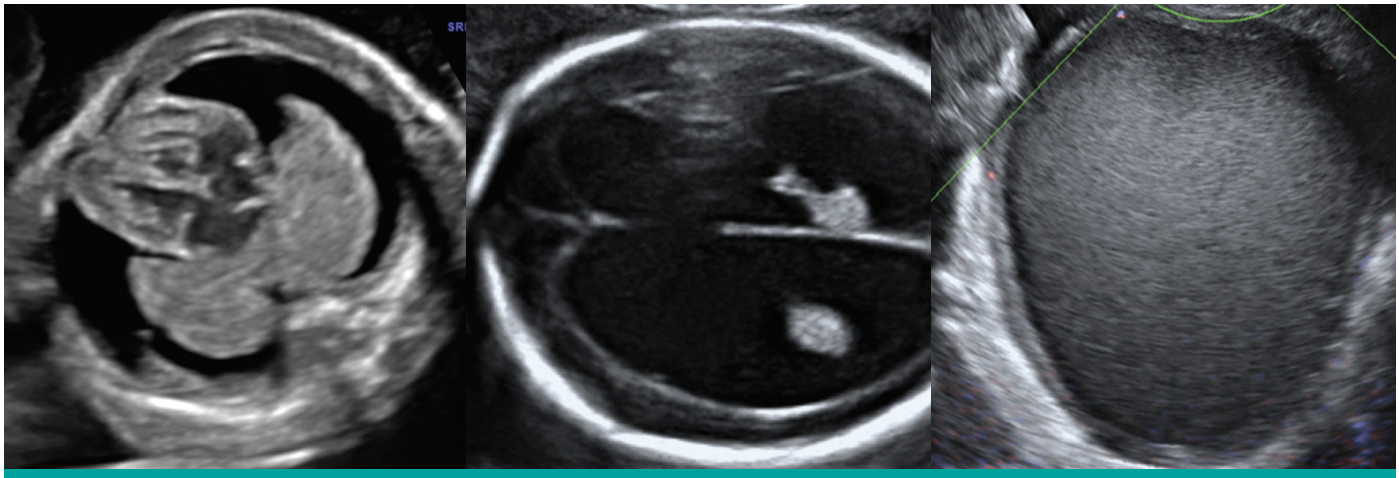

Advances in Ultrasound **in Obstetrics and Gynecology**

Friday, October 14, 2016

Omni Shoreham Hotel • Washington, DC

ESI Educational Symposia

Accreditation with Commendation

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Friday, October 14, 2016

Symposia At Sea
Practical Approach to Ultrasound in Obstetrics and Gynecology
Book By March 26, 2017

Coding in Ultrasound Imaging: Ensuring Compliance with Guidelines and Optimizing Reimbursement

James M. Shwayder, M.D., J.D.
Professor and Chair
Department of Obstetrics and Gynecology
University of Mississippi Medical Center
Jackson, MS

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Coding in Ultrasound Imaging: Ensuring Compliance with Guidelines and Optimizing Reimbursement

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Department of Obstetrics and Gynecology
University of Mississippi
Jackson, Mississippi

Coding in Ultrasound Imaging: Ensuring Compliance with Guidelines and Optimizing Reimbursement

James M. Shwayder, M.D., J.D.

Disclosures: None

Outline

- CPT coding
- ICD-9 and ICD-10
- Supervision requirements
- Appropriate documentation and coding
  - Obstetrical ultrasound
  - Gynecologic ultrasound
  - 3D/4D sonography

Coding Resources

- Procedures
  - Current Procedural Terminology
  - CPT® 2016
- Diagnosis
  - International Classification of Diseases
  - ICD-10-CM
- Resources
  - ACOG, AMA, AIUM

Procedural Coding

- CPT book sets the rules
- Descriptions are imperfect

ICD-10-CM Diagnosis Coding

- Diagnostic services during an encounter/visit
  - Sequence: diagnosis, condition, problem, or other reason (symptom) for encounter/visit
- Outpatient encounters for diagnostic tests and procedures and the final report is available at the time of coding
  - ***Code any confirmed or definitive diagnosis documented in the interpretation.***
  - Do not code related signs and symptoms as additional diagnosis

www.cdc.gov.nchs

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International Classification of Diseases (10th Revision) - ICD-10

- ICD-10 promotes international comparability in the collection, classification, processing and presentation of mortality statistics.
- Developed collaboratively between WHO and 10 international centers
- Effective Oct. 1, 2013 (Now delayed to 10/2015)
- The code-set will grow from its current 17,000 codes to more than 141,000, and the format is new with seven alpha-numeric codes instead of five numeric digits.

www.cdc.gov.nchs

CPT Coding and RVU's

CPT
- Professional component
- Technical component

RVU
- Relative value unit associated with each service
- 2016 Conversion $35.8043

www.cms.gov

Professional Component (-26)

The physician
- Supervises the test
- Interprets the test
- Prepares the written report

Technical Component (-TC)

Costs associated with
- The technician's salary/benefits
- The equipment
- Any necessary supplies

Fully Implemented Non-Facility Billing
A code reported without a modifier

Combines
- Professional component
- Technical component
- Any necessary supplies
- Image storage

Physician Supervision

- General Supervision
- Direct Supervision
- Personal Supervision

Medicare Requirements for Physician Supervision of Sonographers.
www.acog.org/departments

Physician Supervision
General Supervision

- Procedure is furnished under the physician's overall direction and control
- The physician's presence is not required during the performance of the procedure.
- The training of the nonphysician personnel who perform the diagnostic procedure and equipment maintenance are the responsibility of the physician

Medicare Requirements for Physician Supervision of Sonographers.
www.acog.org/departments

Physician Supervision
Direct Supervision

- The physician must be present in the office suite and immediately available to furnish assistance and direction throughout the performance of the procedure.
- The physician's in-room presence is not required during the performance of the procedure.

Medicare Requirements for Physician Supervision of Sonographers.
www.acog.org/departments

Physician Supervision
Personal Supervision

- Physician must be in attendance in the room during the performance of the procedure.

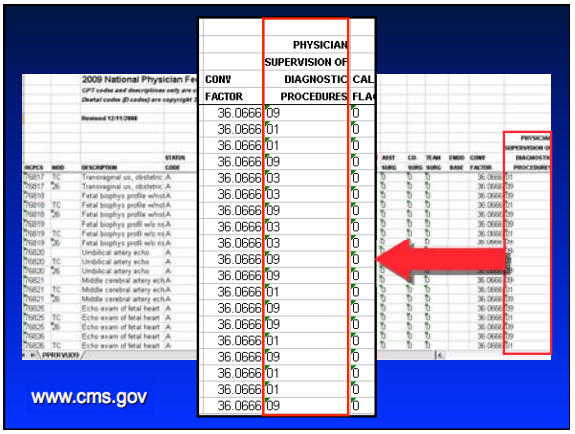
Medicare Requirements for Physician Supervision of Sonographers.
www.acog.org/departments

2013

Physician Supervision
Personal Supervision of Gyn US

- Sonohysterography (ultrasound)
  - 76831 - TC

Medicare Requirements for Physician Supervision of Sonographers.
www.acog.org/departments
www.cms.gov



Medicare Fee Schedule
Supervision Requirements

- 0 Procedure is not a diagnostic test or procedure is a diagnostic test that is not subject to the physician supervision policy.
- 1 Procedure must be performed under the general supervision of a physician.
- 2 Procedure must be performed under the direct supervision of a physician.
- 3 Procedure must be performed under the personal supervision of a physician.
- 9 Concept does not apply.

Coding – Ob Sonography
1st Trimester

- 76801 Ultrasound pregnant uterus, real time with image documentation, fetal and maternal evaluation, first trimester (< 14 weeks 0 days), *transabdominal approach*; single or first gestation
- 76802 ; each additional gestation. Add on code to 76801.

Coding – Ob Sonography
Vaginal Sonography

- 76817 Ultrasound pregnant uterus, real time with image documentation, transvaginal
- No contingency for multiple gestations
- If transvaginal examination is done in addition to transabdominal obstetrical ultrasound exam, use 76817 in addition to the appropriate transabdominal code

Coding – Ob Sonography
2nd/3rd Trimester

- 76805 Ultrasound pregnant uterus, real time with image documentation, fetal and maternal evaluation, after first trimester (≥ 14 weeks 0 days), transabdominal approach; single or first gestation
- 76810 ; each additional gestation.
  - Add on code to 76805

~~Level 1 Scan~~

Survey
- Viability (cardiac activity)
- Fetal number
- Fetal presentation
- Amniotic fluid volume
- Placental position

Fetal biometry
- BPD, HC, AC, FL, EFW

76805
Standard Content: Basic Scan

Survey
- Viability (cardiac activity)
- Fetal number
- Fetal presentation
- Amniotic fluid volume
- Placental position

Fetal biometry
- BPD, HC, AC, FL, EFW

Anatomic survey
- Head, face and neck, chest, abdomen, spine, extremities, gender

Maternal anatomy
- Cervix, adnexa, uterine anomalies

76805
Essential Elements of Anatomy

Head, face and neck
- Cerebellum, choroid plexus, cisterna magna, lateral ventricles, midline falx, lips

Chest
- 4-chamber cardiac view
- Outflow tracts

Abdomen
- Stomach, kidney, bladder, cord insertion, cord vessels (adrenal glands)

Spine
- Cervical, thoracic, lumbar, sacral

Extremities
- Legs and arms present or absent
- (comment on inability to visualize all extremities)

Fetal Imaging

Executive Summary of a Joint *Eunice Kennedy Shriver* National Institute of Child Health and Human Development, Society for Maternal-Fetal Medicine, American Institute of Ultrasound in Medicine, American College of Obstetricians and Gynecologists, American College of Radiology, Society for Pediatric Radiology, and Society of Radiologists in Ultrasound Fetal Imaging Workshop

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

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

Inability to Visualize Anatomy

Obese women

- Ultrasound at 20-22 weeks
  - 2 weeks later than in the nonobese patient

Inability to Visualize Anatomy

- If fetal anatomy cannot be assessed completely
- Follow-up examination in 2-4 weeks
- Comment on any limitation of the exam
- Follow-up
- Only as clinically indicated

Coding – Ob Sonography 2nd/3rd Trimester

- 76811 Ultrasound pregnant uterus, real time with image documentation, maternal evaluation plus *detailed fetal evaluation*, transabdominal approach; single or first gestation
- 76812 ; each additional gestation.
  - Add on code to 76811

Detailed Anatomic Examination 76811

Performed when an anomaly is suspected on the basis of history, biochemical abnormalities, or the results of either the limited or standard [basic] scan

SMFM Statement on 76811

Because this code is assigned more RVUs than the basic obstetrical sonogram (76805), the SMFM believes the code describes an examination involving significantly more work, and requiring greater expertise than that required for 76805.

SMFM Statement on 76811

Additionally, sophisticated equipment, rather than typical office level ultrasound machines, will be required to obtain the necessary imaging detail.

SMFM Statement on 76811

The level of expertise required to perform this examination can generally only be obtained through the extended education beyond residency that is acquired in a fellowship in Maternal-Fetal Medicine or Radiology…Use of this code by general obstetricians should be the exception rather than the rule.

AIUM – 76811
Consensus Statement

- Previous fetus or child with a congenital, genetic, or chromosomal abnormality
- Known or suspected fetal anomaly or known growth disorder in current pregnancy

76811 Task Force. J Ultrasound Med 2014; 33:189-195.

AIUM – 76811
Consensus Statement

Fetal at increased risk for a congenital anomaly:

- Maternal pregestational diabetes or gestational diabetes before 24 weeks
- High BMI (≥ 35 kg/m^2)
- Multiple gestation
- Abnormal maternal serum analytes
- Teratogen exposure
- 1st trimester NT ≥ 3.0 mm

76811 Task Force. J Ultrasound Med 2014; 33:189-195.

AIUM – 76811
Consensus Statement

Other conditions affecting the fetus:

- Congenital infections
- Maternal drug dependence
- Isoimmunization
- Oligohydramnios
- Polyhydramnios

76811 Task Force. J Ultrasound Med 2014; 33:189-195.

76811

655.8

- Suspected or known chromosomal abnormality

796.5

- Abnormal finding on antenatal screening

278.01

- Severe obesity (BMI ≥ 35)

Coding – Ob Sonography
Limited study

- 76815 Ultrasound pregnant uterus, real time with image documentation, limited (e.g., fetal heart beat, placental location, fetal position and/or qualitative amniotic fluid volume), one or more fetuses
- Use 76815 only once per exam and not per element

Coding – Ob Sonography

76815 Limited Examination
A limited examination is performed when a specific question requires investigation. For example, a limited examination could be performed to confirm fetal heart activity in a bleeding patient or to verify fetal presentation in a laboring patient. In most cases, limited sonographic examinations are appropriate only when a prior complete examination is on record.

Coding – Ob Sonography
2nd/3rd Trimester, Follow-up study

- 76816 Ultrasound pregnant uterus, real time with image documentation, follow-up (e.g., re-evaluation of fetal size by measuring standard growth parameters and amniotic fluid volume, re-evaluation of organ system(s) suspected or confirmed to be abnormal on a previous scan), transabdominal approach, *per fetus*
- Report 76816-59 for each additional fetus examined in a multiple pregnancy.

Coding – Ob Sonography
2nd/3rd Trimester

- What about the patient who presents for a repeat study later in the pregnancy?
- Code by status of indication
  - If new indication, use 76805
  - If not new, use 76816
    - Even if complete biometry and amniotic fluid assessment performed

Coding – Ob Sonography
Biophysical Profile

- 76818 Fetal biophysical profile; with non-stress testing
- 76819 Fetal biophysical profile; without non-stress testing

Coding – Ob/Gyn Sonography
Fetal Echocardiography

- 76825 Fetal initial (2D +/- m-mode)
- 76826 F/U or repeat (2D +/- m-mode)
- 76827 Doppler echo - initial
- 76828 Doppler echo – F/U or repeat
  - Add to 76825, 26826
- 93325 Color mapping
  - Add to 76825, 76826, 76827, 76828

Coding – Ob/Gyn Sonography
Fetal Evaluation

- 76820 Umbilical artery Doppler
- 76821 Middle cerebral artery Doppler

Coding – Ob/Gyn Sonography
3-D Rendering

- 76376 and 76377
  3-D rendering with interpretation and reporting of computed tomography, magnetic resonance imaging, ultrasound, or other tomographic modality
- Add on codes to appropriate ultrasound code(s)

Coding – Ob/Gyn Sonography
3-D Rendering

- 76376 3-D rendering…not requiring image postprocessing on an independent workstation.
- 76377 3-D rendering…requiring image postprocessing on an independent workstation.

Coding in Ob-Gyn Sonography
Modifiers

- 22 Unusual complexity
- 26 Professional component
- 52 Reduced services
- 59 Distinct procedural service, same day (e.g., referral for suspected fetal anomaly on the same day.
  - Ob uses 76805
  - Consultant uses 76811-59

Coding – Ob Sonography
Nuchal Translucency

- 76813 Ultrasound pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal; single or first gestation (List separately in addition to code for primary procedure)

Coding – Ob Sonography
Nuchal Translucency

- 76814 Ultrasound pregnant uterus, real time with image documentation, first trimester fetal nuchal translucency measurement, transabdominal or transvaginal; each additional gestation (List separately in addition to code for primary procedure)

ICD-9 Codes

- Use all that apply
- Prioritize
- Sparingly use "V codes" (screening codes) as a primary indication
- Note: Advanced maternal age may not be accepted as an indication for ultrasound or amnio
  - Can use "suspected or known chromosomal abnormality" (655.8)
  - May use diagnosis as reflected on final report

Coding - Gyn Ultrasound

- Vaginal sonography
  - Dimensions
  - Morphology
  - Dynamic studies
  - 3-D
- Abdominal sonography
- Sonohysterography

76830 –Echography, transvaginal

- Complete evaluation of the female pelvic anatomy – vaginal study
- Elements
  - Description and measurements of uterus and adnexal structures (cervix)
  - Measurement of the endometrium
  - Description of the cul-de-sac and fluid
  - Description of the bladder (if applicable)
  - Description of any pelvic pathology

Adnexa

Ovaries
- Dimension
  - Length
  - Width
  - Depth
- Morphology
- Motion
- Doppler

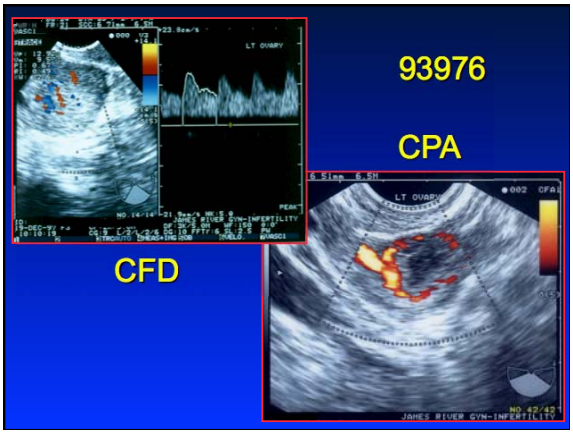
- Fallopian Tubes
  - Usually not visualized

76856 – Gyn Abdominal (add to TVS)

- Complete evaluation of the female pelvic anatomy – abdominal study
- Elements
  - Description and measurements of uterus and adnexal structures
  - Measurement of the endometrium
  - Measurement of the bladder (when applicable)
  - Description of any pelvic pathology

76857 – Gyn Limited or follow-up

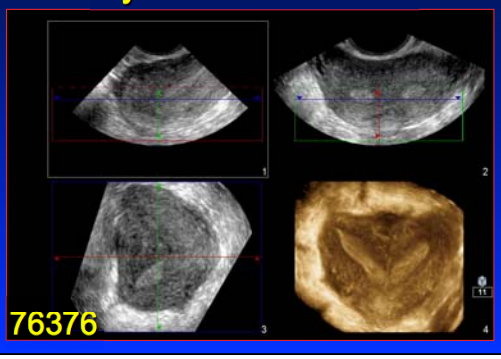
- Ultrasound, pelvic (nonobstetric), real-time with image documentation; limited or follow-up (e.g. for follicles)
- 76857
  - Used if follow-up of urinary bladder alone, i.e. post-void residual, imaged
- 51798
  - Used for post-void residual non-imaging: i.e. Bladder scan



Coding Gyn Sonography
Doppler Studies

- 93975 Duplex scan of A/V flow: Abdomen and pelvic – Complete
- 93976 Duplex scan of A/V flow: Abdomen and pelvic - Limited

Gyn ultrasound – 3D



76942

- 76942 Ultrasonic guidance for needle placement imaging supervision and interpretation)

76998 – Intraoperative Ultrasound

- Ultrasound guidance, intraoperative
- 76998
  - Ultrasound guided follicular aspiration
  - Ultrasound guided transfer
  - Ultrasound guided insemination

76998 – Intraoperative Ultrasound

Ultrasound guidance, intraoperative

- Documentation may be incorporated into the operative report. A separate report is not required
  - Reimbursement for TC = 0.00

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Sonohysterography

- 76831 Hysterosonography; with or without color flow Doppler
  - Includes elements of TVS, therefore is no separate charge for TVS
- 58340 Introduction of contrast agent or saline

Sonosalpingography

- 76831 Saline infusion sonohysterography (SIS), including color flow Doppler, when performed
  - Includes all elements of 76830 (TVS)
- 58340 Catheterization and introduction of saline or contrast material for saline infusion sonohysterography (SIS) or hysterosalpingography

Endometrial Cryoablation

- 58356 Endometrial cryoablation with ultrasonic guidance, including endometrial curettage, when performed
- Code 58356 cannot be reported with CPT codes 58100, 58120, 58340, 76700, 76856

CPT Coding Rules

- Pre-service work can be reported only if "significant and separately identifiable."
- Discussions of procedure & obtaining informed consent is NOT reported separately

CPT Coding Rules

- Pre-service work can be reported if:
  - Performing another procedure or evaluating another problem
  - Evaluating the patient and decide to perform an ultrasound during the visit

Coding in OB-Gyn Sonography

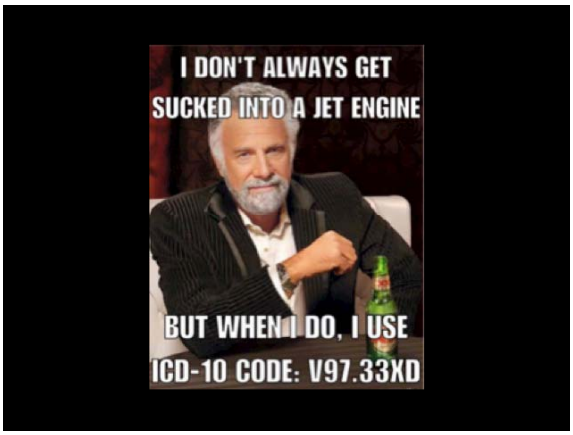
- Physician interpretation and signed final report are components of all codes
- A signed note in the progress notes or patient chart is adequate
- It is preferable to take photographs and place with the note (compliance issues)
- It is preferable to have a formal, final report, retaining all images for the SOL

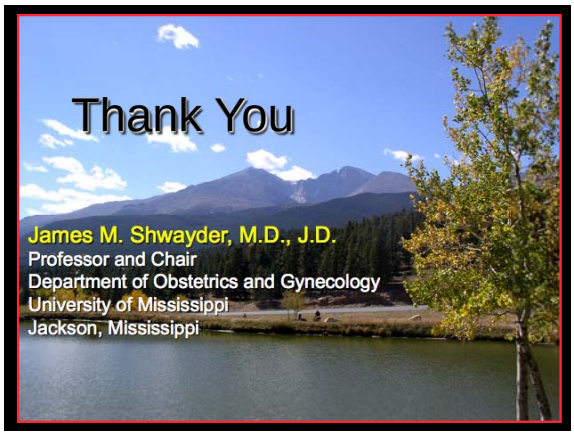
CPT General Coding Rules

- The diagnosis code should demonstrate the medical necessity for the procedure
- Report only the procedures that were performed and documented

CPT Coding Rules

- Do not change the codes reported in order to obtain reimbursement for non-covered services.
- Report the highest valued procedure code first on the claim form.





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The Second Trimester Genetic Sonogram: What Does It Entail and Is It Still Useful?

Jude P. Crino, M.D.
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Contribution Not Received in Time for Inclusion.

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The Normal and Abnormal Fetal Skeleton

Lawrence D. Platt, M.D.
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Professor (Clinical) Ob Gyn David Geffen School of Medicine at UCLA
Los Angeles, CA

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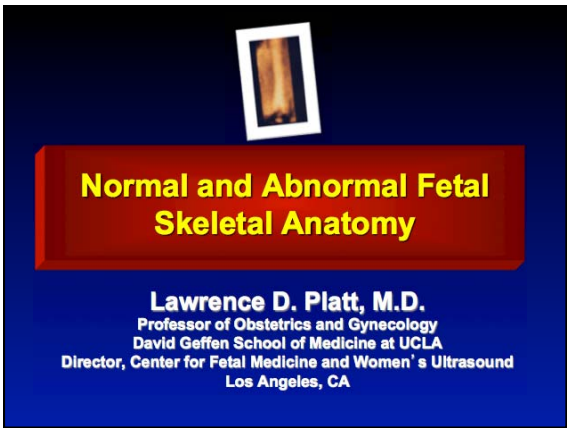
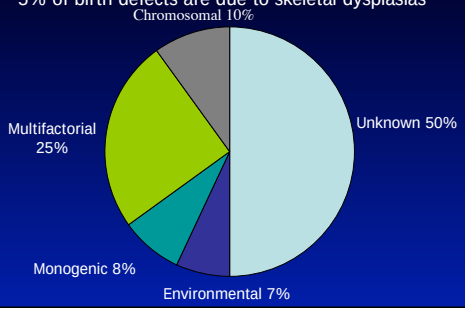


Normal and Abnormal Fetal Skeletal Anatomy

Lawrence D. Platt, M.D.
Professor of Obstetrics and Gynecology
David Geffen School of Medicine at UCLA
Director, Center for Fetal Medicine and Women's Ultrasound
Los Angeles, CA

Approximate Birth Prevalence

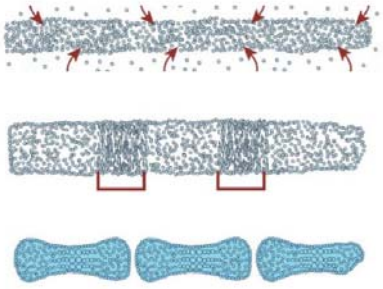
Overall birth incidence:
3% of newborns are affected by a birth defect
5% of birth defects are due to skeletal dysplasias



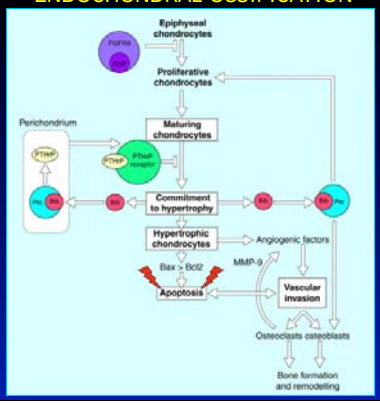
Approximate Birth Prevalence

Dysplasia	Prevalence
Achondroplasia	1/10,000
Thanatophoric dysplasia	1/10,000
Osteogenesis imperfecta type II	1/20,000
Achondrogenes	1/40,000
Asphyxiating thoracic dysplasia	1/70,000
Congenital hypophospatasia	1/100,000
Campomelic dysplasia	1/110,000
Chondrodysplasia punctate (rhizo)	1/150,000

ENDOCHONDRAL OSSIFICATION

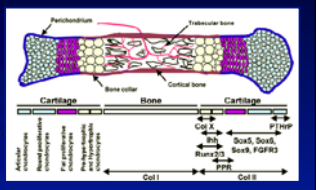


ENDOCHONDRAL OSSIFICATION



Endochondral Ossification

- Clavicle and mandible (8 wks)
- Appendicular long bones, phalanges, ileum and scapula (12 wks)
- Metacarpals and metatarsals (12-16 wk)
- Secondary ossification centers calcaneus (20 wks)
- Distal femur, proximal tibia and proximal humerus (>28 wks)



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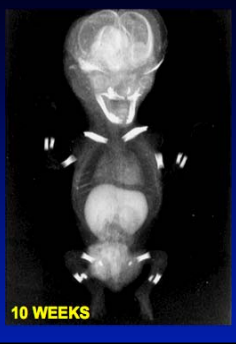
Endochondral Ossification

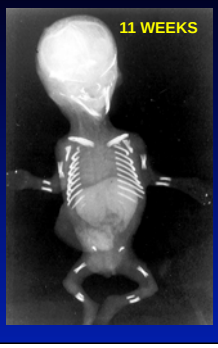
19 WEEKS

Courtesy of Rieteke M. van Zalen-Sprock

Endochondral Ossification



10 WEEKS



11 WEEKS

Endochondral Ossification

12 WKS

14 WKS

Skeletal Bones: Normal

Profile: 16 Weeks

Hand: 16 Weeks 2D

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Sternum: 16 Weeks

Feet: 16 Weeks 2D

Ischia: 19 Weeks 2D

Skeletal: 20 Weeks

LE: 20 Weeks

Metatopic Suture: 20 Weeks

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Hands: 20 Weeks 3D VCI Omni View

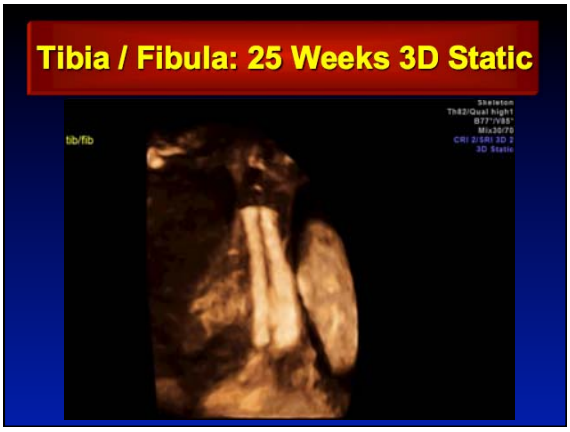

Ileum: 21 Weeks 2D


Sternal Center: 21 Weeks


Radius / Ulna: 25 weeks 2D

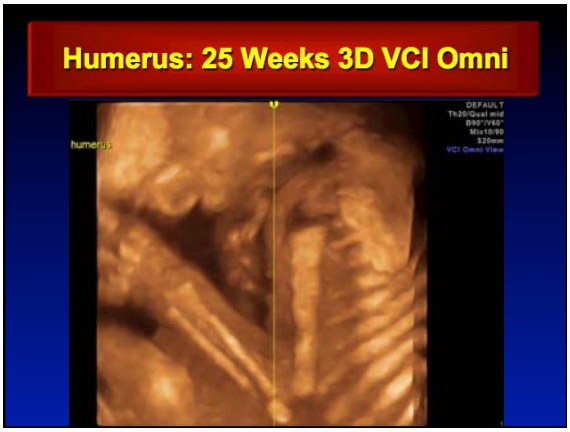

Radius / Ulna: 25 Weeks 3D Static

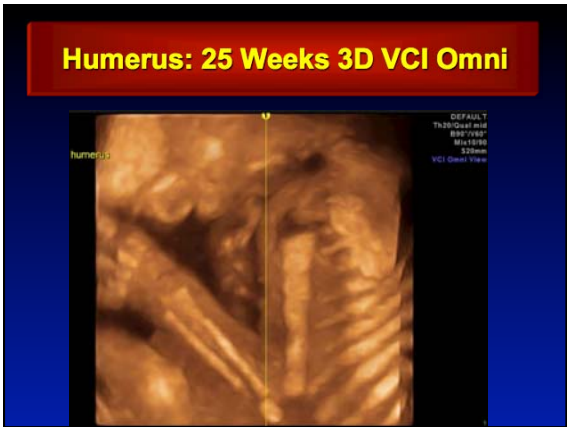

Tibia / Fibula: 25 Weeks 3D VCI Omni

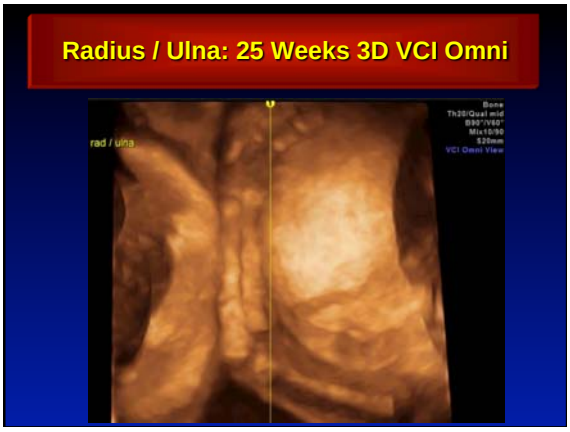

Tibia / Fibula: 25 Weeks 3D Static
tib/fib


Humerus: 25 Weeks 2D
humerus


Humerus: 25 Weeks 3D Static
humerus

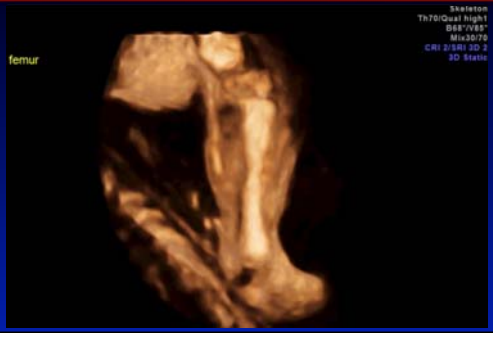

Humerus: 25 Weeks 3D VCI Omni
humerus


Humerus: 25 Weeks 3D VCI Omni
humerus


Radius / Ulna: 25 Weeks 3D VCI Omni
rad / ulna

Femur: 25 Weeks 2D

Femur: 25 Weeks 3D Static



Femur: 25 Weeks 3D VCI Omni View

Sternal Ossification

Sternal Ossification

Scapula: 2D

Scapula: 3D Static

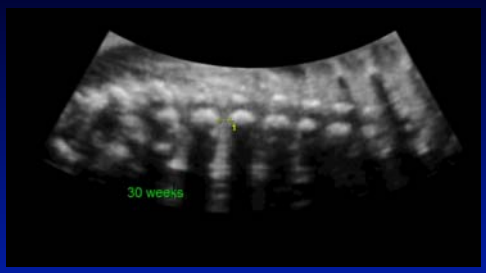
Hand: 28 Weeks 2D

Feet: 28 Weeks

Hand: VCI A

Hand / Arm: 2D

Spine: 30 Weeks



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Hand: 3D Static

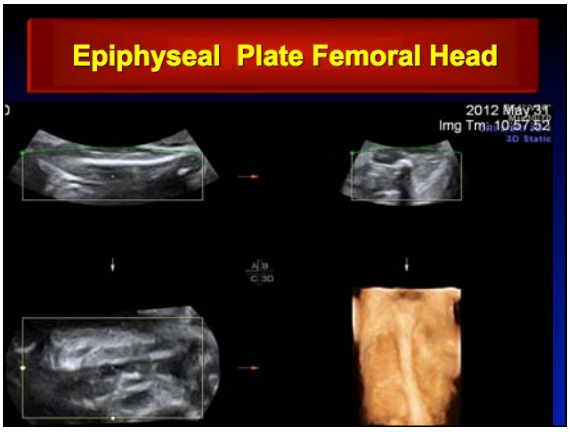
Normal Long Bones

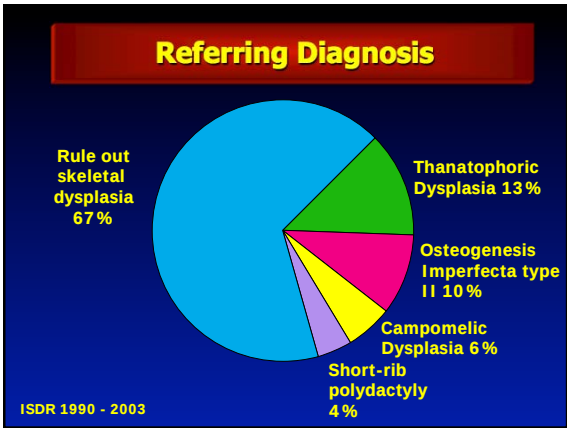
Normal Long Bones

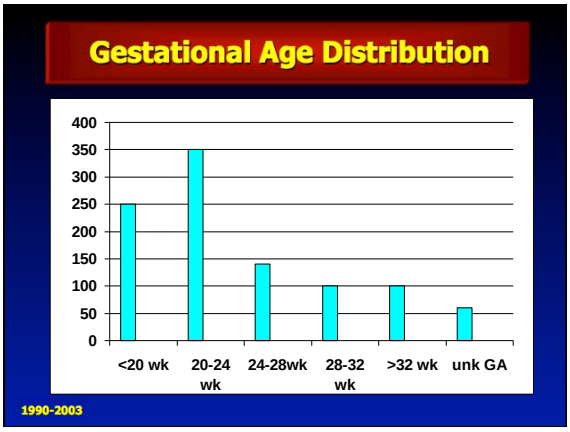
Normal Profile

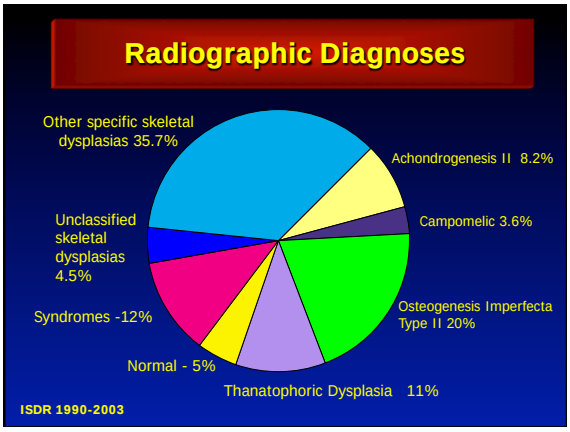
Normal Profile

Normal Profile









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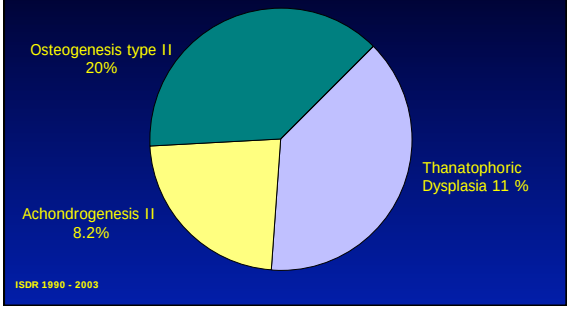
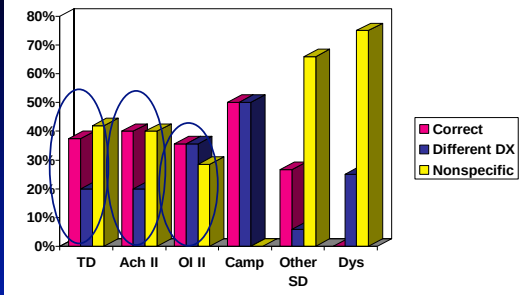
Most Common Diagnoses in Prenatal Onset Osteochondrodysplasias



ISDR 1990 - 2003

Comparison of U/S vs. Radiographic Diagnosis



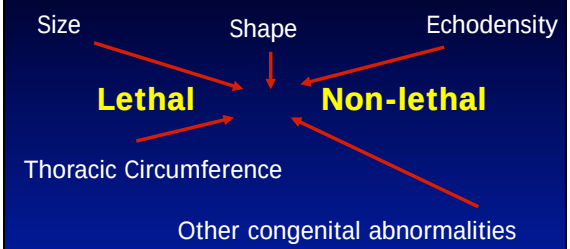
Difficulties in Ultrasound Diagnosis

- Relative rarity of the skeletal dysplasia (1.1-7.6/10,000)
- Findings on U/S may not be pathognomonic of a particular osteochondrodysplasia
- Varying degrees of expertise in U/S
- Findings can be GA dependent
- Most occur sporadically in LR pregnancies
- Overlap of radiographic and U/S findings in different disorders (EVC vs. SRPIII)
- Biologic variability

Lethal Skeletal Dysplasias

- Definition - Without heroic measures, the majority of affected individuals will die in the first few months of life.
- Death is secondary to pulmonary insufficiency or associated anomalies.
- There are almost always milder forms of the disorder.
  - Asphyxiating Thoracic Dystrophy
  - Type II collagen disorders: Achondrogenesis II, Hypochondrogenesis, Spondyloepiphyseal Dysplasia Congenita
  - Diastrophic Dysplasia Sulfate Transporter disorders: Achondrogenesis IB, Atelosteogenesis II, Diastrophic Dysplasia and recessive Multiple Epiphyseal Dysplasia

Ultrasound Imaging



Lethality and Chest Circumference

- Most critical parameter to determine in the prenatal period
- Can be done subjectively
- Can be done based on diagnosis
- Chest circumference to abdominal circumference < .6
- Heart circumference is > 50% of the chest circumference

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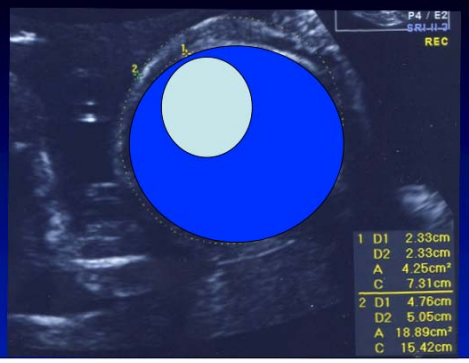
Heart Circumference vs. Chest Circumference



Ultrasound Parameters

Key Measurements
BPD, HC, FL, AC
All long bones
Foot
Scapula
Clavicle

Other Parameters
Cranium, profile, facies
Vertebrae/pelvis
2° epiphyseal centers
Hands, phalanges

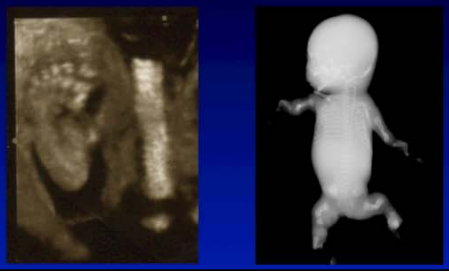
Other Findings
Cystic hygroma
Edema, hydrops
Abnl posturing

Ratios that aid in the diagnosis of disproportion

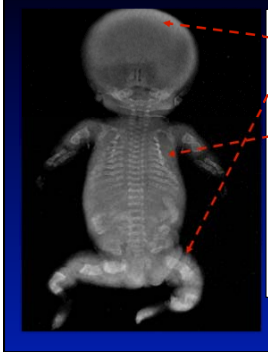
- AC to HC
- FL to AC (<0.16)
- FL to Foot Length
- FL to HC
- HC to Thoracic Circumference
- AC to Thoracic Circumference
- Rib Cage Perimeter to Thoracic Circumference

It's all about disproportion, dysmorphology,

And of course, the radiology!

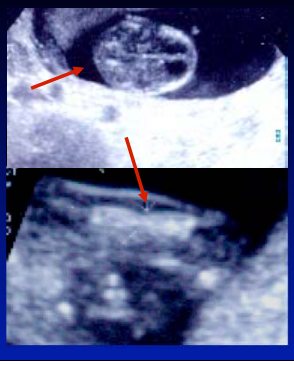


OI Type II/III



- Lethal
- Poor mineralization of the calvarium
- "Crumpled" long bones
- Platyspondyly
- Thoracic hypoplasia
- "Beaded ribs"
- Equinovarus
- Relative normal appearing hands
- Hydrops
- COL1A1, COL1A2, CRTAP,P3H1 mutations
- Recurrence risk 2-6% due to
- gonadal mosaicism or 25% due to autosomal recessive inheritance

OI type II/III



Ultrasound Findings:
Early onset of findings
Hypomineralization of calvarium
FRACTURES
BENDS
VARIABILITY

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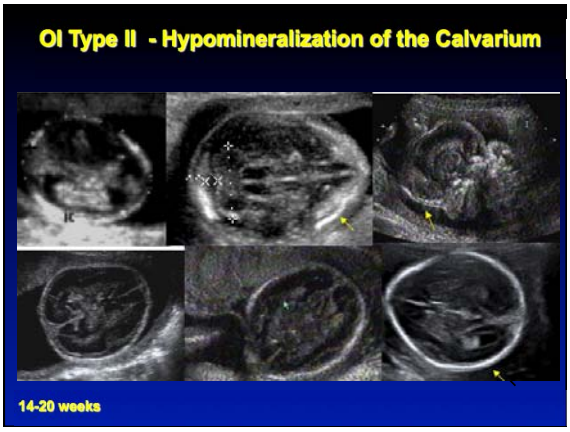
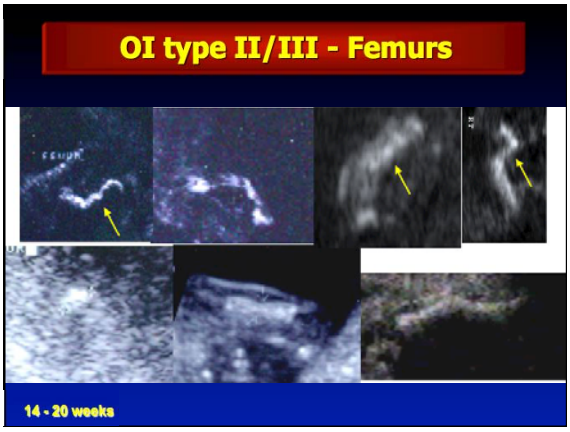
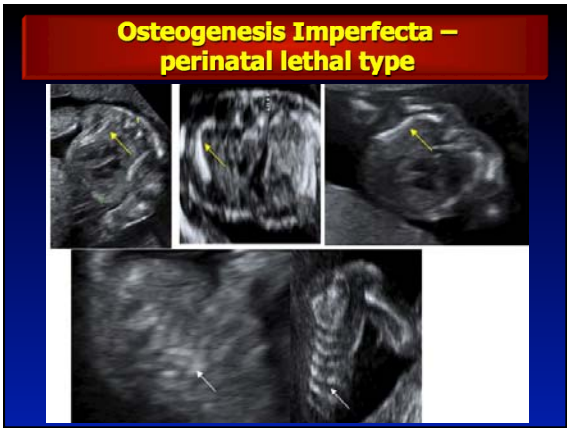
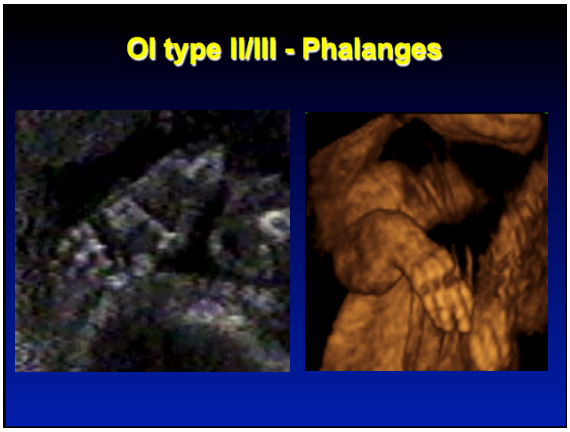
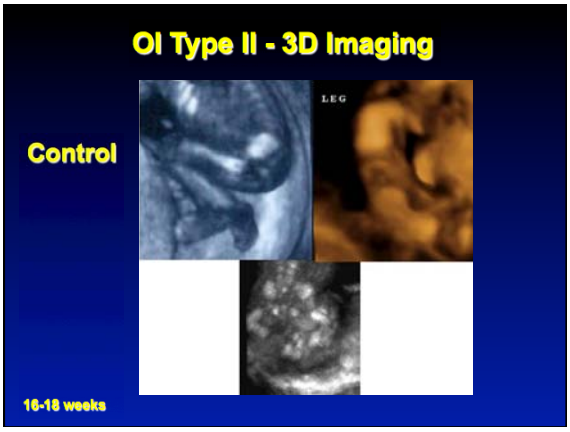
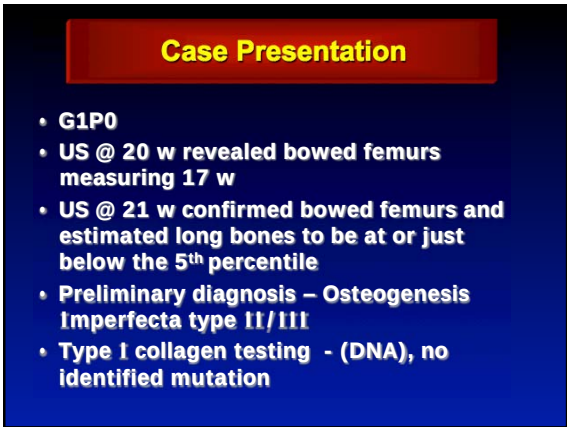

OI Type II - Hypomineralization of the Calvarium
14-20 weeks


OI type II/III - Femurs
14 - 20 weeks

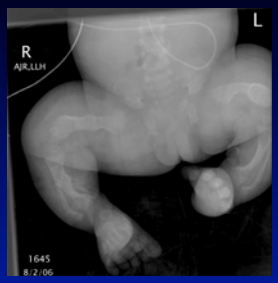

Osteogenesis Imperfecta – perinatal lethal type


OI type II/III - Phalanges


OI Type II - 3D Imaging
Control
LEG
16-18 weeks

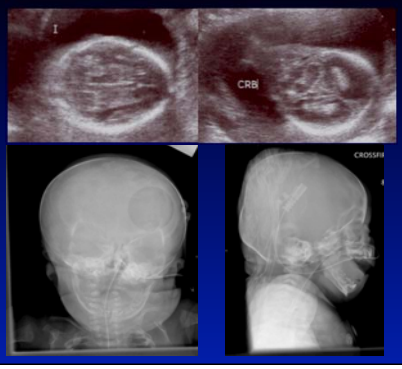

Case Presentation
G1P0
US @ 20 w revealed bowed femurs measuring 17 w
US @ 21 w confirmed bowed femurs and estimated long bones to be at or just below the 5th percentile
Preliminary diagnosis – Osteogenesis Imperfecta type II/III
Type I collagen testing - (DNA), no identified mutation

Lower extremities

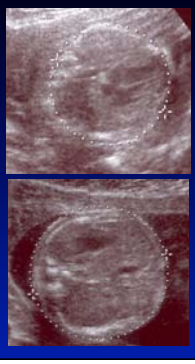


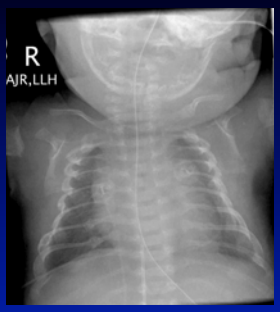
Images courtesy of Akron Children's Hospital: Haynes Robinson, MD

Calvarium

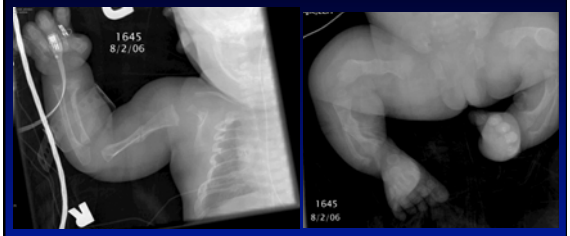


Chest circumference/abdominal circumference - .89





Newborn A/P Upper and Lower Extremities



Take home message!

- Ultrasound and postnatal radiographs consistent with Osteogenesis Imperfecta type II/III
- Molecular analysis showed that this fetus has a recessive form of OI due to mutations in *CRTAP*
- Familial recurrence risk now 25%, not 2% as previously thought based on germline mosaicism

Differential Diagnosis of Hypomineralization Disorders

Osteogenesis Imperfecta
Demineralized calvarium
Fractures
Platyspondyly
Normal appearing hands

Cleidocranial Dysplasia
Autosomal Dominant
Demineralization of the calvarium
Clavicle hypoplastic
Relatively normal appearing long bones

Hypophosphotasia
Autosomal Recessive
Fractures
"Chromosome-appearing" bones
Very poor mineralization of the hands

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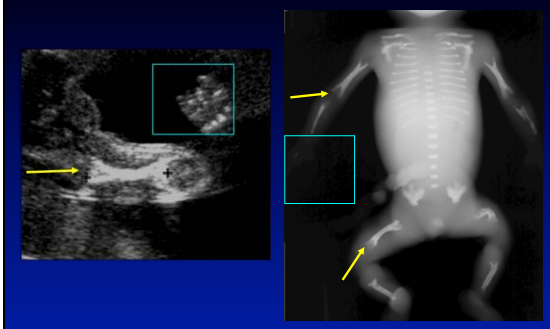
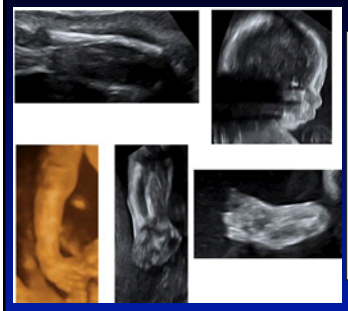
Hypophosphotasia



Hypophosphotasia

OI vs. Campomelic Dysplasia

Campomelic Dysplasia



- AD
- Heterozygosity SOX9
- Recurrence - germline mosaicism
- Micrognathia
- Micro/macrocephaly
- Equinovarus
- Brachydactyly
- Multiple organ system involvement
- Not uniformally lethal

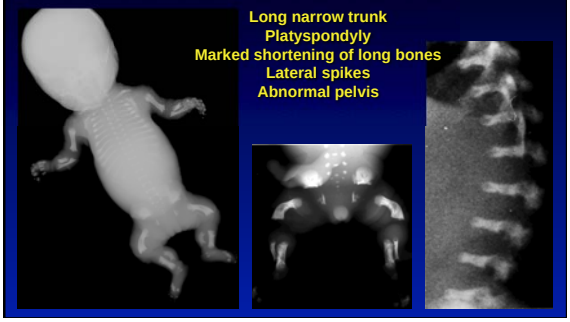
Thanatophoric Dysplasia

- Lethal
- Disproportion
- Severe micromelia
- Relative Macrocephaly
- Craniosynostosis
- Frontal bossing
- Midface hypoplasia
- Narrow chest
- Straight versus versus
- curved long bones
- No epiphyseal delay
- Rare other organ
- system anomalies
- FGFR3 mutation
- Paternal in origin

Thanatophoric Dysplasia

RADIOLOGIC FINDINGS

Long narrow trunk
Platyspondyly
Marked shortening of long bones
Lateral spikes
Abnormal pelvis



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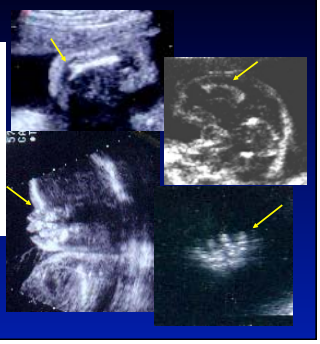
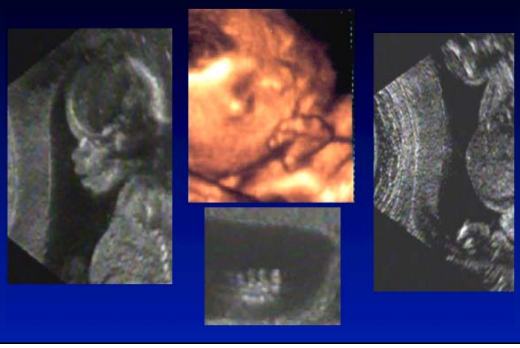
Thanatophoric Dysplasia

U/S Findings:
Cloverleaf skull
Curved or straight femurs
Generalized micromelia
Small narrow thorax
Platyspondyly
Very small hands and feet
Polyhydramnios



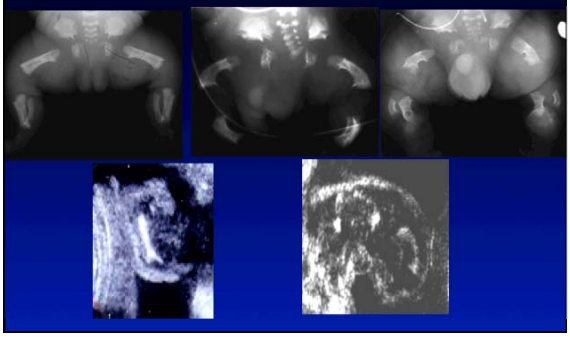
Thanatophoric Dysplasia



Thanatophoric Dysplasia

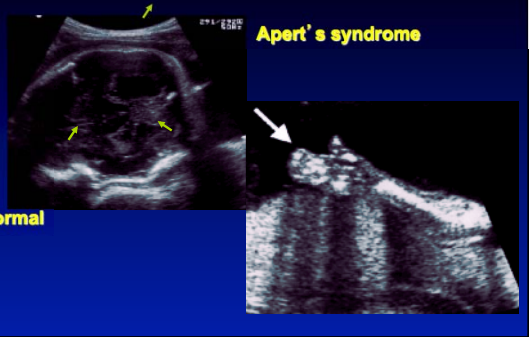
24 weeks

Thanatophoric Dysplasia



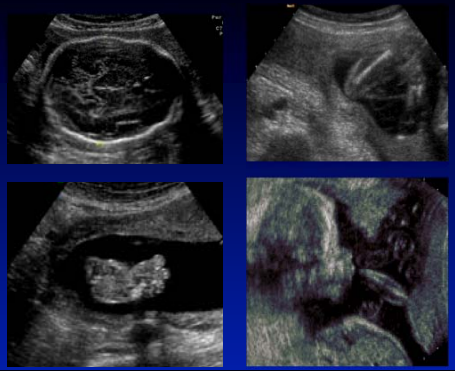
FGFR2 disorders

Apert's syndrome



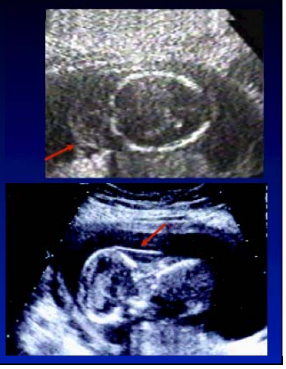
Normal

Craniosynostosis

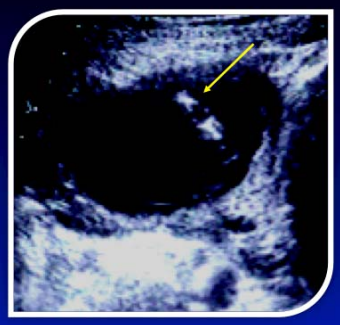


Achondrogenesis II

- Lethal
- Defects in COL2A1
- Cystic hygroma
- Hydrops
- Mid-face Hypoplasia
- Micrognathia
- Severe Micromelia
- (all segments)
- Equinovarus
- Absent mineralization of the vertebral bodies
- Metaphyseal “spikes”



Achondrogenesis II -14 weeks

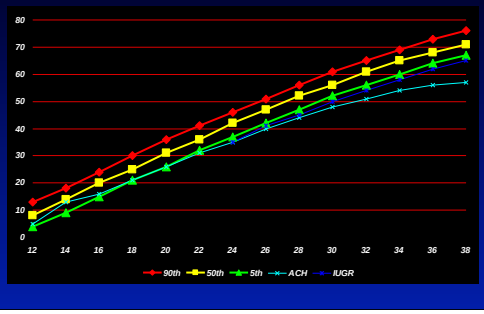


Achondrogenesis II

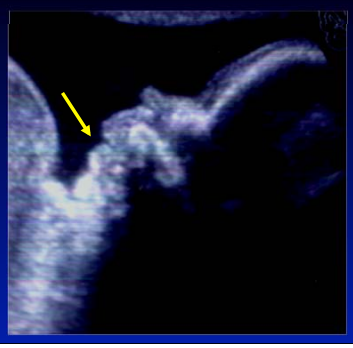
Achondroplasia

- Autosomal Dominant
- 80% cases arise *de novo*
- Exclusively on the paternal allele
- Frequently not diagnosed until the 3rd trimester
- Frequent complications: orthopedic, ENT, and neurologic

Fetal femur growth curves in Achondroplasia



Achondroplasia



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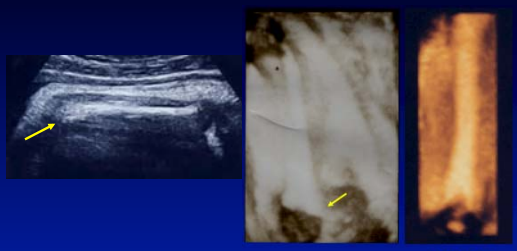
Achondroplasia – Distal Femurs

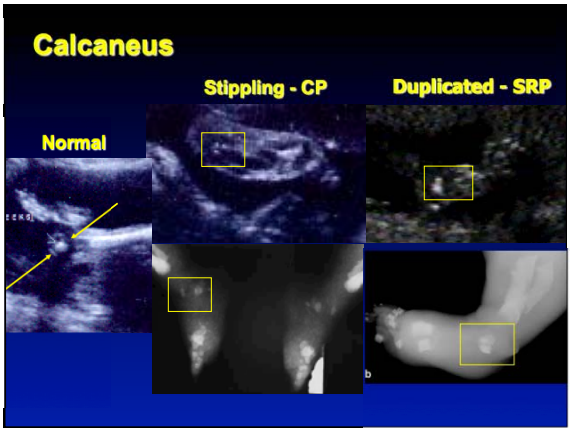


Achondroplasia - facies

Abnormal Calcaneus

- Phenytoin
- Warfarin
- Alcohol
- Rubella
- Trisomy 21;Trisomy 18;Mosaic risomy 9
- Mucolipidosis, type II
- Gangliosidosis I
- Smith-Lemli Opitz syndrome
- Chondrodysplasia punctata, all types
- Thanatophoric Dysplasia
- Short-rib polydactyly syndromes
- Chondrectodermal dysplasia
- Asphyxiating thoracic dysplasia
- Larsen syndrome
- Atelosteogenesis types I and III
- Roberts-SC Phocomelia

Calcaneus



Calcaneus - Robert's SC Phocomelia

Fetal Hands

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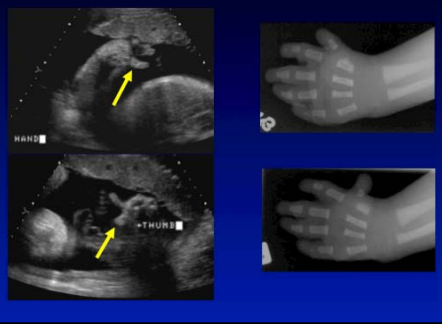
Fetal Hands

Robert SC Phocomelia | OI Type II

Roberts SC Phocomelia

Courtesy of Maureen Bocian

Diastrophic Dysplasia



Fetal Hands

Conclusions

- Ultrasound can be an effective method for the diagnosis in the prenatal-onset skeletal dysplasias.
- Improved accuracy in diagnosis can be achieved by application of the established radiographic findings in the specific disorders.
- Ultrasound parameters need to be expanded to look for a constellation of findings for more precise diagnosis.
- 3D ultrasound is an effective modality for visualization of the dysmorphic findings in the skeletal dysplasias.
- Final diagnosis of a prenatal-onset skeletal dysplasia should be based on radiology and histomorphology.
- Some diagnoses cannot be accurately made in the prenatal period.

Management of a fetus with a skeletal dysplasia

- Determine lethality
- Educate obstetrical team
- Delivery: previable versus at term
- Role of adoption
- Mode of delivery
- Does the newborn need higher level of care
- Post-delivery diagnosis
- Post-delivery management
- Parental support groups (LPA, grief-loss groups)

Management of a fetus with a skeletal dysplasia

- Delivery: pre-viable versus at term
- Will awaiting DNA diagnosis affect parental decisions?
- Mode of delivery/Availability of trained personal
- Importance of counseling regarding NEED for postnatal diagnosis by autopsy (x-rays, histomorphology of relevant tissues, DNA diagnosis as appropriate)
- Coordination of pediatric personal regarding resuscitation including pre-delivery agreement on management plan
- Mode of delivery at term: issues regarding breech presentation, relative macrocephaly and intrapartum fetal distress
- Counseling re: lethality is not frequently, not immediate

Management of the fetus with a non-lethal skeletal dysplasia

- Is the diagnosis definitive?
- What is the differential diagnosis?
- Within the differential diagnosis is the natural history associated with a small percentage of lethality or respiratory compromise?
- Is there a possibility the child will need a higher level of care?
- Osteogenesis imperfecta - no evidence of increased fractures with vaginal versus C/S, do not recommend instrumental delivery
- High incidence of C/S due to relative macrocephaly
- Cord blood for a source of DNA
- Full genetic survey

Management of maternal patients with short stature

OBSTETRICS AND OBSTETRICAL ANESTHESIA ISSUES IN WOMEN WITH DWARFISM

JE Hoover-Fong, G Oswald, D Miller, J Leadroot, H Barnes, J Rossiter, D Penning, I Berkowitz, D Krakow

- ✓ Advocacy for reproductive rights of individuals with skeletal dysplasias (short stature)
- ✓ Education of the obstetrical and anesthesia communities
- ✓ What are the risks to the mother, the fetus? Are they real?

Management of maternal patient with short stature

- What is the maternal diagnosis?
- What is the mode of inheritance?
- Non-assortive mating in the short stature community
- What is the diagnosis in the partner?
- Has mutational analysis been performed on both parents prior to pregnancy? Is it even possible?
- What are the couple's feeling regarding prenatal diagnosis and termination if necessary?
- Should a short stature individual take the risk of pregnancy for a lethal disorder?
- Who is going to care for the patient?

Management of maternal patient with short stature

- Maternal diagnosis
  - Long trunk versus short trunk
    - Achondroplasia, pseudoachondroplasia
    - SEDC, OI type III, Cartilage Hair Hypoplasia, Diastrophic Dysplasia
- Normal pregnancy associated with increased pulmonary dead space and increased heart rate.
- The increased incidence of pulmonary compromise in short trunk dwarfism. Increasing fundal pressure on diaphragm!

Management of maternal patient with short stature

- Non-assortive mating
  - Compound heterozygosity
    - Achondroplasia/Achondroplasia
    - Achondroplasia/SEDC
    - SEDC/pseudoachondroplasia
    - Achondroplasia/acrolaryngeal

Management of maternal patient with short stature

- Non-assortive mating
  - Preimplantation diagnosis for determining affected embryo status
  - Is PGD effective in determing compound heterozygosity? Which diagnosis would the couple prefer?
  - Chorionic villous sampling in short stature women can be challenging due to high incidence of retroverted or anteverted uteri.
    - May need abdominal CVS or 15-20 wk amniocentesis

Management of maternal patient with short stature

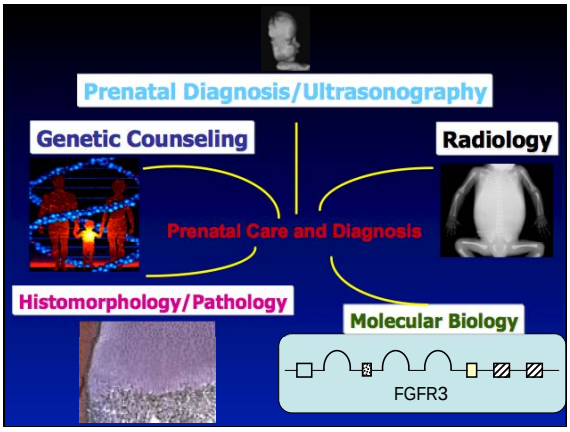
- Length of gestation
  - 8% of the general population delivers preterm
    - 16% if 1 previous PTD
    - 32% if 2 previous PTD,
    - >50% with 3 or more PTD
  - Most LP decrease physical activity around 24-28 weeks
  - Delivery at 34-36 weeks after documentation of fetal lung maturity
  - Vast majority of LP are delivered by C/S

Management of maternal patient with short stature

- Management of Delivery
  - Anesthesia
    - Maternal Diagnosis
    - Epidural versus spinal versus general anesthesia; adult or weight doses???
    - For disorders associated with spinal stenosis or spinal abnormalities, pre-pregnancy MRIs can be helpful
    - For disorders with risk for odontoid hypoplasia, general anesthesia with intubation should be done with care
    - Pre-delivery anesthesia consult with anesthesia

Pregnancy in Short Stature Women

- Most short stature women tolerate pregnancy well!
- Major concerns include proper care, compound heterozygosity, preterm labor, shortness of breath, back pain (achondroplasia),mode of delivery and anesthesia
- Osteogenesis Imperfecta: bisphosphonates are contraindicated in pregnancy, what are the long term effects are bone in pregnancy and lactation?



International Skeletal Dysplasia Registry

David L. Rimoin
Daniel H. Cohn
William R. Wilcox
Ralph S. Lachman
Deborah Krakow
Yasemin Alanay

MaryAnn Priore, coordinator
Tara Funari
Arleen Hernandez
Sue Lief
Betty Melkkian

Medical Genetics Institute
Cedars-Sinai Medical Center

Department of Human Genetics
UCLA School of Medicine

Supported by NIH HD22657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Ultrasound of the Fetal Genitourinary Tract

Jude P. Crino, M.D.
Assistant Professor, Maternal-Fetal Medicine
Director, Perinatal Ultrasound
Department of Gynecology and Obstetrics
Johns Hopkins University School of Medicine
Baltimore, MD

Contribution Not Received in Time for Inclusion.

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The 11 - 14 Weeks Obstetrical Scan: Current Concepts and Future Directions

Jude P. Crino, M.D.
Assistant Professor, Maternal-Fetal Medicine
Director, Perinatal Ultrasound
Department of Gynecology and Obstetrics
Johns Hopkins University School of Medicine
Baltimore, MD

Contribution Not Received in Time for Inclusion.

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Fetal Neurosonography in Early Gestation: What Can Be Diagnosed Today?

Elena Sinkovskaya, M.D., Ph.D.
Assistant Professor
Director of Research
Division of Maternal-Fetal Medicine
Department of Obstetrics & Gynecology
Eastern Virginia Medical School
Norfolk, VA

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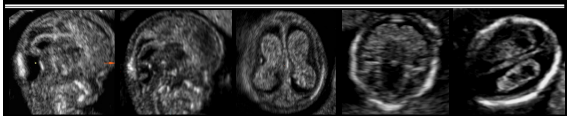
EVMS Eastern Virginia Medical School

Advances in Ultrasound in Obstetrics and Gynecology
October 14-15, Washington DC

FETAL NEUROSONOGRAPHY IN EARLY GESTATION: what can be diagnosed today?

Elena Sinkovskaya, M.D., PhD



What is Fetal Neurosonography?

Ultrasound Obstet Gynecol 2007; **29**: 109–116
Published online in Wiley InterScience (www.interscience.wiley.com). **DOI:** 10.1002/uog.3909

THE INTERNATIONAL SOCIETY OF ULTRASOUND in Obstetrics & Gynecology

GUIDELINES

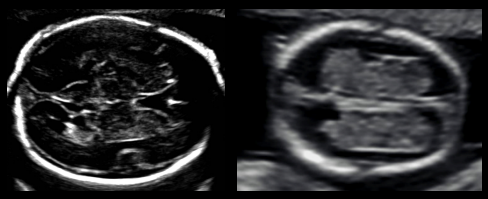
Sonographic examination of the fetal central nervous system: guidelines for performing the 'basic examination' and the 'fetal neurosonogram'

What is Fetal Neurosonography?

Basic Examination	Fetal Neurosonogram
1. Screening (performed on everybody)	1. Performed by indications
2. Usually transabdominal ultrasound	2. Transabdominal and transvaginal ultrasound
3. Three axial planes	3. Multiple axial, coronal and sagittal planes
	4. Extra training and expertise

Basic Examination

transventricular plane

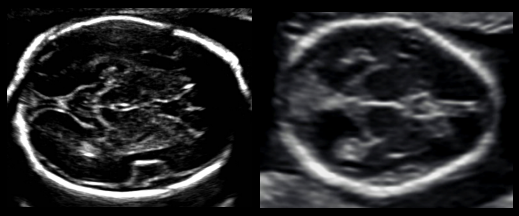


23+6 weeks

13+6 weeks

Basic Examination

transthalamic plane

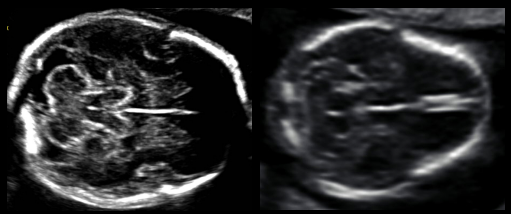


23+6 weeks

13+6 weeks

Basic Examination

transcerebellar plane



23+6 weeks

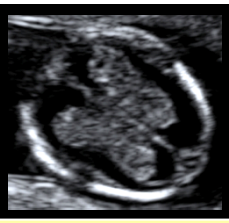
13+6 weeks

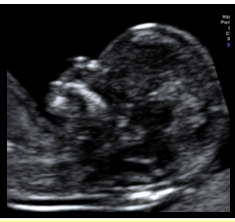
First Trimester Fetal Imaging:

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





Transverse View	Midsagittal View
Presence of: – skull bones – 2 hemispheres – 2 symmetrical ventricles – Homogenous appearance of choroid plexus	Assessment of: – the brainstem – 4th ventricle – IT? – cisterna magna

Intracranial Translucency (IT)

Ultrasound Obstet Gynecol 2010; **35**: 133–138
Published online in Wiley InterScience (www.interscience.wiley.com). **DOI:** 10.1002/uog.7552

Editorial

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

R. CHAOUI†*and K. H. NICOLAIDES‡

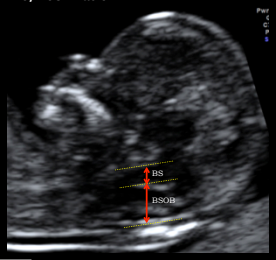
†*Prenatal Diagnosis and Human Genetics, Berlin, Germany and ‡Harris Birthright Research Centre for Fetal Medicine, King's College Hospital and Department of Fetal Medicine, University College, London, UK* **Correspondence (e-mail: chaoui@fetdiagnostik.de)*

Nuchal translucency (NT)

- Fetus in mid-sagittal plane
- Fetal head& neck region occupies majority of image
- Fetal head in neutral position
- Fetus observed away from amnion
- Margins of NT edges clear
- (+) calipers used
- Horizontal crossbars placed correctly
- Calipers placed ⊥ to long axis of fetus
- Measurement at widest NT space

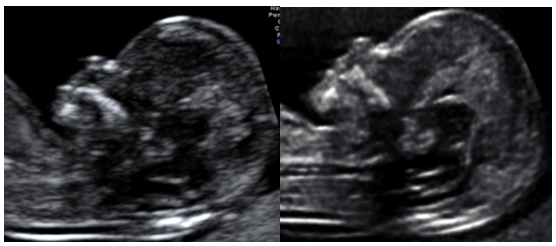
Measurements of the Posterior Brain

- brain stem diameter (BS)
- brain stem to occipital bone diameter (BSOB)
- BS/BSOB ratio



* Prenat Diagn 2011;31:103-106

Limitations of the IT interpretation



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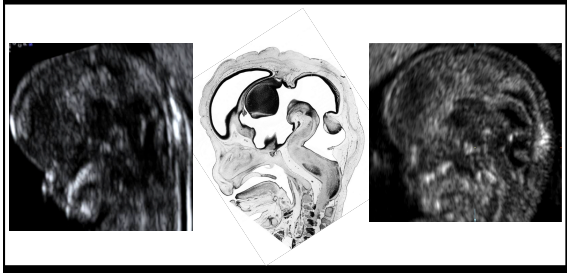
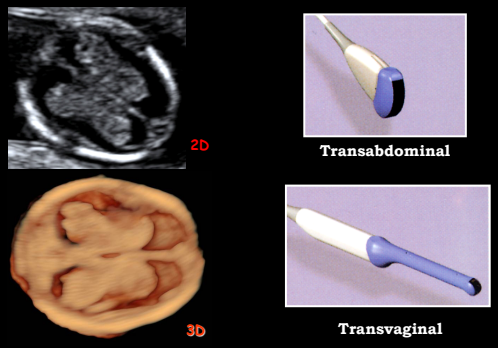
Transabdominal vs. Transvaginal

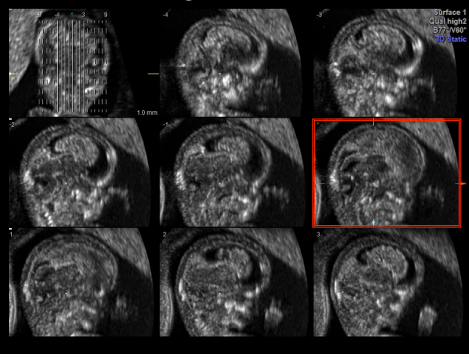


Fetal Neurosonogram: *instrumentation*

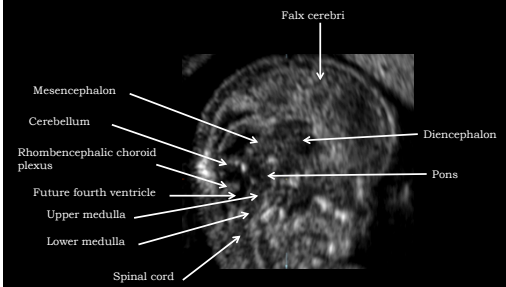


Fetal Neurosonogram: *how does it work?*

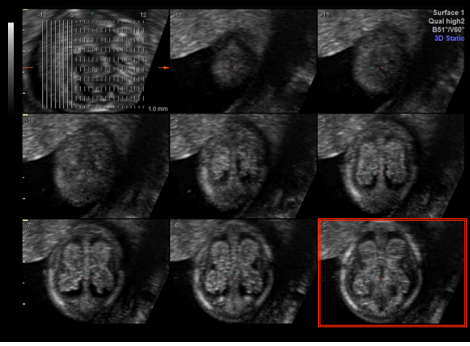
Fetal Neurosonogram: *Sagittal views*



Midsagittal view



Fetal Neurosonogram: *Axial views*



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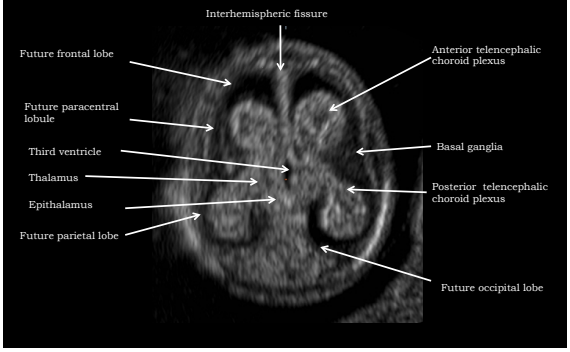
Transthalamic view



First Trimester Fetal Neuro Imaging:

Several studies have suggested that 1st trimester CNS screening for ..

- **acrania**
- **excencephaly/anencephaly**
- **encephalocele**
- **Holoprosencephy**
- **hydrocephaly**

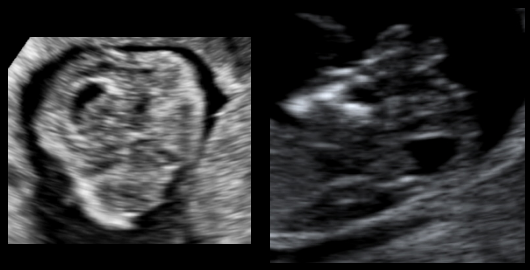
...can be just as effective in diagnosing these conditions as 2nd trimester screening

Recent studies have also demonstrated that performance of detailed evaluation of fetal brain between 11^{+0} -13^{+6} weeks may allow detection of...

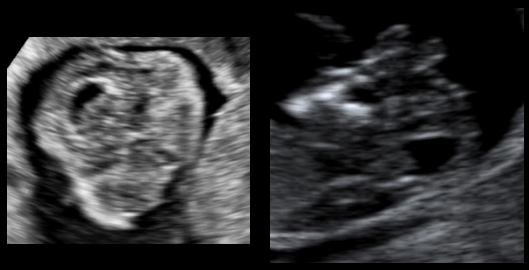
- **Spina bifida**
- **Dandy-Walker malformation**
- **Agenesis of corpus callosum**

Blaas H-GK, Eik-Nes SH. Sonoembryology and early prenatal diagnosis of neural anomalies. Prenatal Diagnosis 2009; 29:312-325.
Vora E, Basturzo B. Sciarrone A. et al. Early diagnosis of Fetal brain anomalies. Ultrasound Rev Obstet Gynecol. 2003; 3: 74-80.

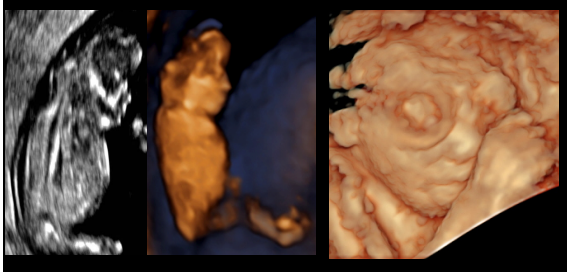
What's wrong?



Anencephaly/Acrania

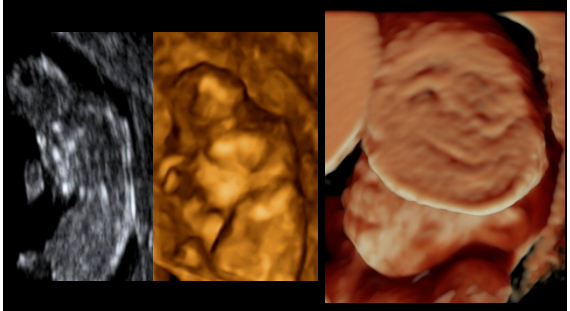


Anencephaly



12 weeks 24 weeks

Acrania



11 weeks 16 weeks

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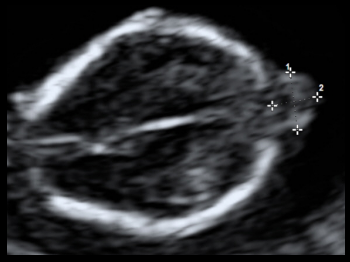
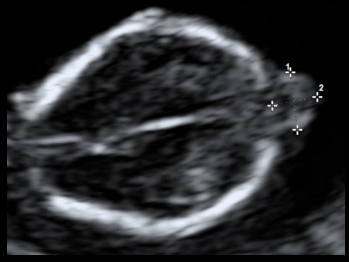
What's wrong?



Encephalocele



Ultrasound Obstet Gynecol 2015; **46**: 29–33
Published online 4 June 2015 in Wiley Online Library (wileyonlinelibrary.com). **DOI:** 10.1002/uog.14661

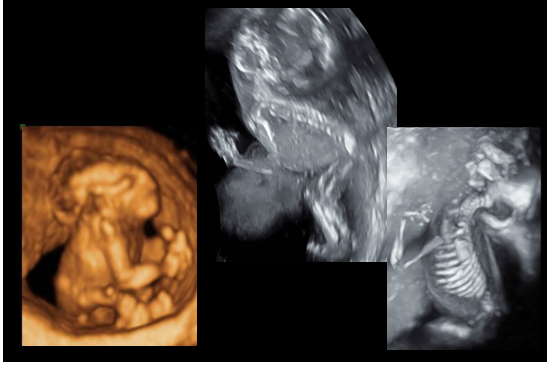
Sonographic spectrum of first-trimester fetal cephalocele: review of 35 cases

W. SEPULVEDA*, A. E. WONG*, E. ANDREEVA†, N. ODEGOVA†, P. MARTINEZ-TEN‡ and S. MEAGHER§

**Fetalmed–Maternal-Fetal Diagnostic Center, Las Condes, Santiago, Chile; †Medical-Genetics Department, Moscow Regions Research Institute of Obstetrics and Gynecology, Moscow, Russia; ‡Delta–Ultrasound Diagnostic Center for Obstetrics and Gynecology, Madrid, Spain; §Monash Ultrasound for Women, Melbourne, Australia*

Location of the lesion	Incidence, %
Occipital	77%
Parietal	9%
Frontal	9%
Multiple	6%

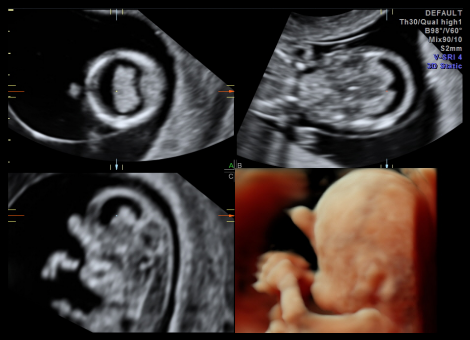
Iniencephaly



What's wrong?

Alobar Holoprosencephaly

Alobar Holoprosencephaly

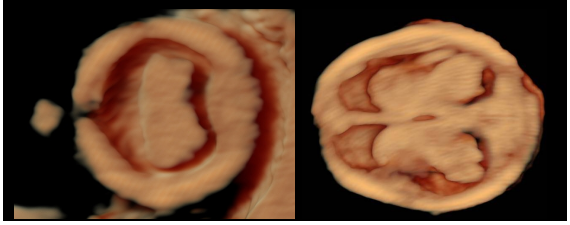


First trimester screening for holoprosencephaly with choroid plexus morphology ('butterfly' sign) and biparietal diameter

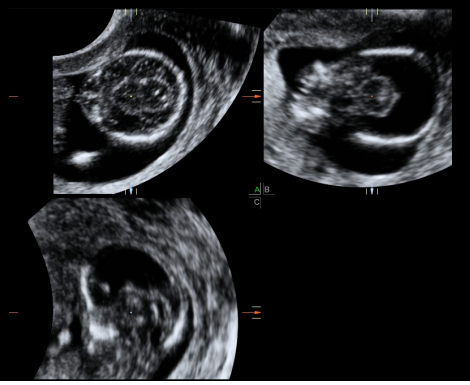
Waldo Sepulveda* and Amy E. Wong

Fetal Medicine Center, Department of Obstetrics and Gynecology, Clinica Las Condes, Santiago, Chile
*Correspondence to: Waldo Sepulveda. E-mail: fetalmed@yahoo.com

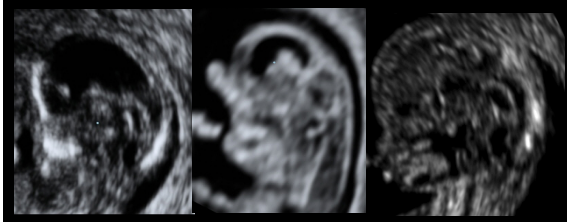
Conclusions The 'butterfly' sign appears to be a highly sensitive marker for HPE in the first trimester. On the other hand, BPD measurements had a lower sensitivity, implying that microcephaly is not a prominent first-trimester feature in these cases. Incorporation of the 'butterfly' sign into the first trimester anatomy scan is simple and can facilitate the identification of the vast majority of fetuses with HPE in the first trimester. © 2013 John Wiley & Sons, Ltd.



Alobar Holoprosencephaly & Hydrocephaly



Alobar Holoprosencephaly & Hydrocephaly



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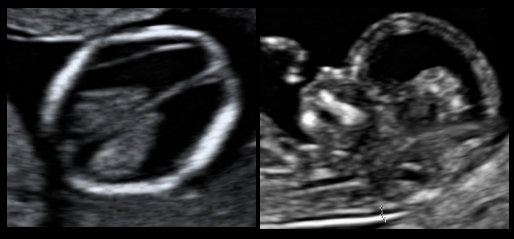
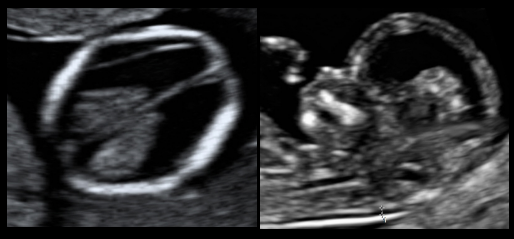


Ventriculomegaly

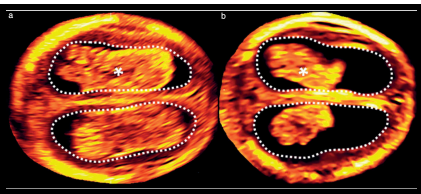


Ultrasound Obstet Gynecol 2012; **40**: 282–287
Published online 30 July 2012 in Wiley Online Library (wileyonlinelibrary.com). DOI: 10.1002/uog.11197

Lateral ventricles in fetuses with aneuploidies at 11–13 weeks' gestation

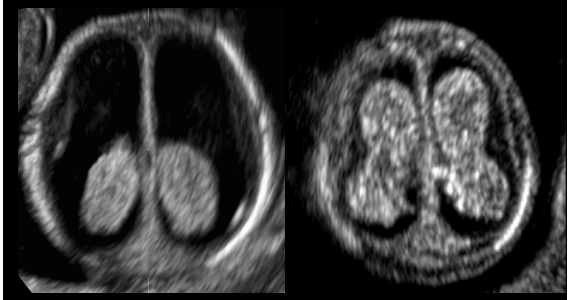
T. LOUREIRO*†, F. USHAKOV†, N. MAIZ‡, N. MONTENEGRO* and K. H. NICOLAIDES†§

**Department of Obstetrics and Gynecology, S. João Hospital, Medical School, University of Porto, Porto, Portugal; †Fetal Medicine Unit, University College Hospital, London, UK; ‡Fetal Medicine Unit, Obstetrics and Gynecology Department, Cruces University Hospital, Biscay, Spain; §Harris Birthright Research Centre for Fetal Medicine, King's College Hospital, London, UK*



The finding of contracted choroid plexus should raise suspicion of ventricular dilation

Ventriculomegaly



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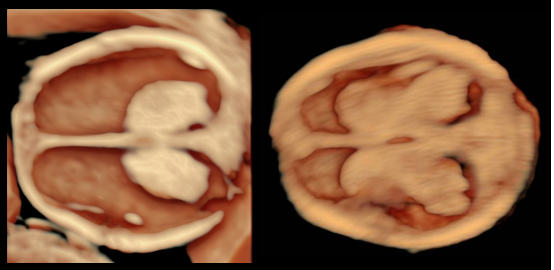
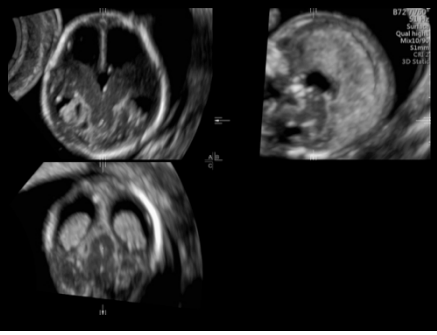
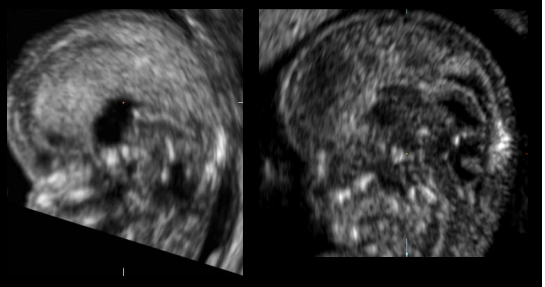


Ventriculomegaly



Ventriculomegaly
What's wrong?



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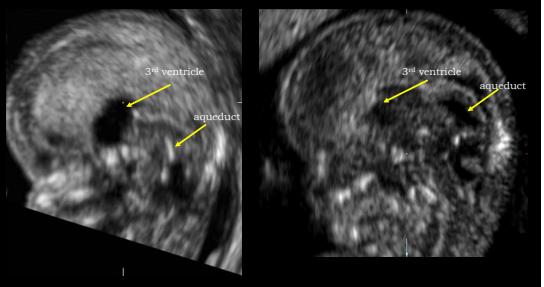

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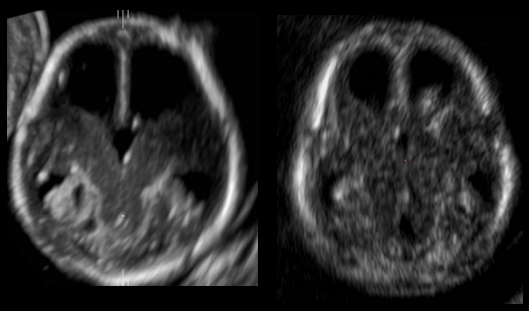
Ventriculomegaly

What's wrong?



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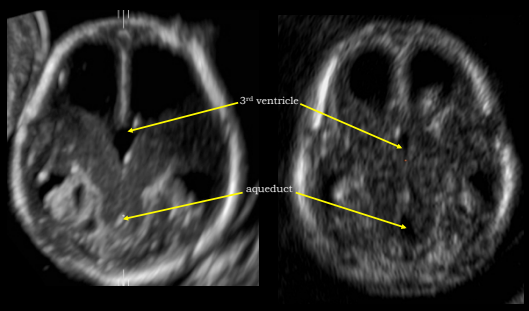
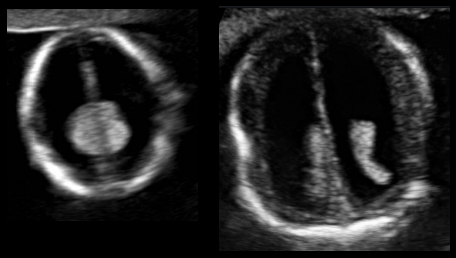


Ventriculomegaly

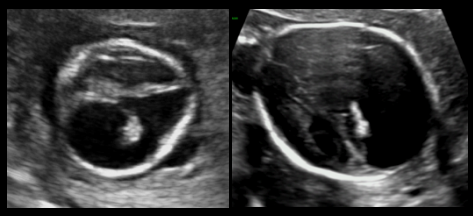
bilateral



12 weeks 19 weeks

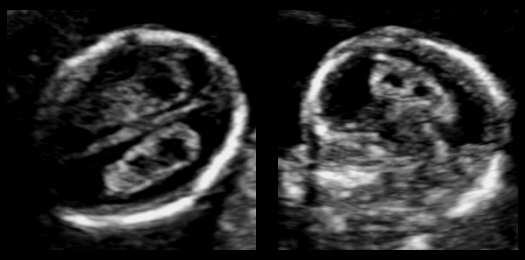
Ventriculomegaly

unilateral



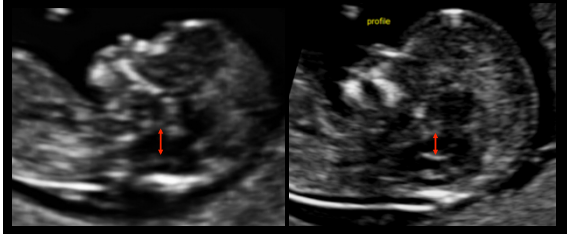
13 weeks 22 weeks

Choroid plexus cysts



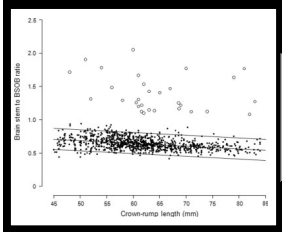
Normal or Abnormal?

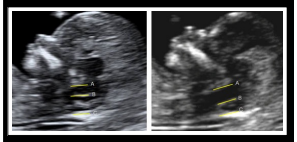
Normal or Abnormal?



Spina Bifida Normal

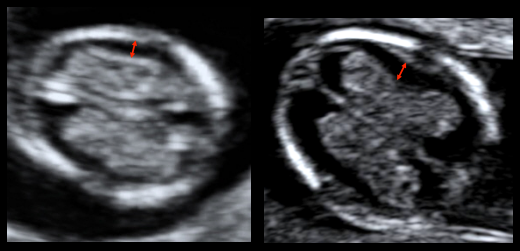
Screening for spina bifida





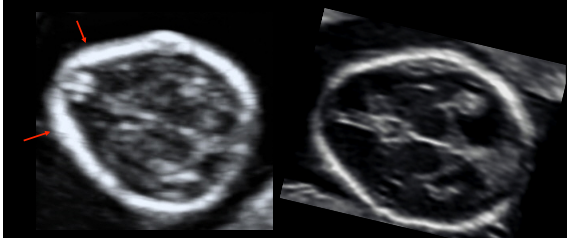
Lachmann et al. *Prenat Diagn* 2011; **31**: 103–106.

Normal or Abnormal?



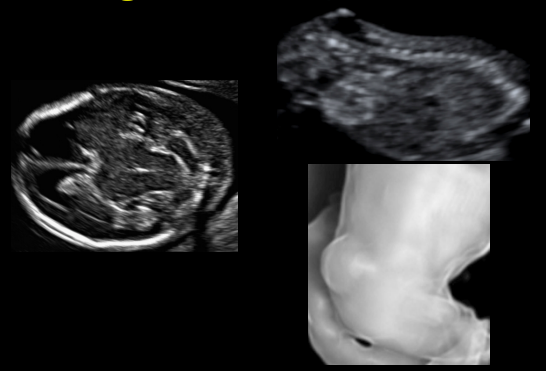
Spina Bifida Normal

Normal or Abnormal?



Spina Bifida Normal

Diagnosis of spina bifida



Normal or Abnormal?

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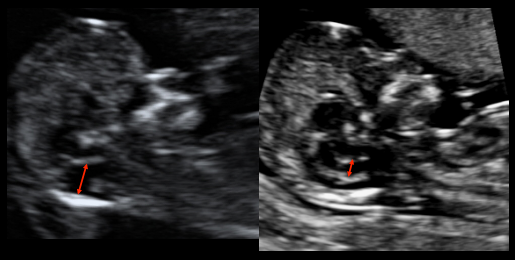
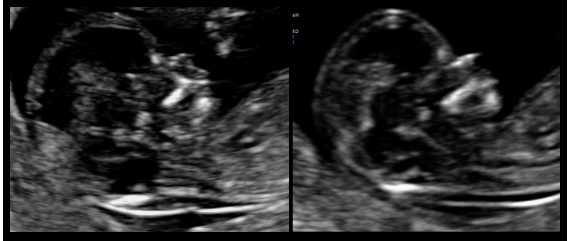
Normal or Abnormal?



Dandy-Walker Malformation

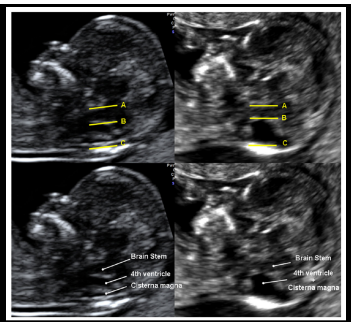
Normal

Normal or Abnormal?

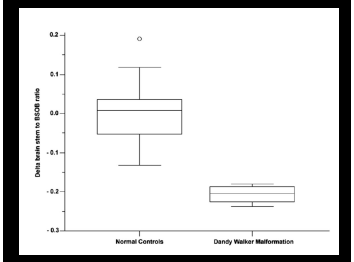


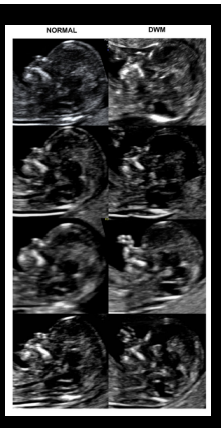
Posterior brain in fetuses with Dandy–Walker malformation with complete agenesis of the cerebellar vermis at 11–13 weeks: a pilot study

Robert Lachmann[1]*,[†], Elena Sinkovskaya[2†] and Alfred Abuhamad[2]

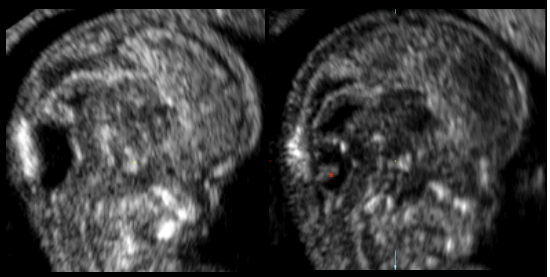


Screening for DWM

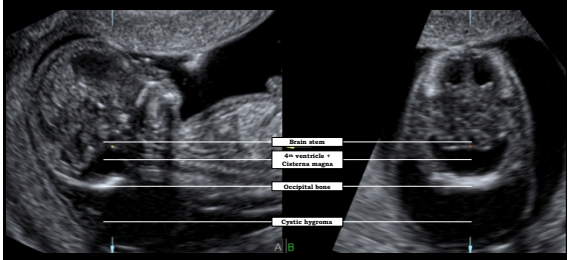




Diagnosis of DWM



Diagnosis of DWM



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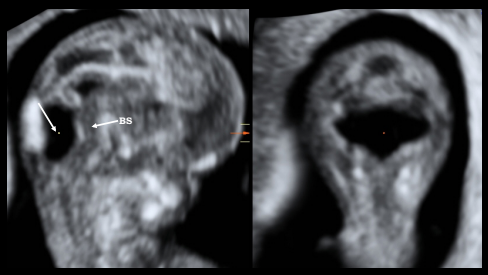
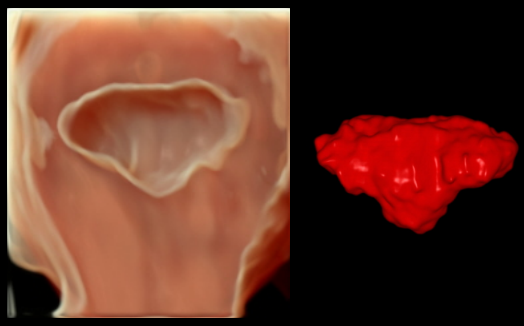
Diagnosis of DWM

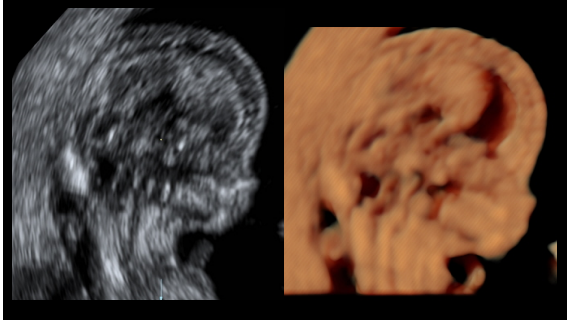


Diagnosis of DWM



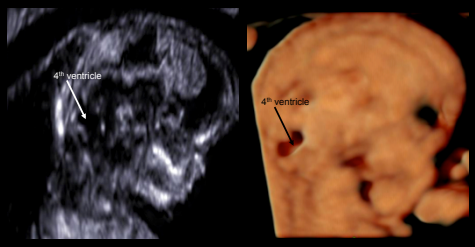
Dandy-Walker Variant

12+3 weeks gestation



Blake's pouch cyst

13+4 weeks gestation



To take home....

1. A screening technique using both transverse and midsagittal view of the fetal head allows for early detection of major fetal CNS anomalies
2. In experienced hands detailed fetal neurosonography can be performed in early gestation using combined TA&TV approach in patients with high-risk for fetal CNS abnormalities

EVMS

First Look

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Notes

The Approach to Fetal Cardiac Imaging in Early Gestation

Alfred Abuhamad, M.D.
Professor and Chairman
Department of Obstetrics and Gynecology
Vice Dean for Clinical Affairs
Eastern Virginia Medical School
Norfolk, VA

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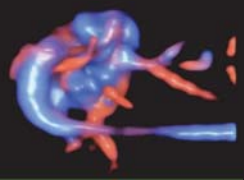
Advanced Evaluation of the Fetal Heart



EVMS Fetal Cardiovascular Center

Alfred Abuhamad, M.D.
Eastern Virginia Medical School

The Top 5 Critical Anatomic Regions in Fetal Cardiac Imaging

Top 5 Critical Anatomic Regions

1. Normal Left Atrium
2. Normal Left Ventricular Outflow
3. Normal Semilunar Valves
4. Normal 3VT View
5. Normal Cardiac Axis in Early Gestation

3

Top Critical Anatomic Regions

1. Normal Left Atrium

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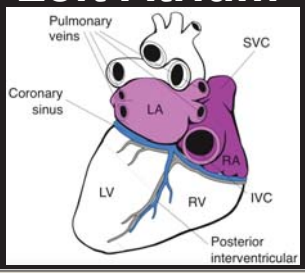
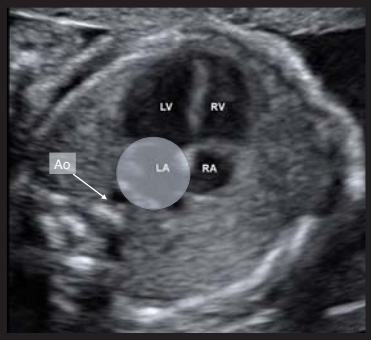
Left Atrium



Posteriorly located, over the spine
Anterior and posterior portion is smooth
Receives four pulmonary veins
Left atrial appendage is narrow, fingerlike with coarse walls

Left Atrium



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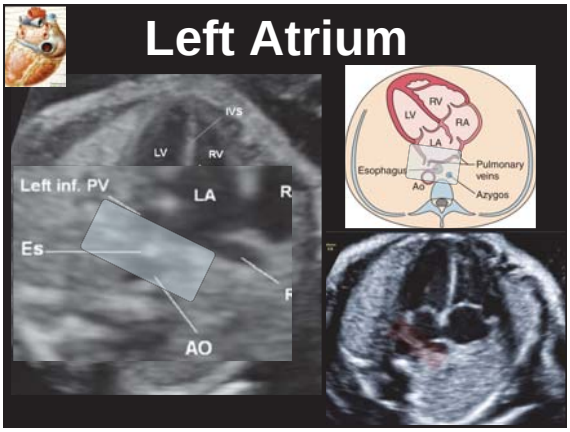
Pulmonary Veins

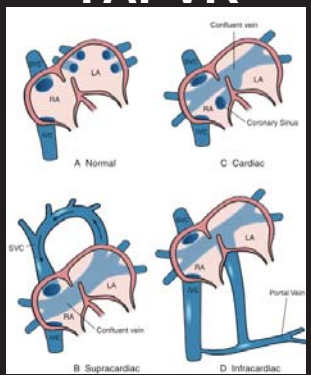
Pulmonary Veins

Left Atrium

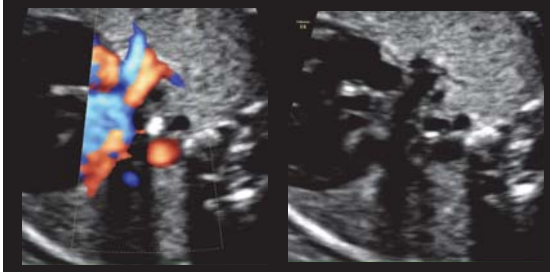


Left Atrium

TAPVR



Left Atrium



Closed Esophagus

Open Esophagus

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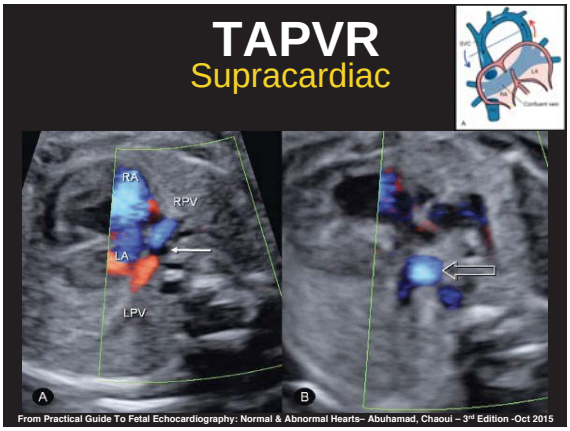
TAPVR
Supracardiac



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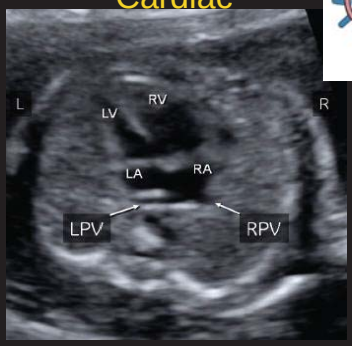
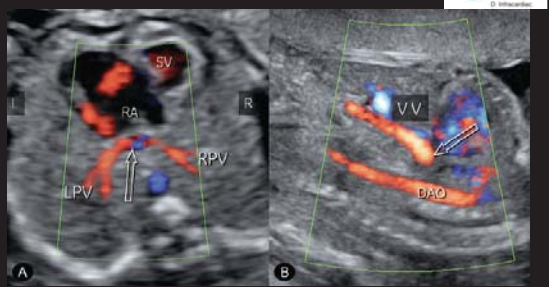
TAPVR
Cardiac



TAPVR
Infracardiac



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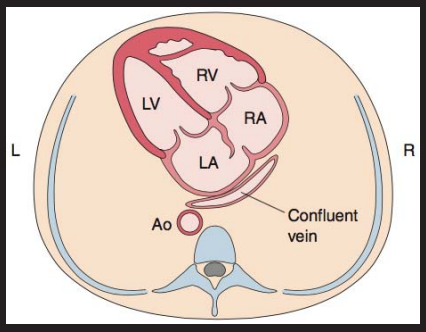
Left Atrium



TAPVR

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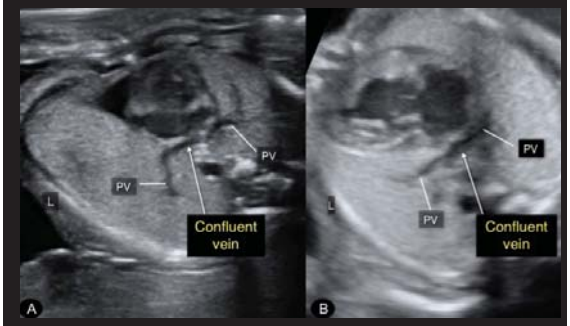
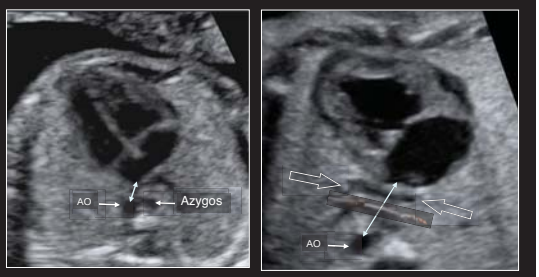
Left Atrium



TAPVR

Left Atrium



Normal TAPVR

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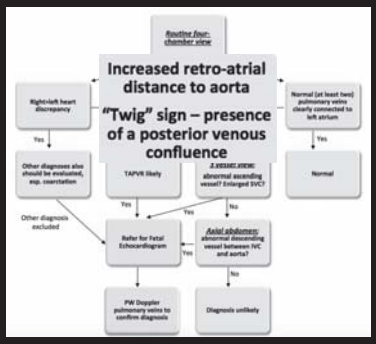
TAPVR



J Ultrasound Med 2014; 33:1193–1207

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TAPVR

TABLE 31.1 Prenatal Ultrasound Findings in Total Anomalous Pulmonary Venous Connection

- Venous confluence chamber behind the left atrium
- No direct connection between pulmonary veins and left atrium
- Enlarged right atrium and right ventricle on four-chamber view
- Bulging of interatrial septum into left atrium
- Enlarged pulmonary artery in the three-vessel-trachea view
- Vertical vein as a fourth vessel in the three-vessel-trachea view (type I)

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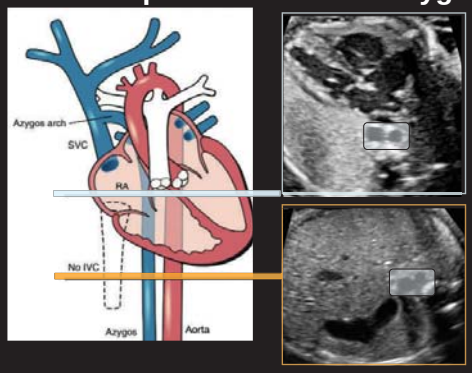
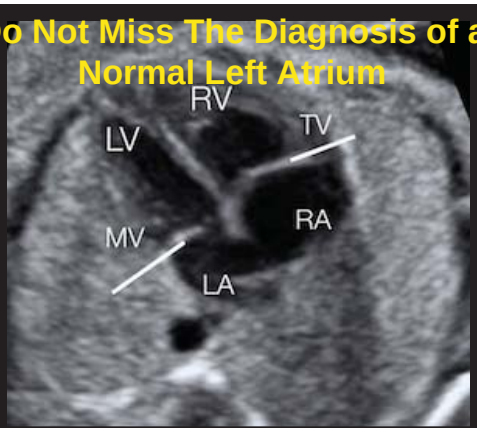
Interrupted IVC- Dilated Azygos



Do Not Miss The Diagnosis of a Normal Left Atrium



Top 5 Critical Anatomic Regions

1. Normal Left Atrium
2. Normal Left Ventricular Outflow

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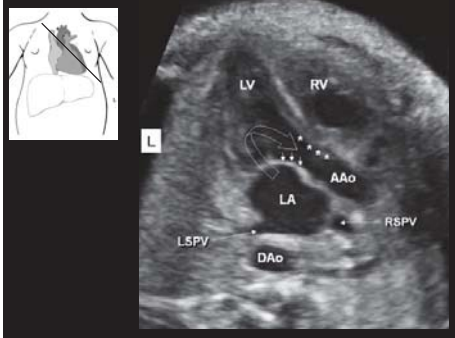
Left Ventricular Outflow



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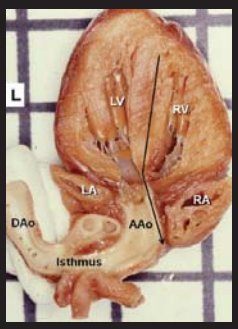
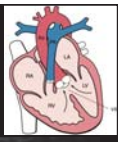
Left Ventricular Outflow

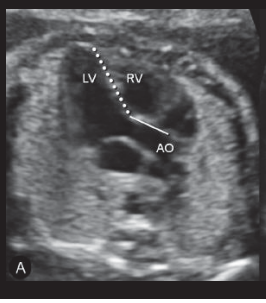


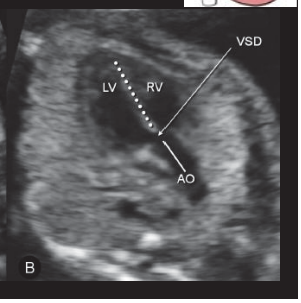
Left Ventricular Outflow

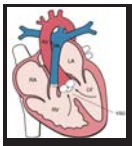
Normal

TOF



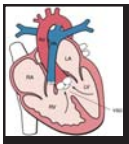






Left Ventricular Outflow

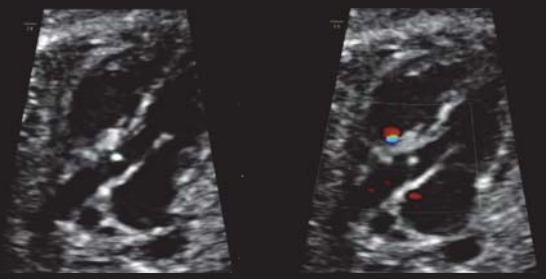
TOF



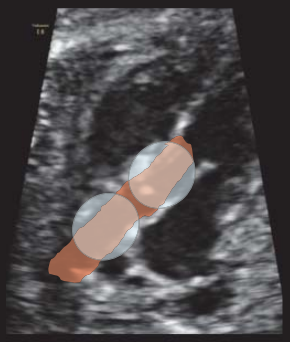
Left Ventricular Outflow

TOF

Left Ventricular Outflow



Left Ventricular Outflow



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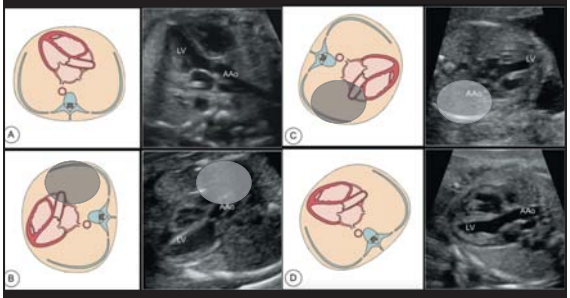
Left Ventricular Outflow



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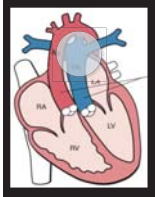
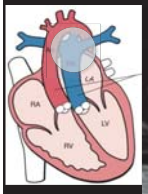
Left Ventricular Outflow

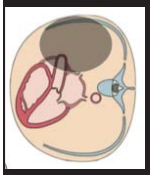
TGA: Left Ventricular Outflow

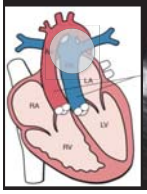


TGA: Left Ventricular Outflow

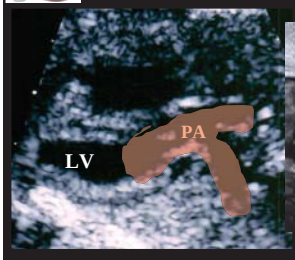


TGA: Left Ventricular Outflow





Transposition of Great Arteries



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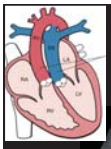
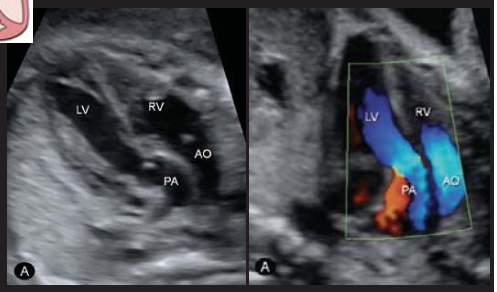
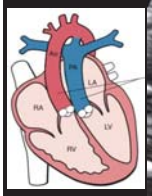
Transposition of Great Arteries



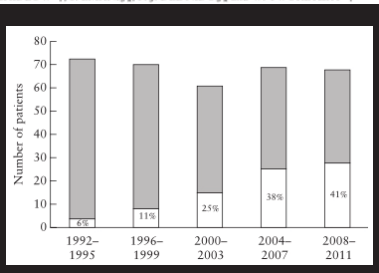


Transposition of Great Arteries



Prenatal diagnosis of transposition of the great arteries over a 20-year period: improved but imperfect

M. C. ESCOBAR-DIAZ*†‡, L. R. FREUD*†, A. BUENO*†, D. W. BROWN*†, K. G. FRIEDMAN*†
D. SCHIDLOW*†, S. EMANI§¶, P. J. DEL NIDO§¶ and W. TWORETZKY*†

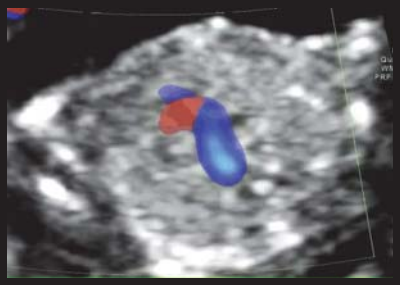


Overall rate of detection 23 %

Normal Left Ventricular Outflow

- Mitral - aortic continuity
- Aorta within left ventricle
- Angle of ascending aorta with ventricular septum
- Aorta does not divide
- Close observation of aortic valves

Transposition of Great Arteries



3VT – 2 vessels seen

Do Not Miss The Diagnosis of a Normal Left Ventricular Outflow

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Top 5 Critical Anatomic Regions

1. Normal Left Atrium
2. Normal Ascending Aorta
3. Normal Semilunar Valves

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Normal Semilunar Valves

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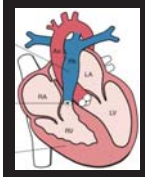
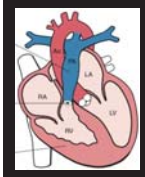
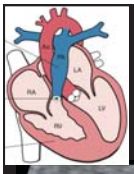
Pulmonary Stenosis

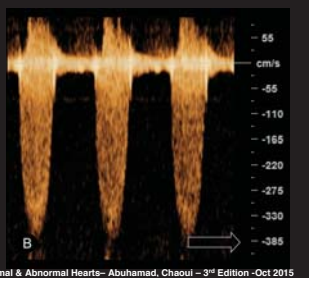


Pulmonary Stenosis



Pulmonary Stenosis





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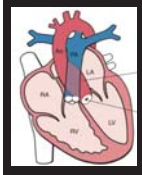
Aortic Stenosis



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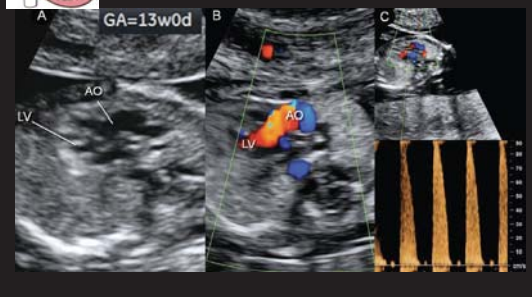
Aortic Stenosis



Aortic Stenosis

Aortic Stenosis

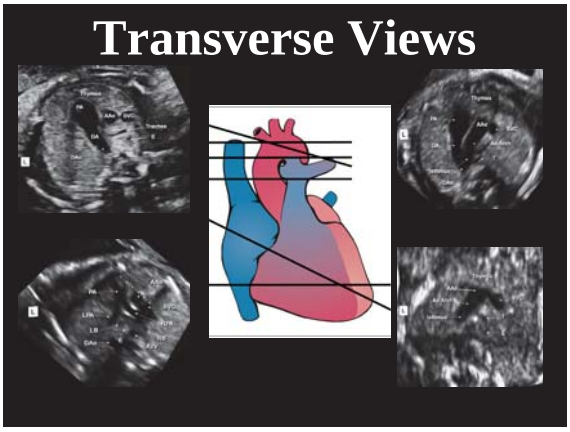
Pulmonary/Aortic Stenosis

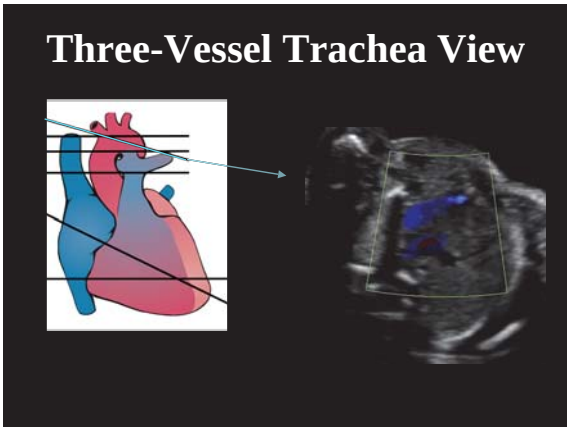
- Look at valve echogenicity
- Look at valve excursion in systole (cineloop should open fully in systole)
- Look at wall of vessel beyond the annulus
- Look at color Doppler for aliasing
- Look at pulsed Doppler for velocity

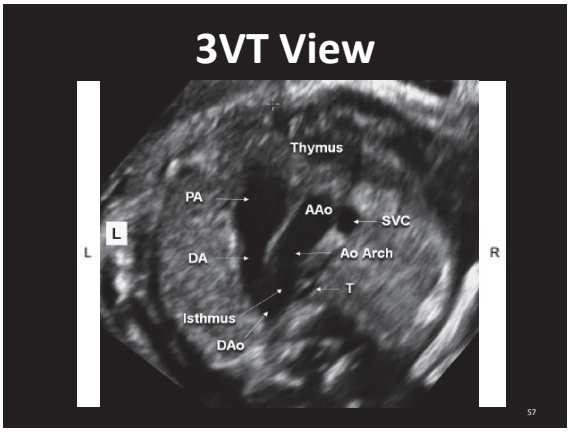
Do Not Miss The Diagnosis of a Normal Semilunar Valve

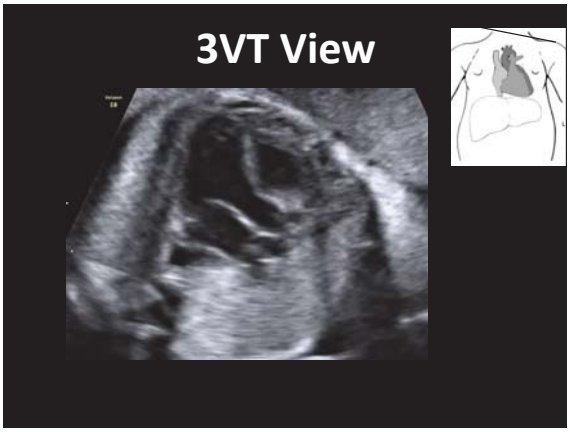
Top 5 Critical Anatomic Regions

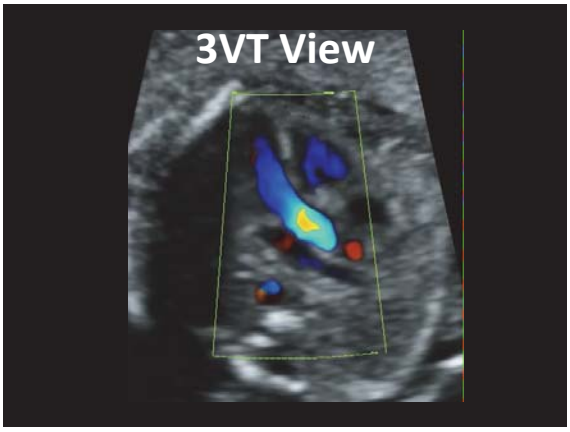
1. Normal Left Atrium
2. Normal Ascending Aorta
3. Normal Semilunar Valves
4. Normal 3VT View

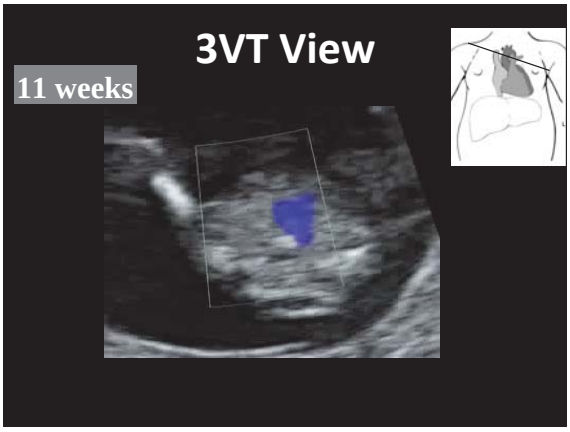

Transverse Views


Three-Vessel Trachea View

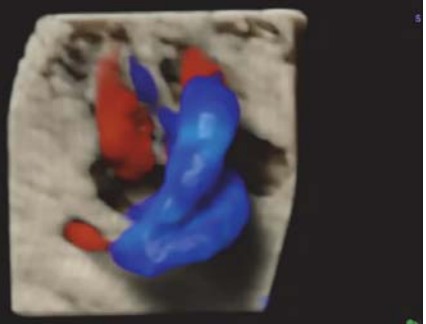

3VT View
Thymus
PA
AAo
SVC
L
DA
Ao Arch
R
T
Isthmus
DAo


3VT View


3VT View


3VT View
11 weeks

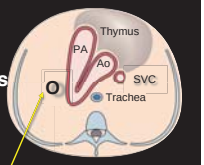
3VT View



Checklist 3-VT-view

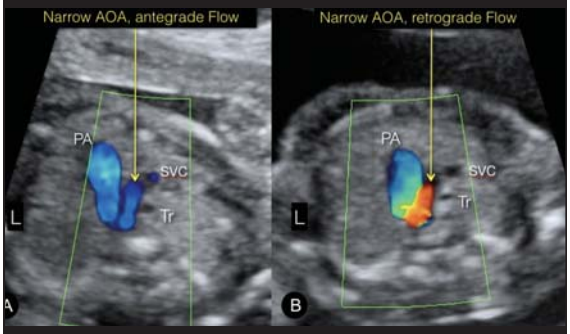
- Course and size of PA, Ao and SVC
- Aortic isthmus and ductus arteriosus
- Aortic arch right or left-sided ?

(Trachea as landmark)



- Thymus visualized
- Assessment with Color Doppler: "Blue V" or "Red V"
- Atypical vessels (left persistent SVC - Vertical Vein)

3VT View: 3 Vessels Seen



Coarctation of the Aorta | Hypoplastic Left Heart

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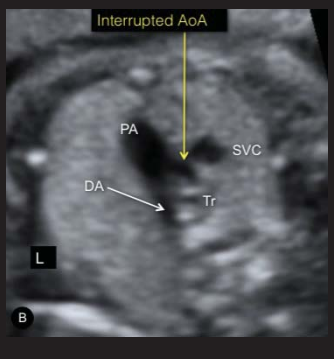
3VT View

HLHS

3 Vessels Seen



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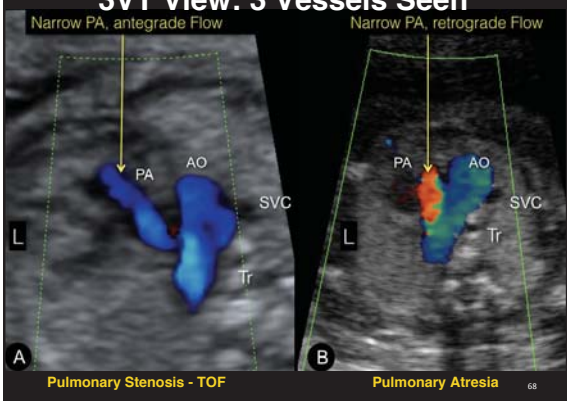
3VT View: 3 Vessels Seen



Pulmonary Stenosis - TOF | Pulmonary Atresia

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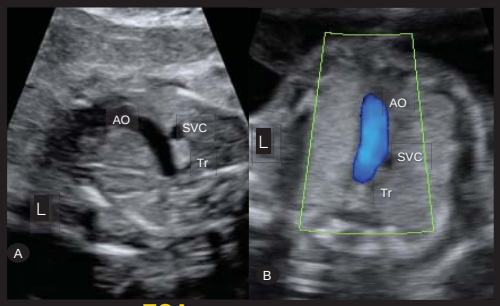
3VT View: 3 Vessels Seen

TOF

3VT View: 2 Vessels Seen
Normal size great vessel



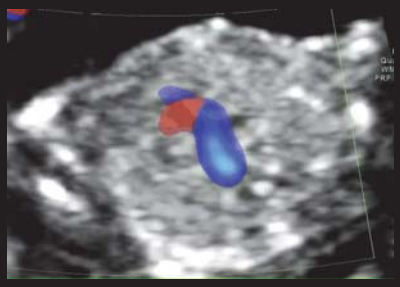
TGA (Convex Course)

Transposition of Great Arteries

13 weeks

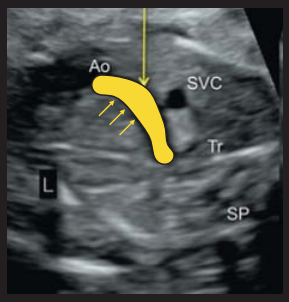
3VT – 2 vessels seen

Transposition of Great Arteries



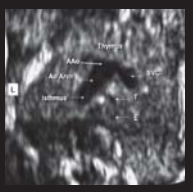
3VT – 2 vessels seen

Transposition of Great Arteries



Convex-shape of Aorta

Normal Aortic arch view



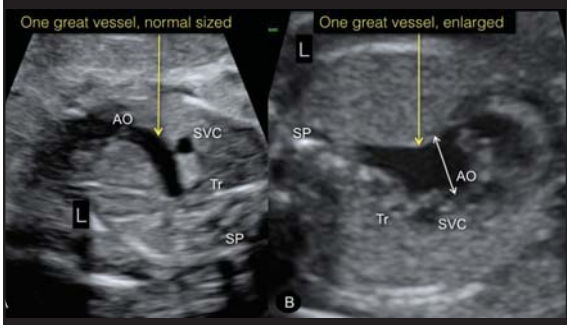
Do not confuse with an abnormal 3VT View

Orientation of Great Arteries

Normal TGA

75

3VT View: 2 Vessels Seen



TGA (Convex Course) CAT

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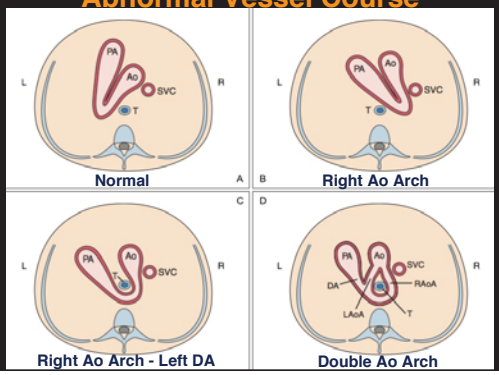
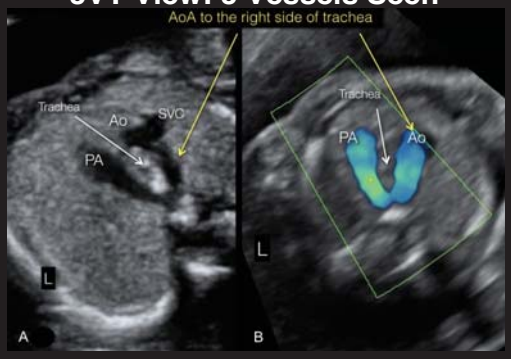
3VT View: 3 Vessels Seen
Abnormal Vessel Course



3VT View: 3 Vessels Seen



Right-Sided Aortic Arch

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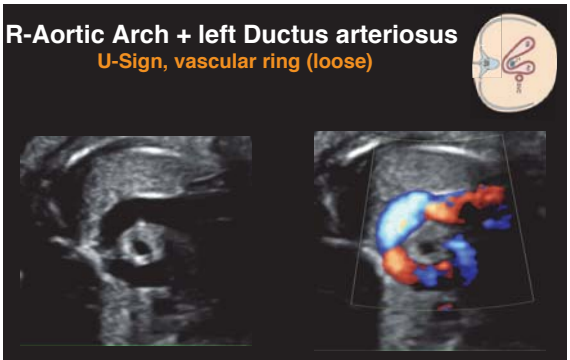
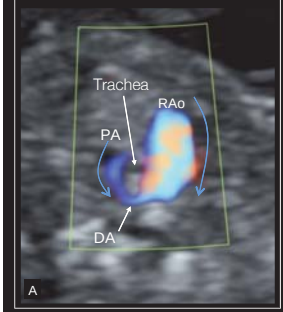
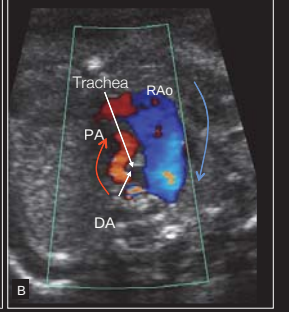
R-Aortic Arch + left Ductus arteriosus
U-Sign, vascular ring (loose)

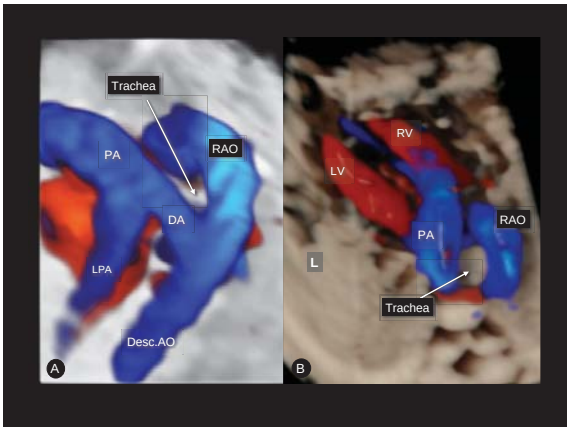


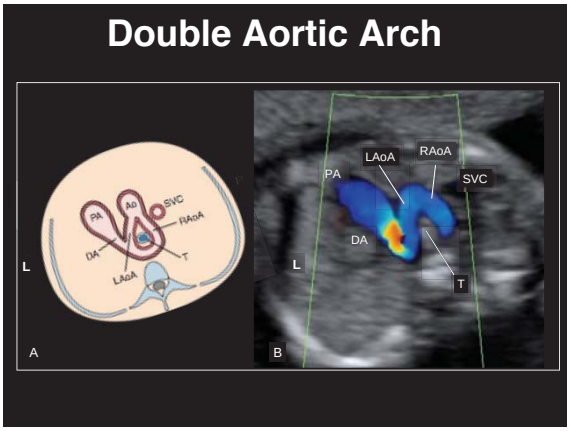
Right Ao.Arch & left DA in Tetralogy of Fallot

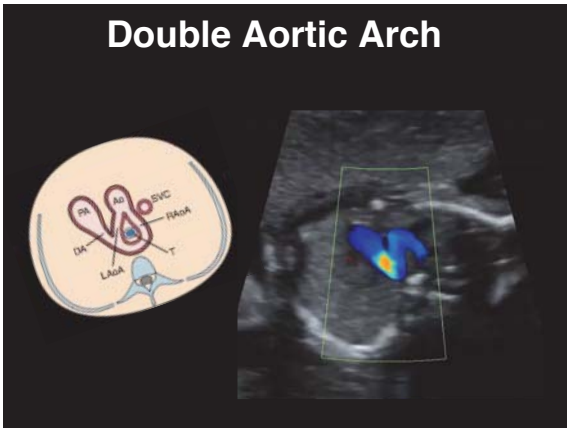


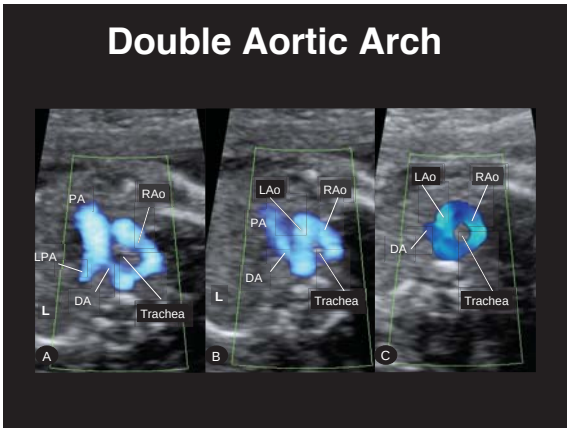
Right Ao.Arch & left DA in Pulmonary Atresia with VSD

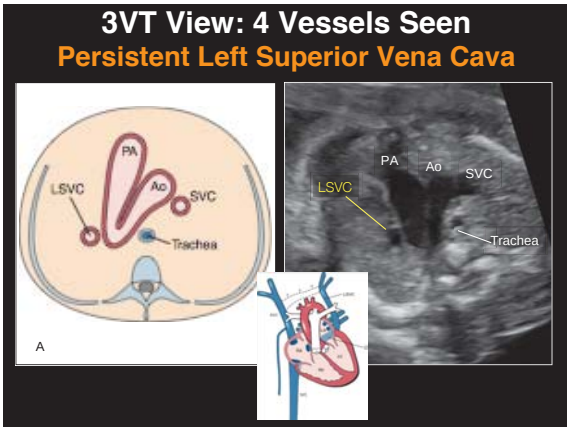


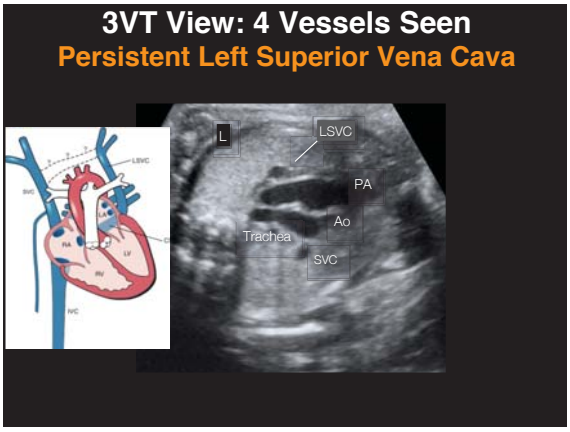












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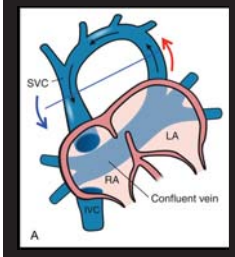
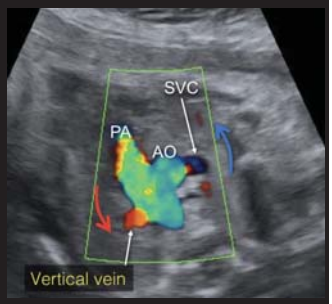
3VT View: 4 Vessels Seen
Supracardiac TAPVR





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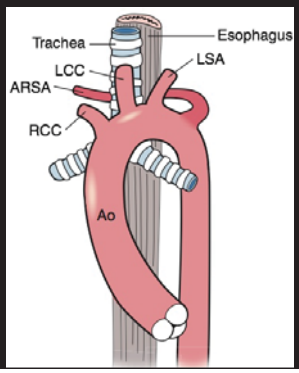
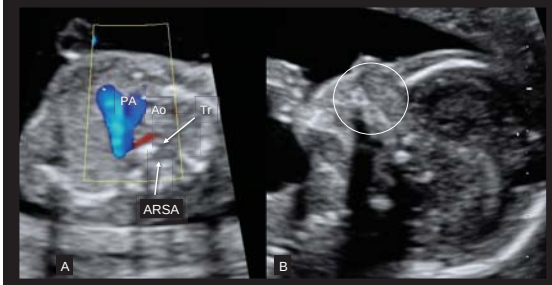
Aberrant Right Subclavian Artery

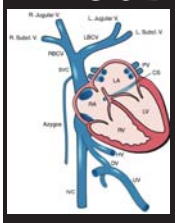


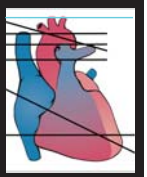
Aberrant Right Subclavian Artery

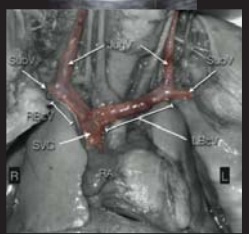
Aberrant Right Subclavian Artery



Left Brachiocephalic Vein



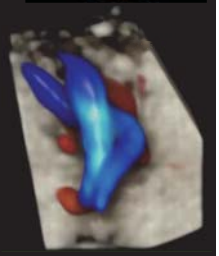




3VT View

Abnormal in:

- HLHS
- HRHS
- TGA
- DORV
- TOF
- CAT
- TAPVR
- PA-VSD
- PS / PA
- Critical Ao Stenosis
- Coarctation of Ao
- ARSA
- LSVC
- TA-VSD
- RAA
- Double Ao Arch
- Ebstein
- Interrupted Ao Arch



Courtesy of Dr. Chaoui

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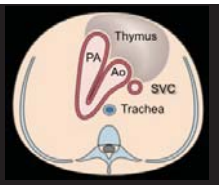
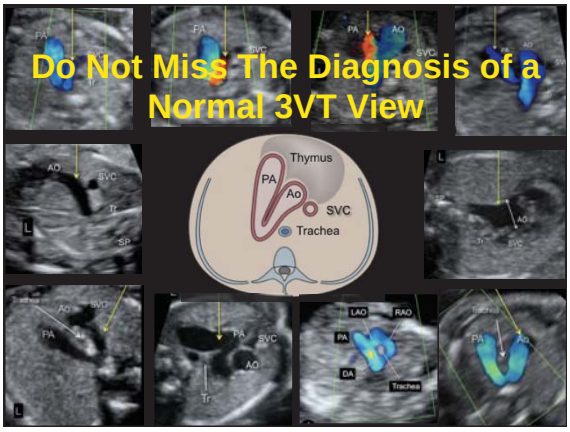
3VT View



- In my opinion, the most important ultrasound view in the fetus
- It is easy to obtain - especially in early gestation
- Anatomic landmarks easy to identify and master
- It is affected in most major CHD
- Strong consideration should be given to its incorporation in screening ultrasound examinations



Top Critical Anatomic Regions

1. Normal Left Atrium
2. Normal Ascending Aorta
3. Normal Semilunar Valves
4. Normal 3VT View
5. Normal Cardiac Axis in Early Gestation

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Top 5 Critical Anatomic Regions

1. Normal Left Atrium
2. Normal Ascending Aorta
3. Normal Semilunar Valves
4. Normal 3VT View
5. Normal Cardiac Axis in Early Gestation

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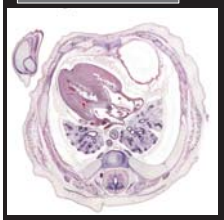
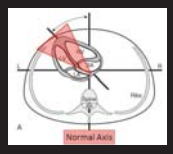
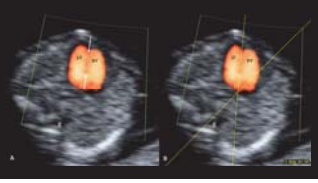
Cardiac Axis

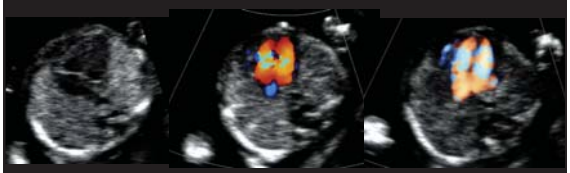
9 weeks - 52 days







Cardiac Axis



2D | 2D+color Doppler | 2D+HD color Doppler

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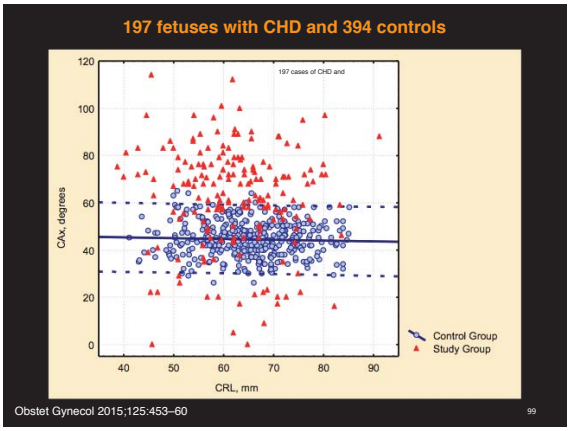
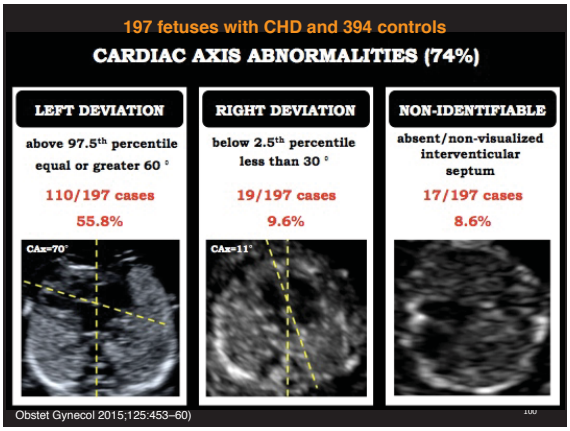
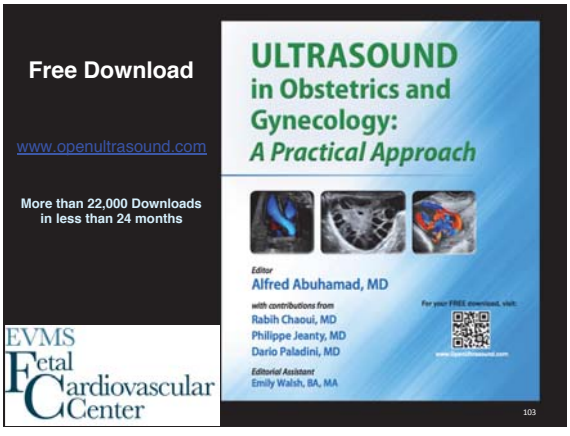




5 Top Anatomic Regions

- **Normal left atrium:** rule out TAPVR, Isomerism, Interrupted IVC
- **Normal ascending aorta:** rule out TOF, TGA, DORV
- **Normal semilunar valves:** rule out AS, PS, HLHS, PA
- **Normal 3VT view:** rule out conotruncal anomalies, RAA, HLHS, coarctation of Ao
- **Normal cardiac axis in early gestation:** rule out major CHD in first trimester




Notes

Hands-On Scanning Demonstration: Fetal Anatomy Review in Early Gestation

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Anomalies of the Fetal Gastrointestinal Tract

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FETAL GASTROINTESTINAL MALFORMATIONS AND ABDOMINAL WALL DEFECTS

Lawrence D. Platt, MD
Clinical Professor of Obstetrics and Gynecology
David Geffen School of Medicine at UCLA
Director, Center for Fetal Medicine & Women's Ultrasound
Los Angeles, CA

Objectives

- Identify normal sonographic appearance of the fetal GI tract, diaphragm and abdominal wall
- Learn normal views of these structures
- Systematic approach to evaluate anomalies
- Understand pitfalls in imaging the fetal abdomen

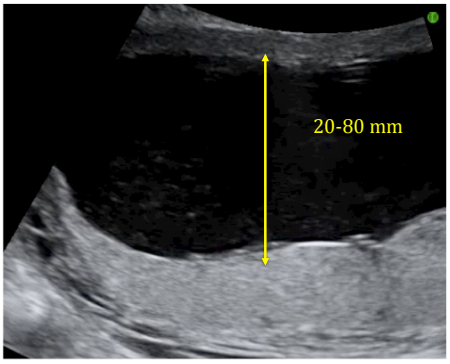
Systematic approach to diagnosing GI malformations

- Become familiar with normal views to detect what is normal
- Abnormal structures in the abdomen
  - Isolated or multiple?
  - If cystic, discrete or corrected?
- Anatomical landmarks to locate region
  - Upper or lower abdomen, midline or lateral?
  - Arising from or near which organ?
- Amniotic fluid

Check the fluid levels

- Amniotic fluid
- Fluid outside the organs (ascites)
- Fluid filled organs
- Too much?
- Too little?

Amniotic Fluid



Amniotic Fluid

- Too much?
  - Look for gastrointestinal obstructions
- Too little?
  - Evaluate possible urogenital disorders

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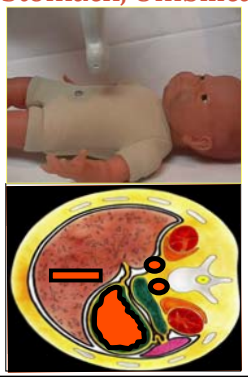
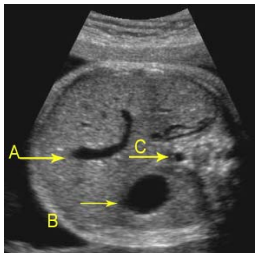
Anatomical Landmarks

- Amniotic fluid
- Diaphragm
- Liver
- Stomach
- Intestine
- Abdominal wall
- Umbilical vein
- Bladder
- Kidneys

Stomach, Umbilical Vein, and Aorta



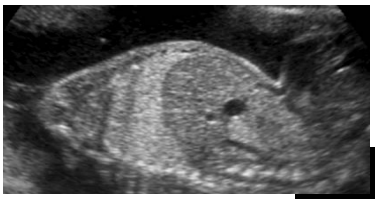


Diaphragm

Small Bowel

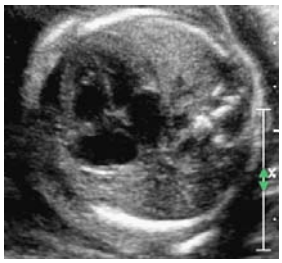
- Middle-to-lower part of the fetal abdomen
- Hyperechoic

Fetal Diaphragm

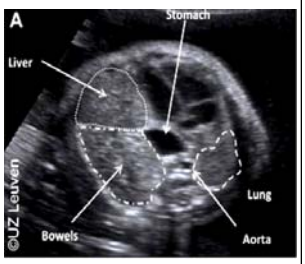


Diaphragm Evaluation from a 4-Chamber View

Normal



Abnormal



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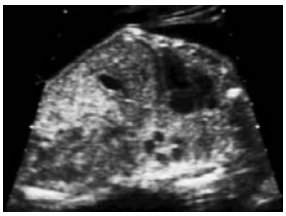
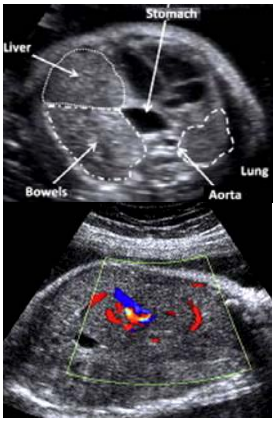
Normal and abnormal diaphragm



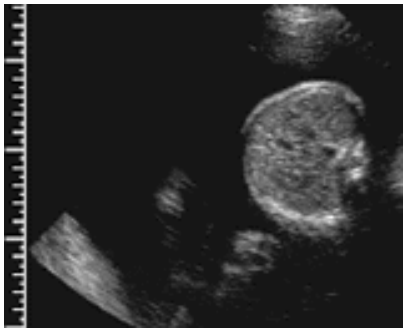


Gastro-intestinal Obstructions

What are the causes?

- Intrinsic
  - Atresia
  - Web
- Extrinsic
  - Bands
  - Volvulus
- Exact cause in not seen antenatally

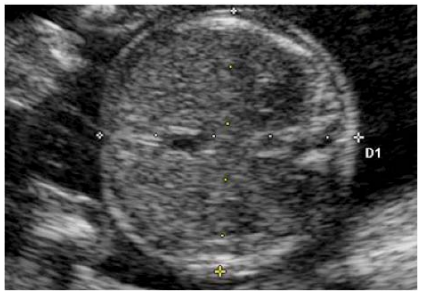
Polyhydramnios with absent stomach

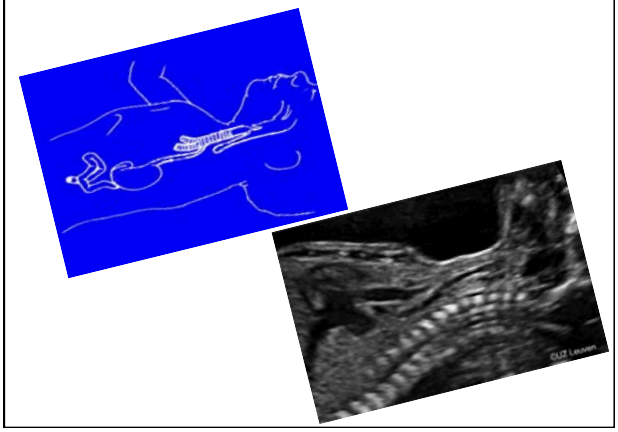


Esophageal atresia

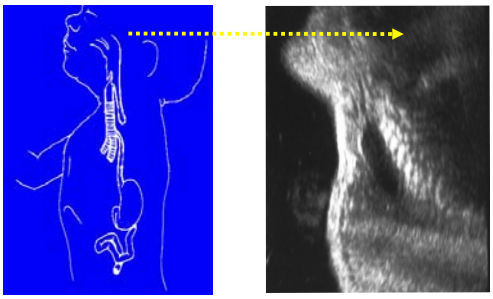
- Indirect signs
  - Small or invisible stomach
  - Polyhydramnios

When the stomach is not visible … wait for an hour and look again





Esophageal atresia



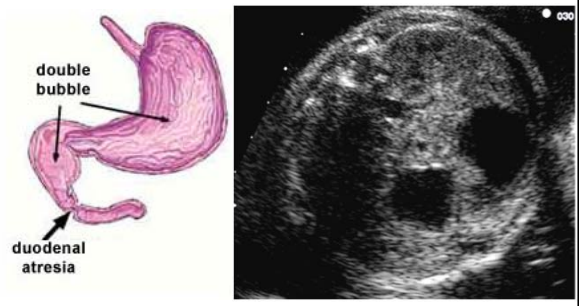
Esophageal atresia

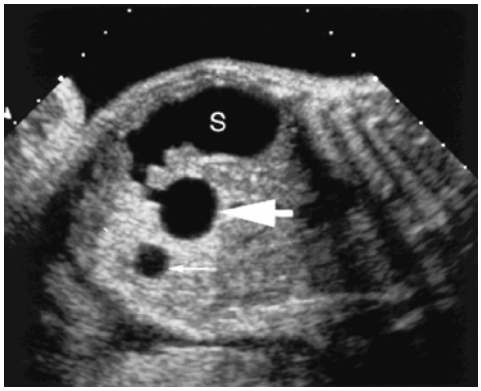
- Prenatal detection rate 40%
- Associated anomalies 80%
- Prenatal diagnosis usually occurs in 3rd trimester

Duodenal atresia - double bubble

- Associated with polydramnios
- "Double bubble" sign is the result of a dilated stomach and proximal duodenum
- Two communicating cystic masses appear in the upper abdomen
- 30% of duodenal atresia cases are associated with T21
- Caution urged not to misinterpret a normal gallbladder adjacent to the stomach as a "double bubble"

Duodenal atresia – double bubble association with Trisomy 21





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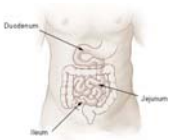
Dilation of Small Bowel & Colon

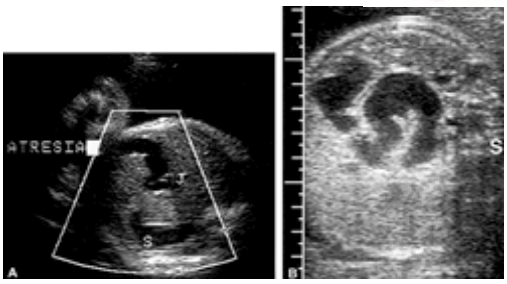
- Diameter of lumen of fetal small bowel rarely >6 mm
- Congenital small bowel syndrome:
  - Fatal, but rare condition
  - Few familial cases reported associated with malrotation
  - U/S presentation: delayed return of midgut to the abdominal cavity, dilated loops ad polyhydrmanios
- Diameter of lumen of fetal colon rarely >20 mm

Jejunal atresia

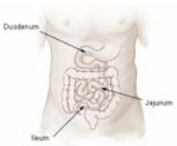
- Most upper bowel obstruction seen with polyhydramnios and dilated loops of bowel may also be seen
- Jejunal atresia may give rise to a "triple bubble" sign
- Typical appearance:
  - Proximal small bowel dilatation appears as multiocular sonolucent masses
  - May be a single, fluid-filled bubble in the abdomen
  - Peristaltic movement of small particles in the fluid-filled lesion
  - No dilatation is visible in other parts of the interesting

Proximal jejunal atresia

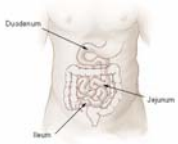




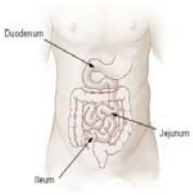
Distal jejunal atresia

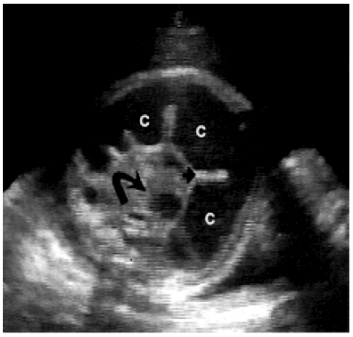


Ileal atresia



Colonia atresia





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Unusual lesions – mesenteric cysts

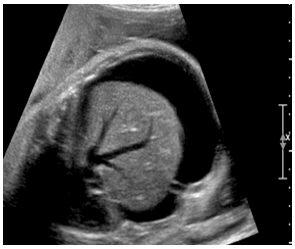
Meconium peritonitis

- Meconium released by a bowel perforation causes chemical peritonitis
- Meconium deposits may calcify creating a "snowstorm" appearance

Causes of Fetal Ascites

- Gastrointestinal causes
  - Bowel atresias
  - Volvulus
  - Meconium peritonitis
  - Bowel obstruction with perforation

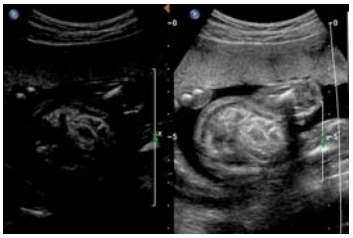
Ascites



Echogenicity / Calcification

NOTE: Calcium has a distal shadow, echogenicity does not

Echogenic Bowel



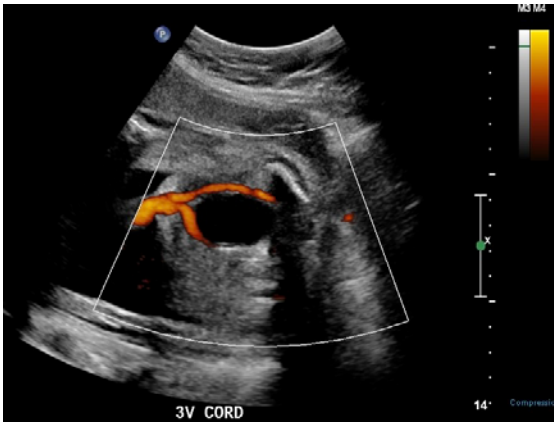
Limb-Body Wall Complex

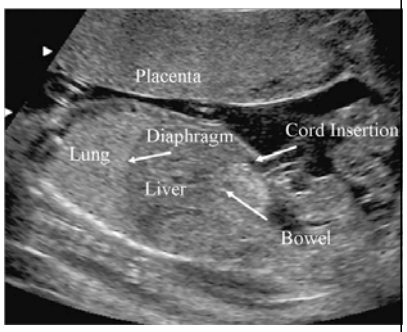
- Lethal condition with severe anterior abdominal wall defect (defect is usually placed laterally involving the umbilical cord insertion size)


Multiple Cavernous Hemangiomas

- Associated with Klippel-Trenaunay-Weber syndrome
- Diagnosed in presence of multiple surface masses producing limb hypertrophy
- Hydrops may occur


Fetal Abdominal Wall: Anterior

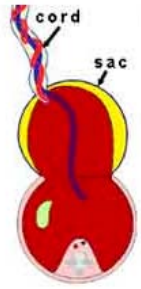



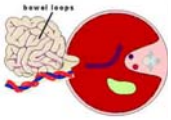
Omphalocele: High-risk Trisomies 13 & 18


Physiologic gut herniation <week 12

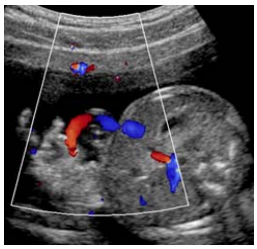
Omphalocele



Gastroschisis: Low risk aneuploidy



Gastroschisis



Gastroschisis

Pitfalls in diagnosing GI abnormalities

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Take home points

- When you find an anomaly …
- Try to complete the examination

- Assess for any other signs of aneuploidy
- If you find one abnormality, there is often another one waiting to be found

Notes

Anomalies of the Fetal Chest

Elena Sinkovskaya, M.D., Ph.D.
Assistant Professor
Director of Research
Division of Maternal-Fetal Medicine
Department of Obstetrics & Gynecology
Eastern Virginia Medical School
Norfolk, VA

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Advances in Ultrasound in Obstetrics and Gynecology
October 14-15, Washington DC

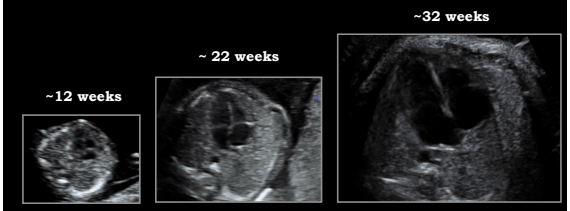
ANOMALIES OF THE FETAL CHEST

Elena Sinkovskaya MD, PhD

Division of Maternal-Fetal Medicine

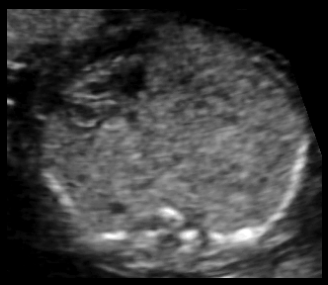
EVMS
Eastern Virginia Medical School

SCREENING EVALUATION

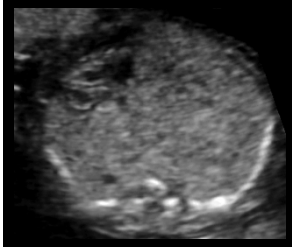


On ultrasound fetal lungs appear as homogeneous and slightly echogenic tissue surrounding the heart; no evidence of effusion

What is wrong?



Congenital high airway obstruction (CHAOS)



Incidence: very rare

Causes:

- Laryngeal atresia
- Tracheal atresia
- Subglotic stenosis
- Laryngeal web

CHAOS: sonographic presentation

Fetal Lungs mass

- Bilateral
- Enlarged
- Symmetric distended
- Echogenic
- Homogeneous

Fetal Heart

- Anterior and midline
- Compressed (restrictive CMP)

CHAOS: sonographic presentation

Other findings:

- Flattened/inverted diaphragm
- fluid-filled bronchi & trachea
- Ascites/Hydrops
- Poly-/Oligohydramnios
- Placentomegaly

CHAOS: prognosis & management

Associated anomalies: **more than 50%** (Fraser' s syndrome)

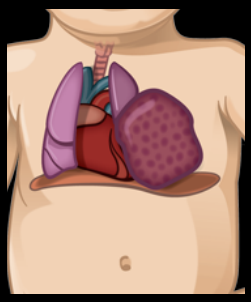
Expectant management: **poor outcome** (<10% survival)

Fetal treatment: **poor outcome**

Postnatal management: ***ex utero*** **intrapartum treatment (EXIT) with tracheostomy** (reported to have 90% survival)

Cavoretto P et al. UOG 2008;32:769-783

Congenital Cystic Adenomatoid Malformation (CCAM)



Incidence: **is most common thoracic mass in the fetus**

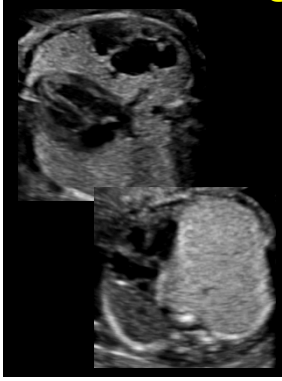
Prenatal studies: 1:4,000

Postnatal studies: 1:25,000

Associated anomalies: **8-12 %**

Risk for chromosomal anomalies is not increased

CCAM: imaging overview



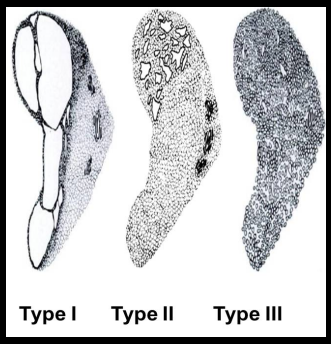
Fetal Lungs mass

- **Usually unilateral**
- **Well defined**
- **Involves part of the lung**
- **Solid or cystic**
- **No systemic blood supply**

Fetal Heart

- **Is displaced right/left**
- **Compressed in severe cases**

CCAM: Histological classification



Stocker JT et al. Human Pathol 1977;8:155-171

CCAM: imaging overview

Macrocystic Mixed Microcystic

CCAM: imaging overview

Macrocystic Mixed Microcystic

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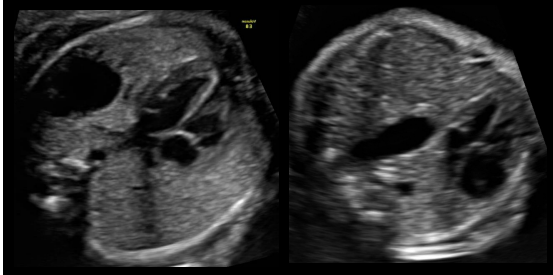
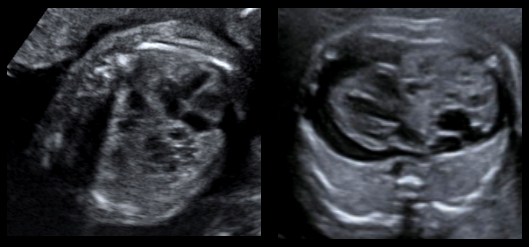
CCAM: differential diagnosis

CCAM vs. CDH



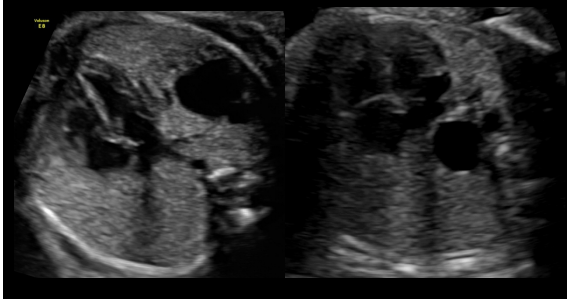
CCAM: differential diagnosis

CCAM vs. Pericardial Teratoma



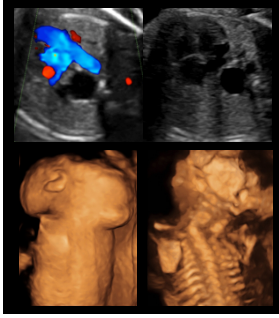
CCAM: differential diagnosis

CCAM vs. Neuroenteric cyst



CCAM: differential diagnosis

Neuroenteric cyst



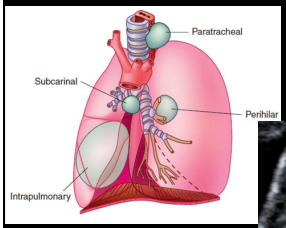
Incidence: unknown

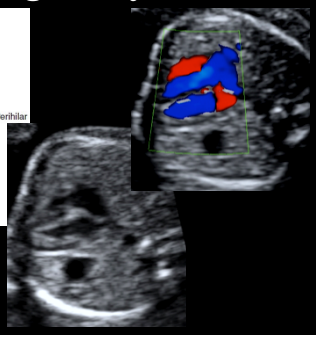
Sonographic appearance:

- cystic mass
- septated or bilobed
- in posterior chest
- spinal anomalies – ALWAYS, and located more cephalad

CCAM: differential diagnosis

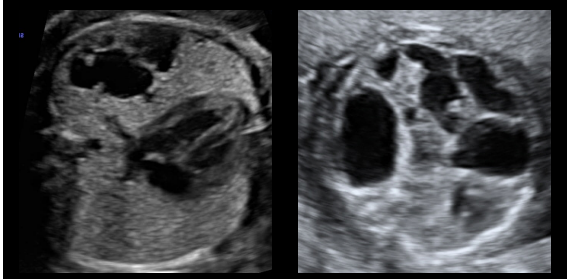
Bronchogenic cyst





CCAM: differential diagnosis

CCAM vs. Lymphangioma



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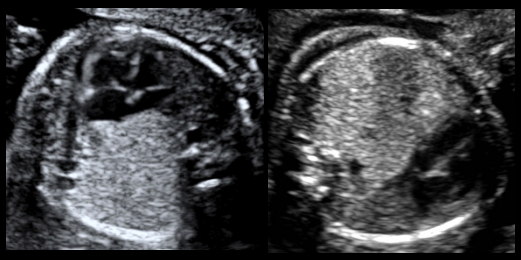
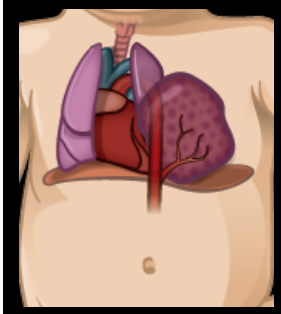
CCAM: differential diagnosis

CCAM vs. PS



Pulmonary sequestration (PS)



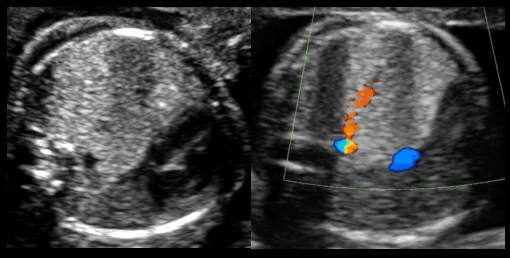
Incidence: 23% of all fetal chest masses

Associated anomalies: rare

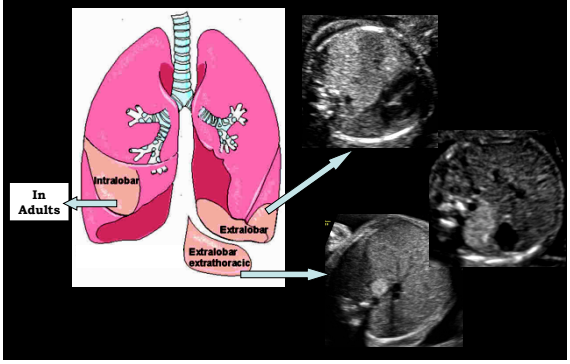
Risk for chromosomal anomalies is not increased

PS: imaging overview

Echogenic mass with systemic blood supply

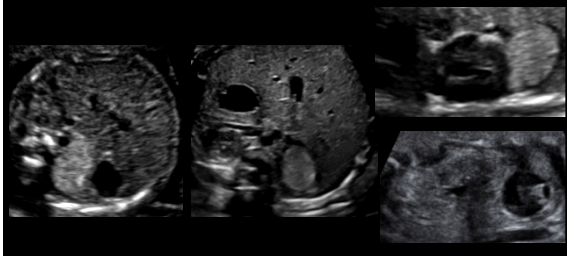


PS: imaging overview

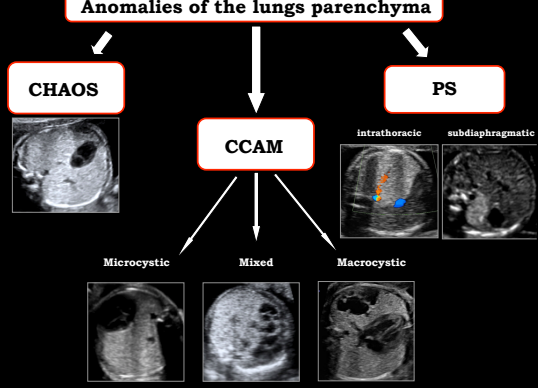


PS: differential diagnosis

PS vs. Adrenal Hemorrhage



Anomalies of the lungs parenchyma



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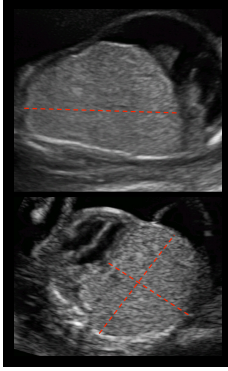
ULTRASOUND PROTOCOL

- Mass location (unilateral, bilateral)
- Mass size (MVR)
- Homogeneity
- Presence of mediastinal shift
- Diaphragm distortion
- Evidence of hydrops
- Evidence of polyhydramnios
- Cardiac function

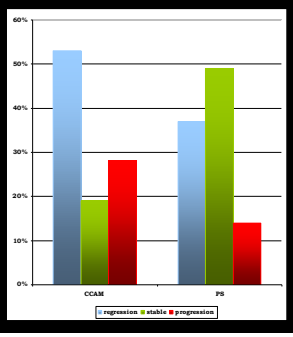
CCAM volume ratio (MVR)



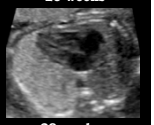
$$\frac{\text{L (cm)xW(cm)xH(cm)x0.52}}{\text{Head Circumference (cm)}}$$

CVR >1.6
predicts Hydrops in 75%

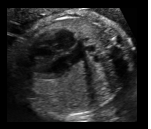
Change with gestation



20 weeks



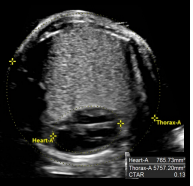
29 weeks

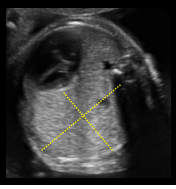


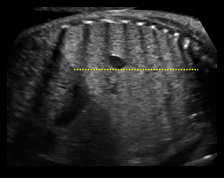
36 weeks

High-risk group

- Mass volume ratio >1.6
- At least 3 cm in at least 2 dementions
- Presence of severe mediastinal shift
- Diaphragm distortion
- Cardiac compression (CTAR < 0.2)



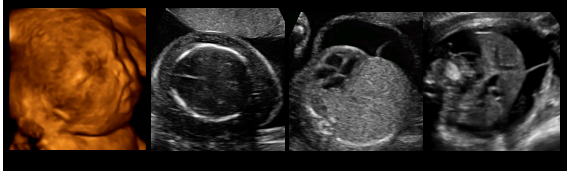




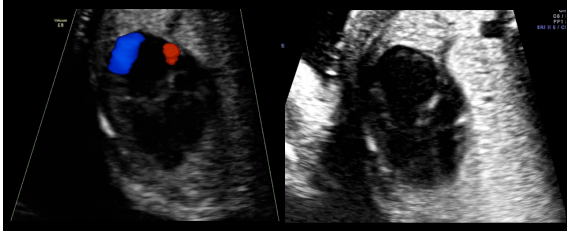
Complications

5.7% cases of CCAM resulted in hydrops fetalis

6.2% cases PS is associated with plural effusion



All fetuses with large lesions have sings of myocardial dysfunction



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CCAM: prognosis & management
STEROIDS

- Administering betamethasone antenatally to fetuses with CCAM and non immune hydrops there was complete resolution of hydrops
- This area needs further research owing to the small data sample.

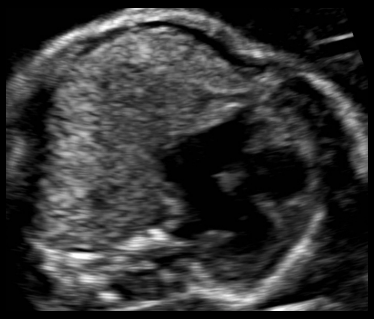
CCAM and PS: prognosis & management

- In the absence of hydrops: >95% survival
- CCAM with hydrops:
  - expectant 95% mortality
  - shunting 65% survival
  - fetal surgery 45% survival
- **Availability of extracorporeal membrane oxygenation (ECMO) is crucial for infant survival with large lesions**

CCAM and PS: Prognosis

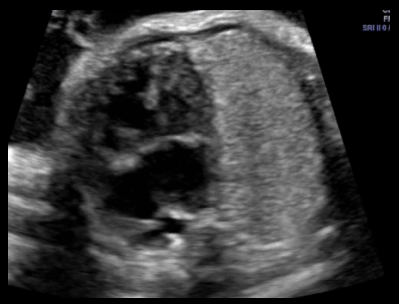
- Outcome is good in the absrnce of the hydrops
- Intrauterin amelioration appears to be common but lesions rarely regress
- All CCAM should be removed
- Large lesions increase risk for scoliosis

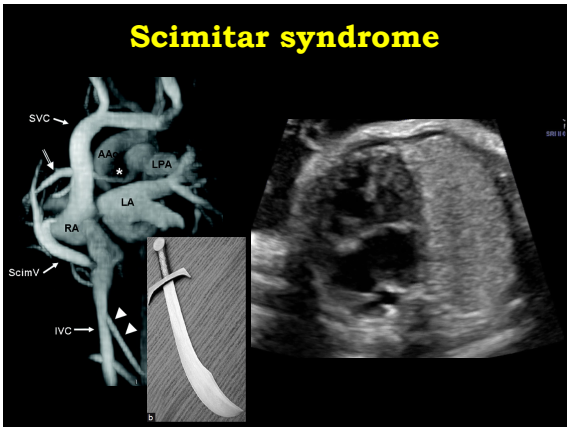
What is wrong?



Unilateral lung agenesis

What is wrong?





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Hands on Scanning Demonstration: The Detailed (76811) Ultrasound Examination

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Debate on the Role of Routine Cervical Length in Preterm Labor Prevention

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