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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

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

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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
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.

CPT Coding and RVU’s

CPT
- Professional component
- Technical component

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

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.

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.

Personal Supervision

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

Personal Supervision of Gyn US

• Sonohysterography (ultrasound)
  • 76831 - TC

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

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


Inability to Visualize Anatomy
Obese women
• Ultrasound at 20-22 weeks
• 2 weeks later than in the nonobese patient

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

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.

Fetal at increased risk for a congenital anomaly:
- Maternal pregestational diabetes or gestational diabetes before 24 weeks
- High BMI (> 35 kg/m²)
- Multiple gestation
- Abnormal maternal serum analytes
- Teratogen exposure
- 1st trimester NT > 3.0 mm

Other conditions affecting the fetus:
- Congenital infections
- Maternal drug dependence
- Isoimmunization
- Oligohydramnios
- Polyhydramnios

Suspected or known chromosomal abnormality

Abnormal finding on antenatal screening
- 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

#### 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

#### Biophysical Profile
- **76818** Fetal biophysical profile; with non-stress testing.
- **76819** Fetal biophysical profile; without non-stress testing.

### 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/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 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

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

Adnexa

Ovaries

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

Fallopian Tubes

- Usually not visualized
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
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

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.

Thank You

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

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Contribution Not Received in Time for Inclusion.
The Normal and Abnormal Fetal Skeleton

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Normal and Abnormal Fetal Skeletal Anatomy

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

- Unknown 50%
- Multifactorial 25%
- Monogenic 8%
- Environmental 7%

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

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)
Endochondral Ossification

19 WEEKS

Endochondral Ossification

10 WEEKS

Endochondral Ossification

11 WEEKS

Endochondral Ossification

12 WKS

Endochondral Ossification

14 WKS

Skeletal Bones: Normal

Profile: 16 Weeks

Hand: 16 Weeks 2D
Femur: 25 Weeks 2D

Femur: 25 Weeks 3D Static

Femur: 25 Weeks 3D VCI Omni View

Sternal Ossification

Sternal Ossification

Scapula: 2D
Hand: 3D Static

Normal Long Bones

Normal Long Bones

Normal Profile

Normal Profile

Normal Profile

Normal Profile

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**Most Common Diagnoses in Prenatal Onset Osteochondrodysplasias**

- Osteogenesis type II: 20%
- Thanatophoric Dysplasia: 11%
- Achondrogenesis II: 8.2%
- 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

**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**

- Size
- Shape
- Echodensity
- Lethal
- Non-lethal
- Thoracic Circumference
- Other congenital abnormalities

**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
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
- Platybasia
- 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

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 - Phalanges

OI Type II - 3D Imaging

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
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**
- Dimineralized calvarium
- Fractures
- Platspondyly
- Normal appearing hands

**Cleidocranial Dysplasia**
- Autosomal Dominant
- Dimineralization of the calvarium
- Clavicle hypoplastic

**Hypophosphatasia**
- Autosomal Recessive
- Fractures
- “Chromosome-appearing” bones
- Very poor mineralization of the hands

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

OI vs. Campomelic Dysplasia

Campomelic Dysplasia

Thanatophoric Dysplasia

**Radiologic Findings**
- Long narrow trunk
- Platypondylia
- Marked shortening of long bones
- Lateral spikes
- Abnormal pelvis

**Clinical Findings**
- AD
- Heterozygosity SOX9
- Recurrence - germline mosaicism
- Micrognathia
- Micro/macrocephaly
- Brachydactyly
- Multiple organ system involvement
- Not uniformly lethal

**Thanatophoric Dysplasia**

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

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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

24 weeks

FGFR2 disorders

Craniosynostosis

Apert’s syndrome

Normal
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

Achondroplasia

Autosomal Dominant
60% 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 – Distal Femurs

Achondroplasia - facies

Abnormal Calcaneus

- Phenytoin
- Warfarin
- Alcohol
- Rubella
- Trisomy 21; Trisomy 18; Mosaic trisomy 9
- Mucolipidosis, type II
- Gangliosidosis I
- Smith-Lemli Opitz syndrome
- Chondrodysplasia punctata, all types

- Thanatophoric Dysplasia
- Short-rib polydactyly syndromes
- Chondroectodermal dysplasia
- Asphyxiating thoracic dysplasia
- Larsen syndrome
- Atelosteogenesis types I and III
- Roberts-SC Phocomelia

Calcaneus

- Stippling - CP
- Duplicated - SRP

Calcaneus - Robert’s SC Phocomelia

Fetal Hands

- Achondroplasia
- Ellis van Creveld

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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 counselling 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

- 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

- 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 determining 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

- 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 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?

Prenatal Diagnosis/Ultrasonography

Genetic Counseling Radiology
Histomorphology/Pathology Molecular Biology

Prenatal Care and Diagnosis

FGFR3

International Skeletal Dysplasia Registry

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ULTRASOUND OF THE FETAL GENITOURINARY TRACT

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Contribution Not Received in Time for Inclusion.
The 11 - 14 Weeks Obstetrical Scan: Current Concepts and Future Directions

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Contribution Not Received in Time for Inclusion.
Fetal Neurosonography
in Early Gestation:
What Can Be Diagnosed Today?

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

Elena Sinkovskaya, M.D., PhD

What is Fetal Neurosonography?

Basic Examination
1. Screening (performed on everybody)
2. Usually transabdominal ultrasound
3. Three axial planes

Fetal Neurosonogram
1. Performed by indications
2. Transabdominal and transvaginal ultrasound
3. Multiple axial, coronal and sagittal planes
4. Extra training and expertise

Basic Examination
13+6 weeks transventricular plane
23+6 weeks transthalamic plane
13+6 weeks transcerebellar plane

Sonographic examination of the fetal central nervous system: guidelines for performing the ‘basic examination’ and the ‘fetal neurosonogram’
First Trimester Fetal Imaging:

- Transverse View
  - Presence of:
    - skull bones
    - 2 hemispheres
    - 2 symmetrical ventricles
    - Homogenous appearance of choroid plexus

- Midsagittal View
  - Assessment of:
    - the brainstem
    - 4th ventricle – IT?
    - cisterna magna

Intracranial Translucency (IT)

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

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

Limitations of the IT interpretation

* Prenat Diagn. 2011;31:103-106
Transabdominal vs. Transvaginal

Fetal Neurosonogram:
how does it work?

Midsagittal view

Fetal Neurosonogram:
Sagittal views

Fetal Neurosonogram:
Axial views
First Trimester Fetal Neuro Imaging:

Several studies have suggested that 1st trimester CNS screening for:
- acrania
- exencephaly/encephaly
- encephalocele
- holoprosencephaly
- 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

What’s wrong?

Anencephaly/Acrania

<table>
<thead>
<tr>
<th>Anencephaly</th>
<th>Acrania</th>
</tr>
</thead>
<tbody>
<tr>
<td>12 weeks</td>
<td>11 weeks</td>
</tr>
<tr>
<td>24 weeks</td>
<td>16 weeks</td>
</tr>
</tbody>
</table>
**What’s wrong?**

**Encephalocele**

<table>
<thead>
<tr>
<th>Location of the lesion</th>
<th>Incidence, %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Occipital</td>
<td>77%</td>
</tr>
<tr>
<td>Parietal</td>
<td>9%</td>
</tr>
<tr>
<td>Frontal</td>
<td>9%</td>
</tr>
<tr>
<td>Multiple</td>
<td>6%</td>
</tr>
</tbody>
</table>

**Iniencephaly**

**What’s wrong?**

**Alobar Holoprosencephaly**
Alobar Holoprosencephaly

What’s wrong?

Ventriculomegaly

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

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 holoprosencephaly is not a prerequisite for the ‘butterfly’ 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 cases with HPE in the first trimester. © 2013 John Wiley & Sons, Ltd.
Lateral ventricles in fetuses with aneuploidies at 11–13 weeks' gestation

The finding of contracted choroid plexus should raise suspicion of ventricular dilation
Ventriculomegaly
What’s wrong?

12 weeks 19 weeks

Ventriculomegaly
bilateral

13 weeks 22 weeks

Ventriculomegaly
unilateral

Choroid plexus cysts

Normal or Abnormal?
Normal or Abnormal?

Screening for spina bifida


Normal or Abnormal?

Normal or Abnormal?

Diagnosis of spina bifida

Normal or Abnormal?
Normal or Abnormal?

Dandy-Walker Malformation

Normal or Abnormal?

Screening for DWM

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

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
The Approach to Fetal Cardiac Imaging in Early Gestation

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Vice Dean for Clinical Affairs
Eastern Virginia Medical School
Norfolk, VA
Advanced Evaluation of the Fetal Heart

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

Left Atrium

Table of Contents
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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Transposition of Great Arteries

LV
PA
Transposition of Great Arteries

Overall rate of detection 23%
Top 5 Critical Anatomic Regions

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

Top 5 Critical Anatomic Regions

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

Pulmonary Stenosis

Pulmonary Stenosis

Pulmonary 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

3VT View

3VT View

3VT View

11 weeks

Table of Contents
• 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

3VT View

3 Vessels Seen
**3VT View: 3 Vessels Seen**

**3VT View: 2 Vessels Seen**
- Normal size great vessel

**TGA (Convex Course)**

**Transposition of Great Arteries**
- 13 weeks

**3VT – 2 vessels seen**

**Convex-shape of Aorta**

**Normal Aortic arch view**
- Do not confuse with an abnormal 3VT View
Orientation of Great Arteries

Normal TGA

3VT View: 2 Vessels Seen

One great vessel, normal-sized
One great vessel, enlarged

TGA (Convex Course) CAT

3VT View: 3 Vessels Seen

Abnormal Vessel Course

3VT View: 3 Vessels Seen

Right Ao Arch

Double Ao Arch

3VT View: 3 Vessels Seen

Right-Sided Aortic Arch

R-Aortic Arch + left Ductus arteriosus

U-Sign, vascular ring (loose)

R-Aortic Arch & left DA in Tetralogy of Fallot

R-Aortic Arch & left DA in Pulmonary Atresia with VSD

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Double Aortic Arch

3VT View: 4 Vessels Seen
Persistent Left Superior Vena Cava
3VT View: 4 Vessels Seen
Supracardiac TAPVR

Aberrant Right Subclavian Artery

Aberrant Right Subclavian Artery

Left Brachiocephalic Vein

3VT View

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


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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

**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
Do Not Miss The Diagnosis of a Normal Cardiac Axis in Early Gestation

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
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

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David Geffen School of Medicine at UCLA
Director, Center for Fetal Medicine & Women’s Ultrasound
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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
- Too much?
  - Look for gastrointestinal obstructions
- Too little?
  - Evaluate possible urogenital disorders

Table of Contents
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
Gastro-intestinal Obstructions
What are the causes?
- Intrinsic
  - Atresia
  - Web
- Extrinsic
  - Bands
  - Volvulus
- Exact cause is 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

- 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
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 polyhydramnios

- 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
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
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
ANOMALIES OF THE FETAL CHEST

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ANOMALIES OF THE FETAL CHEST

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Division of Maternal-Fetal Medicine

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

SCREENING EVALUATION

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

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**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**

**Prenatal lung mass**

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

**Prenatal Heart**

- Is displaced right/left
- Compressed in severe cases

**Type I**

**Type II**

**Type III**

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

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**CCAM: Histological classification**

**Microcystic**

**Mixed**

**Macrocystic**

---

**CCAM: imaging overview**

**Macro cystic**

**Mixed**

**Micro cystic**

M A R O C Y S T I C

M I C R O C Y S T I C
**CCAM: differential diagnosis**

**CCAM vs. CDH**

**CCAM vs. Pericardial Teratoma**

**CCAM: differential diagnosis**

**CCAM vs. 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 vs. Lymphangioma**

Table of Contents
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: differential diagnosis
PS vs. Adrenal Hemorrhage

Anomalies of the lungs parenchyma
CHAOS
CCAM
Microcystic
Mixed
Macrocystic
PS

Table of Contents
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)

\[ L \text{ (cm)} : W \text{ (cm)} : H \text{ (cm)} : 0.52 \]

Head Circumference (cm)

CVR >1.6 predicts Hydrops in 75%

High-risk group

• Mass volume ratio >1.6
• At least 3 cm in at least 2 dimensions
• 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 signs of myocardial dysfunction
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: 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 absence 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

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Scimitar syndrome
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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Practical Approach to Ultrasound in Obstetrics and Gynecology

June 24 – July 1, 2017

Canada North East Discovery aboard Ms. Veendam Holland America
Sail from Boston, Massachusetts to Montreal, Canada

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