ULTRASOUND OF THE FETAL GENITOURINARY TRACT

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RENAL DEVELOPMENT

Nephron formation

- Pronephros (3rd-5th week)
  - nonfunctional
- Mesonephros (5th-12th week)
  - able to form urine
  - renin production
- Metanephros
  - final stage
  - mature metanephric kidney

RENAL DEVELOPMENT

Metanephros

- Tubular function begins 9th-12th week
- Functional loop of Henle by 14th week
  - tubular reabsorption
- New nephron formation through 36 wks

AMNIOTIC FLUID DYNAMICS

Determinants of AFV

NORMAL SONOGRAPHIC ANATOMY

Fetal Kidneys

- Visualization - transvaginal
  - earliest 9 weeks
  - 100% by 13 weeks
- Visualization - transabdominal
  - earliest 13-14 weeks
  - most patients by 16-18 weeks
NORMAL SONOGRAPHIC ANATOMY
Fetal Kidneys

• Paraspinous
• Circular/ elliptical shape
• Hypoechoic
• Echogenic rim more prominent with advancing GA

Renal pelvis
– slit-like, central, anechoic

Medullae
– hypoechoic
– arranged in A-P orientation around pelvis

Cortex
– echogenicity similar to surrounding tissues

measurements

↑ gain, ↓ dynamic range

normal echogenicity transverse
NORMAL SONOGRAPHIC ANATOMY

Fetal Kidneys – 18 weeks
renal arteries

Fetal Kidneys – 36 weeks

Fetal Bladder
• Visualization
  – earliest 10-12 wks (TA or TV)
  – almost 100% by 16 wks (TA)
• Appearance
  – rectangular, anechoic
  – thin wall

Level of iliac crest in transverse
• Int iliac arteries along lateral walls
  – can aid in identification
• Variation in volume
  – filling - emptying cycle 20-30 min

Fetal Bladder – 13 weeks

Fetal Bladder – 18 weeks
NORMAL SONOGRAPHIC ANATOMY
Fetal Bladder – 30 weeks

Fetal Genitalia

- Visualization GA, position dependent
- Must distinguish labia from scrotum
  - Testicles descend 28-34 weeks
  - Testicles in scrotum 100% reliable
- ID of penis provides further evidence
- Pitfalls: prominent clitoris, small penis, undescended testicles

FIRST TRIMESTER GENITALIA

Table 1: Gender identification according to crown-rump length (CRL)

<table>
<thead>
<tr>
<th>Gestational age (weeks)</th>
<th>CRL (mm)</th>
<th>Patients identified by ultrasound (n)</th>
<th>Known gender at birth (n)</th>
<th>Lost to follow-up (n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>12 to 12 + 3</td>
<td>55.4-62.5</td>
<td>180</td>
<td>153 (85)</td>
<td>135 18</td>
</tr>
<tr>
<td>12 + 4 to 12 + 6</td>
<td>62.6-67.9</td>
<td>218</td>
<td>209 (96)</td>
<td>194 15</td>
</tr>
<tr>
<td>13 to 13 + 6</td>
<td>68.0-83.9</td>
<td>258</td>
<td>253 (97)</td>
<td>226 25</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td>656</td>
<td>613 (92.6)</td>
<td>555 59</td>
</tr>
</tbody>
</table>

Table 2: Accuracy of sonographic determination of fetal gender

<table>
<thead>
<tr>
<th>CRL (mm)</th>
<th>Male at birth (n)</th>
<th>Female at birth (n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>35.0-45.0</td>
<td>144 (89.3)</td>
<td>10 (6.7)</td>
</tr>
<tr>
<td>45.1-50.0</td>
<td>186 (99.4)</td>
<td>1 (0.5)</td>
</tr>
<tr>
<td>50.1-60.0</td>
<td>100 (98.1)</td>
<td>2 (2.0)</td>
</tr>
<tr>
<td>Total</td>
<td>430 (99.2)</td>
<td>12 (2.6)</td>
</tr>
</tbody>
</table>

(45.1-50.0): CRL of the fetus was 45.1 mm, (50.1-60.0): CRL of the fetus was 50.1 mm.
SECOND/THIRD TRIMESTER GENITALIA

MALE

FEMALE

HYDROCELE

URINARY TRACT ABNORMALITIES Classification

• Severe hypoplasia/agenesis
• Ectopia
• Dilatation/obstructive uropathy
  – UPJ obstruction
  – UVJ obstruction
  – bladder outlet obstruction
• Renal cystic disease
  – cystic dysplasia
  – multicystic dysplastic kidney disease
  – polycystic kidney disease
  – syndromes with cysts
• Tumors
• Abnormal genitalia

BILATERAL RENAL AGENESIS

BILATERAL RENAL AGENESIS
**Goals of the UTD Classification System**

- To propose a unified description of UT dilation that can be applied pre- and postnatally.
  - Simple but detailed enough to be meaningful for both clinical use and future research endeavors.
  - Allow for communication of information between specialists, providing consistent terminology.
- To propose standardized schema for the perinatal evaluation of these patients based on sonographic criteria.
  - Intended to be a starting point for observation and study.
  - Will be modified over time based on the accumulated evidence.

**Recommendation #1: Terminology**

- Discourage the use of non-specific terms in describing UT dilation (e.g., hydronephrosis, pyelectasis, pelviectasis, urenephrosis, UT fullness or prominence, pelvic fullness).
- Suggest the consistent use of the term "UT dilation".
- Further determination of the severity of UT dilation is characterized by specific sonographic findings, delineated by the UTD Classification System.

**Participants**

- **Prenatal**
  - Society for Maternal-Fetal Medicine
    - Anthony Odibo
    - Jude Crino
  - American Institute of Ultrasound in Medicine
    - Bryan Bromley
  - American College of Radiology
    - Beverly Coleman
  - Society of Radiologists in Ultrasounds
    - Carol Benson

- **Postnatal**
  - Society for Fetal Urology and Society for Pediatric Urology
    - Anthony Herndon
    - Jeffrey Campbell
    - Christopher Cooper
  - Society for Pediatric Radiology
    - Jeannie Chow
    - Kassa Darge
  - American Society of Pediatric Nephrology
    - Michael Somers
    - Deborah Stein
**Recommendation #3: Defining Normal**

<table>
<thead>
<tr>
<th>Ultrasound Findings</th>
<th>Time at Presentation</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>16-27w</td>
</tr>
<tr>
<td>Anterior posterior diameter</td>
<td></td>
</tr>
<tr>
<td>Central</td>
<td>&lt; 4mm</td>
</tr>
<tr>
<td>Peripheral</td>
<td>No</td>
</tr>
<tr>
<td>Parenchymal thickness</td>
<td>Normal</td>
</tr>
<tr>
<td>Parenchymal appearance</td>
<td>Normal</td>
</tr>
<tr>
<td>Bladder</td>
<td>Normal</td>
</tr>
<tr>
<td>Oligohydramnios</td>
<td>No</td>
</tr>
</tbody>
</table>

**Recommendation #3: Stratification of Risk**

- **Risk of what??**
  - Defined as the presence of postnatal urological pathology
- Further research will be needed to correlate risk stratification to clinical outcomes (UTI, pain, stone, need surgery, renal function, urological pathologies)
- Propose follow up recommendation based on risk stratification

**Prenatal Presentation**

- Central calyceal dilation with APRPD = 4 to < 7 mm (≤ 16-27 wk.)
- Central calyceal dilation with APRPD = 7 to < 10 mm (≥ 28 wk.)
- Central calyceal dilation with APRPD ≥ 7 mm (> 48h)

**Risk-Based Management, Prenatal Diagnosis**

- **Prenatal Period:** One additional US ≥ 32 weeks
  - After birth: Initially in 4 to 6 weeks*
    - US: ≥ 48 hrs to 1 month
    - ≥ 6 months later
    - Anomaly risk modifier: 4 included
- **Prenatal Period:** Two additional US ≥ 48 hrs to 1 month
  - ≥ 6 months later
  - Anomaly risk modifier: 4 included

* Certain situations (e.g., posterior urethral valves, bilateral severe hydrocephaly) may require more expedient follow up.
CAUSES OF URINE FLOW IMPAIRMENT

- UPJ anomaly
- UVJ anomaly
- Post urethral valves
- Duplex systems
- Ureterocele/ ectopic ureter
- Urethral atresia
- Cloacal anomaly
- Vesicoureteral reflux
- Megaureter
- Megacystis microcolon hypoperistalsis synd
- Sacrococcygeal teratoma
- Hydrometacolpos
- Other pelvic masses

DUPLEX SYSTEM

URETEROCELE

OBSTRUCTIVE UROPATHY Prognostic Factors

- Site of obstruction
- Degree of dilatation
- Cortical appearance
- Amniotic fluid volume
- Associated anomalies
- Urine biochemistry
OBSTRUCTIVE UROPATHY
Evaluation & Management

- Look for associated anomalies
- Offer karyotype
- Patient counseling
- Pediatric subspecialty consultation
- If urethral level obstruction (LUTO):
  - consider urine biochemistry
  - consider vesicoamniotic shunt

LUTO – SONOGRAPHIC FINDINGS

- Urinary tract dilation
  - renal pelves ≥ 10 mm
- Hydroureter
- Dilated bladder
  - thick wall
  - “keyhole sign”
- ± signs of renal dysplasia

LUTO – PATHOLOGIC FINDINGS

Potter, Pathology of the Fetus and the Newborn, 1952

HYDROURETER

LUTO – “KEYHOLE” BLADDER

Potter, Pathology of the Fetus and the Newborn, 1952
LUTO – RENAL FINDINGS

LUTO – URINE ASCITES WITH THICK-WALLED BLADDER

LUTO – EVALUATION
- Karyotype
  - amniotic fluid, fetal urine, fetal blood, chorionic vili
- Detailed sonography
- Serial urine testing
  - sodium, chloride, calcium, osmolality, total protein, β-2 microglobulin
  - at least 3 samplings 48-72 hrs apart

LUTO – BLADDER ASPIRATION

LUTO URINE VALUE THRESHOLDS
- Sodium < 100 mmol/L
- Chloride < 90 mmol/L
- Osmolality < 200 mOsm/L
- Calcium < 8 mg/dL
- β-2 microglobulin < 6 mg/dL
- Total protein < 20 mg/dL
OBSTRUCTIVE UROPATHY
Criteria for in utero Rx

- Lower urinary tract obstruction (LUTO)
- Normal male karyotype
- Oligohydramnios or ↓ AFV
- No other significant anomaly
- Improving fetal urine values

LUTO – CRITERIA FOR IN UTERO Rx
NORMAL MALE KARYOTYPE

VESICOAMNIOTIC SHUNTING IN FETAL LUTO: TECHNIQUE
**VESICOAMNIOTIC SHUNTING IN FETAL LUTO: PROBLEM AREAS**

- Natural history is highly variable
- Limited accuracy of antenatal assessment of prognostic factors:
  - etiology
  - renal function
- Procedure related complications
- Poor quality of available evidence of efficacy

**LUTO: CLINICAL EXAMPLE**

**LUTO: IS A STENT JUSTIFIED?**

- YES, with the following caveats:
  - antenatal assessment must be systematic and complete
  - patients must be thoroughly counseled and informed of both short term and long term outcomes
  - ideally should be included in a clinical trial or registry

**MULTICYSTIC DYSPLASTIC KIDNEY DISEASE**

- Complete proximal obstruction or atresia before 10 weeks
- Sonographic appearance
  - enlarged kidney, irregular contour
  - multiple cysts, various sizes
  - no communication between cysts

**MULTICYSTIC DYSPLASTIC KIDNEY DISEASE**

- Dysplastic kidney nonfunctional
- May diminish in size or disappear
- 40% contralateral abnormality
  - UPJ most common
- Prognosis
  - good if unilat, other kidney nml
  - fatal if bilateral
MULTICYSTIC DYSPLASTIC KIDNEY DISEASE

Pathologic findings

Autosomal recessive (infantile)
  - usually evident in utero
  - symmetrically enlarged, echogenic appearance
  - presentation depends on fraction of renal tubules affected
  - hepatic fibrosis inversely proportional to renal involvement

Gynecology and Obstetrics

Potter, Pathology of the Fetus and the Newborn, 1952

POLYCYSTIC KIDNEY DISEASE

autosomal recessive
POLYCYSTIC KIDNEY DISEASE

- Autosomal recessive

Potter, Pathology of the Fetus and the Newborn, 1952

• Autosomal dominant (adult onset)
  - commonly presents in young adults
  - occasionally seen in utero
  - enlarged, echogenic kidneys, ± cysts
  - family history crucial

AMBIGUOUS GENITALIA

- Genitalia not typical for male or female
  - Cannot differentiate penis from clitoris
  - Cannot differentiate scrotum from labia
    • Empty scrotum resembles labia
    • Fused labia resemble scrotum
  - Secondary structures rarely seen in fetus
    - Uterus, ovaries, undescended testes

AMBIGUOUS GENITALIA MORPHOLOGY

- Male
  - Hypospadias / epispadias
  - Micropenis
  - Chordee (ventral curvature of penis)
  - Cryptorchidism (undescended testes)
- Female
  - Clitoromegaly
  - Prominent or fused labia

AMBIGUOUS GENITALIA ETIOLOGY

- Congenital adrenal hyperplasia (CAH)
  - Treatable
- Female pseudohermaphroditism
  - 46,XX, fetal or maternal androgen source
- Androgen insensitivity syndrome
  - 46,XY, ↓ end organ testosterone effect
  - Complete – female external genitalia
  - Incomplete – ambiguous genitalia

AMBIGUOUS GENITALIA ETIOLOGY

- Mixed gonadal dysgenesis
  - 45,X/46,XY
- Pure gonadal dysgenesis
  - Variable karyotype
- True hermaphroditism
- Aneuploidy
- Duplication and deletion syndromes
**AMBIGUOUS GENITALIA EVALUATION**

- Determine genetic sex
  - cfDNA, amniocentesis
- Evaluate for aneuploidy, duplication and deletion syndromes
  - Karyotype, microarray
- Evaluate for CAH if virilized female
  - Molecular genetics, amniotic fluid 17 OHP
  - Maternal dexamethasone if affected female

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**HYPOSPADIAS**

- Urethral orifice on ventral side of penis
  - 50% anterior near glans
  - 30% middle
  - 20% posterior
- Blunt ended or bulbous penis
- “Tulip” sign – small penis between scrotal folds
- Other urogenital anomalies in 40%
HYPOSPADIAS

MEGALOURPETHRA

MEGALOURPETHRA

SCROTAL MASS

• Inguinal hernia
  – Indirect (bowel passes into scrotum through processus vaginalis)
  – Echogenic mass separate from testis
    • May see peristalsis – pathognomonic
• Testicular torsion
  – Variable echotexture, size, shape of testis
• Tumor (rare)

INGUINAL HERNIA

HYDROCOLPOS

• Vaginal obstruction
• Distension of vagina with secretions
• Unilocular retrovesicular cystic mass funneling to perineum
• Look for evidence of cloacal anomaly
HYDROCOLPOS

OVARIAN CYST
- Fetal ovarian response to maternal hormones
- Abdominal cyst in female fetus
  - Usually in lower abdomen / pelvis
  - Variable in size
  - May be simple, complex, septate
- GI and urinary tracts normal
- May resolve spontaneously
- Hemorrhage, torsion may occur

OVARIAN CYST
- Abdominal cyst in female fetus
  - Usually in lower abdomen / pelvis
  - Variable in size
  - May be simple, complex, septate
- GI and urinary tracts normal
- May resolve spontaneously
- Hemorrhage, torsion may occur

BLADDER EXSTROPHY
- Lower abdominal wall defect
- Exposed bladder
  - Soft tissue mass – posterior bladder wall
- Abdominal cord insertion at superior margin of exposed bladder
- Wide iliac wing angle, separated pubic symphysis
- Abnormal genitalia
  - Bifid penis, separated labia
CLOACAL MALFORMATION

- Complex malformation – failure of cloacal division
- Spectrum of abnormal anatomy
- Septated retrovesicular mass with fluid-fluid level
- Genitourinary, bowel, spine anomalies common
PROTOCOL FOR GENITOURINARY ANOMALIES

- Genitourinary system evaluation
  - measure renal pelves, renal lengths
  - assess renal appearance (contour, echogenicity, cysts)
  - demonstrate renal artery blood flow (color or power Doppler), consider pulsed Doppler study
  - image bladder in transverse and coronal/sagittal planes, measure bladder volume and/or wall thickness if appropriate

- Genitourinary system evaluation (cont’d)
  - look for ureteral dilatation
  - if kidneys or bladder are enlarged, measure fetal abdominal circumference at maximum level in addition to standard level
- Evaluate amniotic fluid volume
- Complete anatomic survey
- Fetal echocardiography
- Consultation as appropriate (genetic counseling, pediatric urology, neonatology)

THANK YOU