester Scan

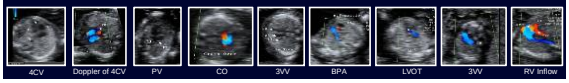
Reem S. Abu-Rustum, MD, M. Fouad Ziade, PhD, Sameer E. Abu-Rustum, MD

Abu-Rustum

Abu-Rustum et al. JUM 2011; 30:695

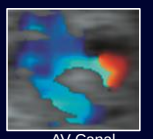
Results



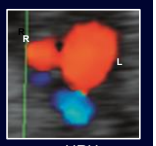


Conclusion

- Fetal cardiac evaluation is feasible in the first trimester
- At least 52 exams and an average time of 10 minutes needed
- Time allocation and gained sonographer experience are the most significant factors



AV Canal



HRH

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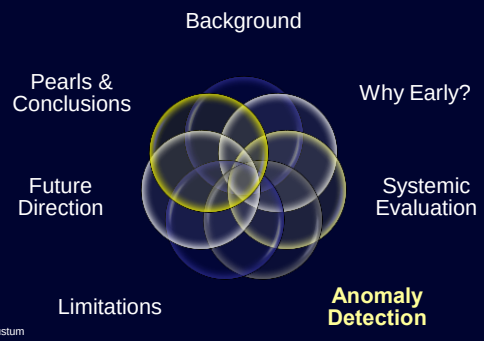
Objectives



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First Trimester Detection of Structural Abnormalities

Various Developmental Limitations with Structural Defects
Always detectable
Somewhat detectable
Never detectable

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•Acrania
•Cystic hygroma
•Exompholos/Gastroschicis
•Ectopia Cordis
•Megacystis
•Sirenomelia/Limb Anbormalities

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Acrania

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Cystic Hygroma

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Abu-Rustums, Dacou. J Ultrasound in Medicine 2010 ; 29:817

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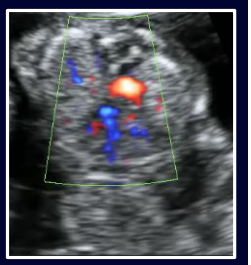
Exompholos



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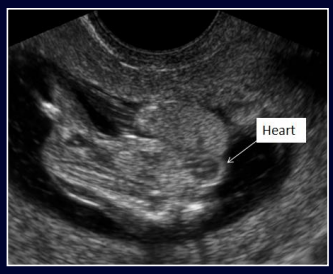
Ectopia Cordis at 11W6D



Pentalogy of Cantrell

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Abu-Rustum, Aoun. UOG 2010; 36:301

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Megacystis

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Abu-Rustums, Daou ,J Ultrasound in Medicine 2010 ; 29:817

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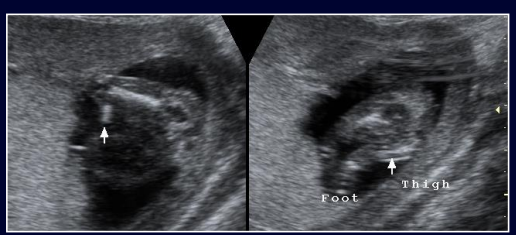
Limb Amputation



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Holoprosencephaly

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Probocis at 12W6d

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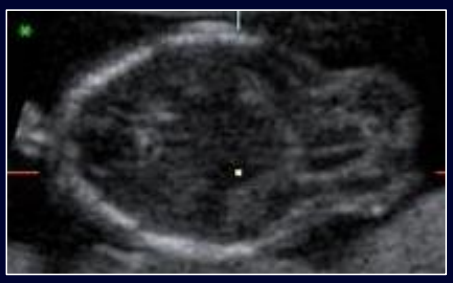
Cephalocele



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Abu-Rustums, Daou. J Ultrasound in Medicine 2010 ; 29:817

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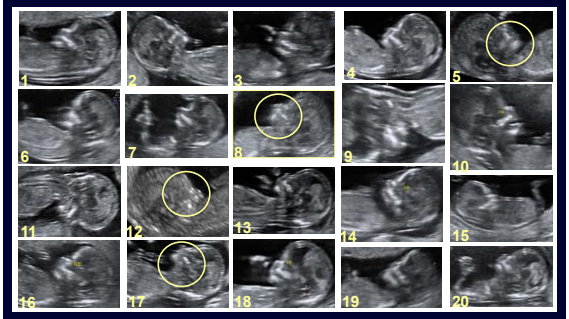
Abu-Rustum

Facial Clefts



Abu-Rustum

Abu-Rustum et al. Abstract presented at the AIUM Annual Convention NY 2016.

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NTD at 12W6D

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NTD at 12W6d

Abu-Rustum

NTD at 12W6D

Abu-Rustum

RS Abu-Rustum. A Practical Guide to 3D Ultrasound. CRC Press 2015

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Hypoplastic Right Heart

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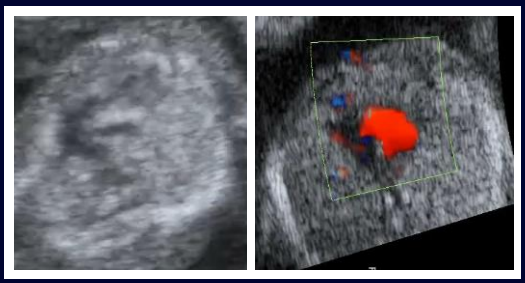
Univentricle



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Dextrocardia at 13W3D

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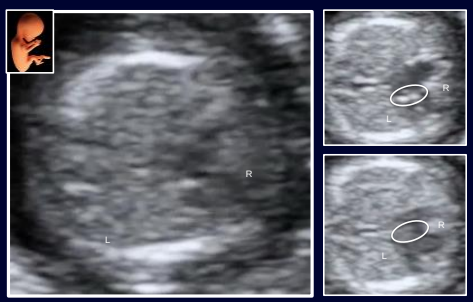
AV Canal



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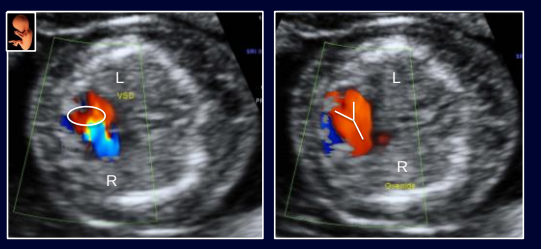
Tetralogy of Fallot at 12W6D



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First Trimester Detection of Structural Abnormalities

Somewhat Detectable
- Holoprosencephaly
- Cephalocele
- Choroid plexus cysts
- Facial clefts
- NTD
- Univentricle/HLHS/HRHS
- Congenital diaphragmatic hernia
- Skeletal dysplasias
- Renal agenesis/hydronephrosis
- Single umbilical artery
- Anal atresia

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P07.02
First trimester diagnosis of congenital diaphragmatic hernia: too good or too bad?

R. S. Abu-Rustum[1], H. Ayoubi[2], J. Jani[3]

[1]Center for Advanced Fetal Care, Tripoli, Lebanon; [2]Ob/Gyn, Monla Hospital, Tripoli, Lebanon; [3]Ob/Gyn, University Hospital Brugmann, Brussels, Belgium

Left CDH at 12w5d

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Abu-Rustum et al. UOG 2011; 38:190

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Rhizomelia at 12W6D

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Abu-Rustum et al. UOG 2010; 36 (supplement 1) 168-305

Talipes at 12W6D

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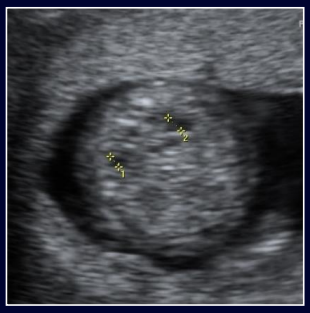
Renal Pelvises



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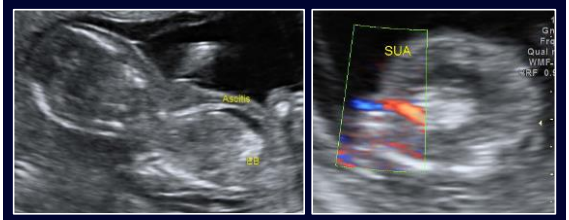
Single Umbilical Artery



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Anal Atresia

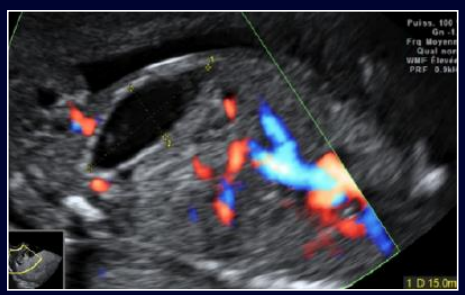

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Image adapted from Bault et al. UOG 2010; 36:11

First Trimester Detection of Structural Abnormalities

Various Developmental Limitations with Structural Defects
Always detectable
Somewhat detectable
Never detectable

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First Trimester Detection of Structural Abnormalities

Never Detectable?
- Dandy-Walker malformation
- Ventriculomegaly
- Agenesis of the corpus callosum
- Vermian agenesis
- Mild valvular heart abnormalities
- Late appearing coarctation of the aorta
- Pulmonary abnormalities
- Duodenal atresia
- Bowel obstruction
- UPJ obstruction and other mild renal abnormalities

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Dandy-Walker Malformation at 13W3D

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First Trimester Detection of Structural Abnormalities

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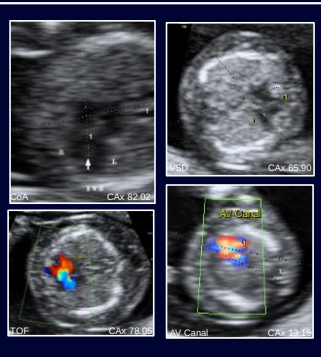
Role of the Cardiac Axis



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Pleural Effusion at 12W1D

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Objectives

Background

Why Early?

Systemic Evaluation

Anomaly Detection

Limitations

Future Direction

Pearls & Conclusions

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Safety in the First Trimester

alum

ISUOG statement on the safe use of Doppler in the 11 to 13 + 6-week fetal ultrasound examination

Bromley at al. JUM 2014; 33:1209

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Technical/Personal Limitations

Training

Machinery

Maternal Body Habitus

Developmental Stage

Time Consuming

Undue Anxiety

Greatest challenge is the LOW RISK PATIENT!

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Objectives

Background

Why Early?

Systemic Evaluation

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Future Direction

Pearls & Conclusions

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Anatomy at 8-10 Weeks? Votino et al UOG 2014; 44: 10

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9W4D Fetus CRL 28.7 mm

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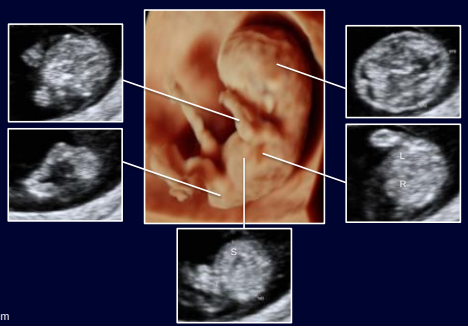
9W4D Fetus CRL 28.7 mm



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Unique to the First Trimester

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Unique to the First Trimester

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Results

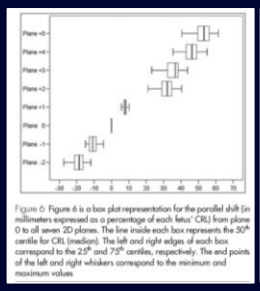

Figure 6 Figure 6 is a box plot representation for the parallel shift (in millimeters expressed as a percentage of each fetus' CRL) from plane 0 to all seven 2D planes. The line inside each box represents the 50th centile for CRL (median). The left and right edges of each box correspond to the 25th and 75th centiles, respectively. The end points of the left and right whiskers correspond to the minimum and maximum values

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Abu-Rustums, Ziade. Prenatal Diagnosis 2012; 32:875

Ultimately

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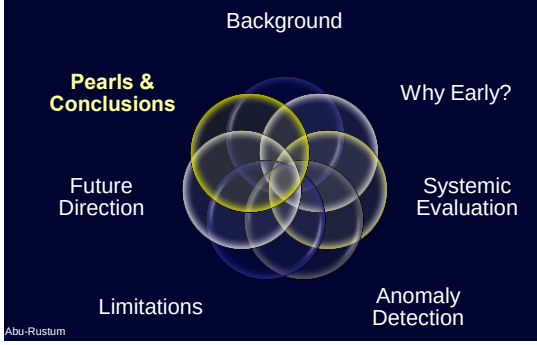
Objectives



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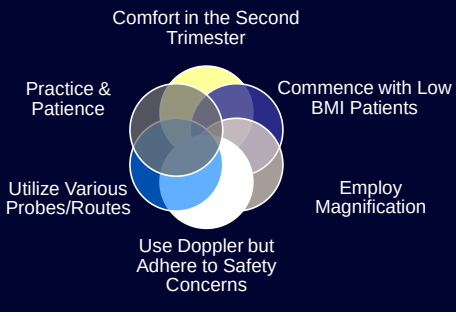
Practical Pearls



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Conclusion

- Detailed first trimester fetal assessment is feasible
- Critical role in the era of NIPT
- Powerful tool for early reassurance
- May diagnose over 70% of major anomalies/CHD
- Does not replace the second trimester scan
- Its incorporation into clinical practice is inevitable

IT IS TIME TO LOOK BEYOND THE NT

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Hippocrates

Learn the Past
and
Research the Present
to Predict
the Future…

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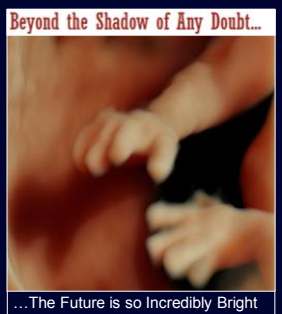
Thank You!



…The Future is so Incredibly Bright

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