FETAL RENAL ANOMALIES
DISCLOSURES

- Fetal Renal Anomalies - Linda Street MD
- I have no financial relationships to disclose
- I will not discuss off label use and/or investigational use in my presentation
OBJECTIVES

- Discuss the components of the Genitourinary (GU) system
- Discuss basic fetal GU physiology and anatomy
- Discuss normal sonographic appearances of GU organs
- Discuss GU system abnormalities
  - Appearance on ultrasound
  - Causes
  - Management
GENITOURINARY SYSTEM (GU) COMPONENTS

- Kidneys
- Ureters
- Bladder
- Urethra
- Adrenal Glands
- Internal and External genitalia
- Fetal skin allows transudation of plasma into amniotic sac
- Urine produced as early as 9 weeks
- Kidneys do not significantly contribute to amniotic fluid volume prior to 16 weeks
Normal kidneys are identifiable trans-abdominally in most fetuses by 20 weeks gestation (as early as 10-12 weeks).

Normal kidneys generally span 4-5 vertebral bodies.

The length of normal kidney in mm is similar to gestational age in weeks.
KIDNEY ANATOMY
LONGITUDINAL VIEW
TRANSVERSE VIEW
TRANSVERSE VIEW
ECTOPIC KIDNEY LOCATIONS

- Thoracic
- Crossed
- Iliac
- Pelvic
PELVIC KIDNEY
PELVIC KIDNEY₁
RENAL ANOMALIES

- Abnormal Kidneys
  - Urinary tract dilation
  - Duplicated collecting system
  - Cystic kidneys
  - Absent Kidneys
  - Echogenic Kidneys
AP measurement of the renal pelvis

Normal varies by gestational age
- <4mm second trimester
- <7mm third trimester

Present in 3% fetuses (Very common, usually resolves)

Recently re-categorized to attempt to standardize terminology

URINARY TRACT DILATION
(PREVIOUSLY PYELECTASIS/HYDRONEPHROSIS)
# Urinary Tract Dilation

## What to Evaluate

<table>
<thead>
<tr>
<th>US parameters</th>
<th>Measurement /findings</th>
<th>Note</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anterior-Posterior Renal Pelvic Diameter (APRPD)</td>
<td>(mm)</td>
<td>Measured on transverse image at the maximal diameter of intrarenal pelvis</td>
</tr>
<tr>
<td>Calyceal dilation</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Central (major calyces)</td>
<td>Yes/No</td>
<td></td>
</tr>
<tr>
<td>Peripheral (minor calyces)</td>
<td>Yes/No</td>
<td></td>
</tr>
<tr>
<td>Parenchymal thickness</td>
<td>Normal/Abnormal</td>
<td>Subjective assessment</td>
</tr>
<tr>
<td>Parenchymal appearance</td>
<td>Normal/Abnormal</td>
<td>Evaluate echogenicity, corticomedullary differentiation, and for cortical cysts</td>
</tr>
<tr>
<td>Ureter</td>
<td>Normal/Abnormal</td>
<td>Dilation of ureter is considered abnormal</td>
</tr>
<tr>
<td>Bladder</td>
<td>Normal/Abnormal</td>
<td>Evaluate wall thickness, for the presence of ureterocele, and for a dilated posterior urethra</td>
</tr>
</tbody>
</table>
URINARY TRACT DILATION TERMINOLOGY

PRENATAL PRESENTATION

16-27 wks AP RPD
4 to <7mm
To <7mm

≥7mm
≥10mm

Central or no
calyceal dilation*
Parenchymal
thickness normal
Parenchymal
appearance normal
Ureters
normal
Bladder
normal
No unexplained
oligohydramnios

Peripheral
calyceal dilation*
Parenchymal
thickness abnl
Parenchymal
appearance abnl
Ureters
abnormal
Bladder
abnormal
Unexplained
oligohydramnios**

UTD A1;
LOW RISK

UTD A2-3;
INCREASED RISK

*Central and peripheral calyceal dilation may be difficult to evaluate
early in gestation
**Oligohydramnios is suspected to result from a GU cause
URINARY TRACT DILATION
UTD A1 (LOW RISK)
URINARY TRACT DILATION
UTD A2-3 (HIGH RISK)
## ETIOLOGY OF URINARY TRACT DILATATION DETECTED ON ANTENATAL ULTRASOUND

<table>
<thead>
<tr>
<th>Etiology</th>
<th>Incidence (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Transient/physiologic</td>
<td>50–70</td>
</tr>
<tr>
<td>Ureteropelvic junction obstruction</td>
<td>10–30</td>
</tr>
<tr>
<td>Vesicoureteral reflux</td>
<td>10–40</td>
</tr>
<tr>
<td>Ureterovesical junction obstruction/megaureter</td>
<td>5–15</td>
</tr>
<tr>
<td>Multicystic dysplastic kidney disease</td>
<td>2–5</td>
</tr>
<tr>
<td>Posterior urethral valves</td>
<td>1–5</td>
</tr>
<tr>
<td>Ureterocele, ectopic ureter, duplex system, urethral atresia, Prune belly syndrome, polycystic kidney diseases</td>
<td>Uncommon</td>
</tr>
</tbody>
</table>
URINARY TRACT DILATION
UTD A2-3 (HIGH RISK)
URINARY TRACT DILATION
UTD A2-3 (HIGH RISK)
If other findings or risk factors are present, the finding of UTD warrants adjusting T21 risk and offering prenatal testing

- Likelihood ratio=1.3 for Trisomy 21

In a low risk population, UTDA1 is likely a normal variant and doesn’t significantly change aneuploidy risk.
URINARY TRACT DILATION MANAGEMENT

RISK-BASED MANAGEMENT, PRENATAL DIAGNOSIS

UTD A1: LOW RISK

PREGNATAL PERIOD:
One additional US ≥ 32 weeks

AFTER BIRTH:
Two additional US:
1. > 48 hrs to 1 month
2. 1-6 months later

OTHER:
Aneuploidy risk modification if indicated

UTD A2-3: INCREASED RISK

PREGNATAL PERIOD:
Initially in 4 to 6 weeks*

AFTER BIRTH:
US at > 48 hours to 1 month of age*

OTHER:
Specialist consultation, e.g. nephrology, urology

* certain situations (e.g. posterior urethral valves, bilateral severe hydronephrosis) may require more expedient follow up
URINARY TRACT OBSTRUCTION

- Urethral obstruction
- UVJ obstruction
- UPJ obstruction
- Ureterocele
- Urethral obstruction
URETERO-PELVIC JUNCTION (UPJ) OBSTRUCTION-LONGITUDINAL
URETERO-PELVIC JUNCTION (UPJ) OBSTRUCTION-LONGITUDINAL
URETERO-PELVIC JUNCTION (UPJ) OBSTRUCTION-TRANSVERSE
URETERO-VESICULAR JUNCTION (UVJ) OBSTRUCTION

The normal fetal ureter is NEVER visible!
URETERO-VESICULAR JUNCTION (UVJ) OBSTRUCTION
URETEROCELE
(CAN CAUSE UVJ OBSTRUCTION)
FETAL BLADDER PHYSIOLOGY/ANATOMY

- Fetal bladder fills and empties every 30-155 minutes
- Fetal bladder wall not usually seen when normal bladder is distended
Etiology:
- Posterior urethral valves (Males)
- Urethral atresia
- Other: Megacystis-microcolon, cloacal malformations

Sonographic findings:
- Bilateral UTD
- Hydroureter
- Distended bladder
- Oligohydramnios
BLADDER OUTLET OBSTRUCTION

“Keyhole Bladder”
A. Transverse UTDA2-3
B. Longitudinal UTDA2-3 and hydroureter
C. Transverse thickened bladder wall
D. “Keyhole sign” and thickened bladder wall
Multicystic dysplastic kidneys
Cystic dysplasia

Autosomal recessive polycystic kidney disease (ARPKD)
Autosomal dominant (ADPKD)
MULTICYSTIC DYSPLASTIC KIDNEY

- Result of an EARLY (first trimester) urinary tract obstruction
- 70-80% unilateral
- Cysts vary in size
- No communication between cysts, no uniformity
- Reniform contour is lost
- Non-functioning kidney
- 25% of the time other anomalies are present
MULTICYSTIC DYSPLASTIC KIDNEY
MULTICYSTIC DYSPLASTIC KIDNEY
MULTICYSTIC DYSPLASTIC KIDNEY

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BILATERAL MULTICYSTIC DYSPLASTIC KIDNEYS

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Result of a LATE (second/third trimester) urinary tract obstruction

Common causes

- UPJ obstruction
- Bladder outlet obstruction

Results in irreversible renal damage

Usually few small cysts with a small, echogenic kidney
CYSTIC DYSPLASIA$_3$
Autosomal recessive mutation of the PKHD1 gene, locus 6p

Symmetric enlargement of both kidneys with innumerable 1-2 mm cysts

+/- hepatic involvement

Typically diagnosable by 24 weeks but varies widely and may not present until postnatal period

Sonographic findings
- Progress with advancing gestational age
- Loss of corticomedullary differentiation
- Low amniotic fluid
AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE - LONGITUDINAL

- 19 weeks
- 24 weeks
- 32 weeks
- 38 weeks
AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE - TRANSVERSE
85% due to PKD1 gene mutation on chromosome 16.
- Can also be due to PKD2 gene mutation
- Severity of parental disease not predictive of offspring disease
- Cystic echogenic kidneys with NORMAL amniotic fluid volume
- Systemic disorder
  - Liver, pancreas, intracranial aneurysms, colonic diverticulae
AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE

- Difficult to differentiate from ARPKD
  - Assess family history
  - Get renal ultrasound on both parents
    - If normal, consider ARPKD
ECHOCgenic KIDNEYS

- Differential Diagnosis (partial)
  - Normal variant
  - Cystic kidney disorders (previously discussed)
  - Bardet Biedl syndrome
  - Meckel Gruber syndrome
  - Trisomy 13 and 18
  - Beckwith-Wiedemann syndrome
ECHOCGENIC KIDNEYS
Unilateral or bilateral

Bilateral renal agenesis
  - M:F ratio 2.5:1
  - 100% mortality (?)

Sonographic findings
  - Oligo/anhydramnios
  - Bladder not visible
  - Kidneys not visible
  - Other anomalies common

Absent Kidneys
UNILATERAL RENAL AGENESIS
BILATERAL RENAL AGENESIS
BILATERAL RENAL AGENESIS

“LYING DOWN ADRENAL” SIGN

Normal

Renal agenesis
BILATERAL RENAL AGENESIS

KIDNEYS

NO RENAL ART SEEN

AO
Upper pole ureter typically implants lower on the bladder, and is associated with a ureterocele.
DUPLICATED COLLECTING SYSTEM
REFERENCES


QUESTIONS?