Developmental Abnormalities of the Kidneys and GU System

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Session Summary

This presentation will provide participants with an overview of the GU system during various stages of fetal development, as well as the identification and treatment options for common renal and other GU abnormalities.

Session Objectives

Upon completion of this presentation, the participant will be able to:

- identify the most common developmental abnormalities of the kidneys;
- manage antenatal hydronephrosis after birth;
- understand the fetal development of the GU system.

References


Session Outline

See presentation handout on the following pages.
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Morphogenesis
- Differentiation starts around 5 weeks of gestation
- First nephrons formed by 8 weeks
- Nephrogenesis continues up to 34-35 weeks
- Kidney and bladder seen on US at 15 weeks
- Precise renal architecture seen at 20 weeks

Unilateral Renal Agenesis
- Isolated, sporadic finding
- 1/500-1/800 live births
- Look for other anomalies:
  - Cleft palate
  - Preauricular pits
  - Cardiac and vertebral defects
  - Müllerian duct aplasia
- If normal growth at 1 yr, no further evaluation

Polycystic Kidneys
- Autosomal Dominant and Recessive forms
- Many cysts in both kidneys
- No evidence of renal dysplasia
  - Continuity of the lumen of the nephron with the uriniferous space to the urinary bladder
- Tuberous sclerosis may have cystic kidneys

Bilateral Renal Agenesis
- 1/3000 births
- Potter Syndrome
  - Potter facies
  - Limb deformities
  - Pulmonary hypoplasia
  - Anuria/oligohydramnios

Palpable abdominal mass
- 0.2-0.6% of infants at birth
- 2/3 of all cases due to renal masses
  - Top 3 causes: polycystic, multicystic, hydronephrosis
  - Remainder: GI, adrenal, liver, female reproductive tract
- Midline masses
  - Bladder obstruction
  - Neurogenic bladder
Autosomal Recessive Polycystic Kidney Disease (ARPKD)

- 1/16000
- Variable expression
- Chromosome 6p
- Affects liver and kidney
- Protuberant abdomen, large palpable kidney
- Severe hypertension

ARPKD

- U/S
  - Large kidney
  - Increased echogenicity of the parenchyma
  - Loss of corticomedullary differentiation
  - Loss of central echo complex
  - Cortex preserved
  - Liver echodense with dilated biliary ducts

ARPKD

- Diagnosed after 24 weeks gestation
  - Large hyperechogenic kidneys, oligohydramnios, empty bladder
- If symptomatic at birth, death due to respiratory or renal causes
- 75% of infants who survive to 1 year can live for more than 15 years

ARPKD

- 1-3/100,000 live births
- Variable expression
- Chromosome 16 or 4
- Rarely manifests with clinical findings at birth

Autosomal Dominant Polycystic Kidney Disease
ADPKD

Prenatal DX of ADPKD

- Oligohydramnios sequence, enlarged kidneys and hematuria
- Endocardial fibroelastosis
- Intracerebral vascular anomaly
- Pyloric stenosis
- Hepatic fibrosis
- If manifesting in utero/first few months of life, ~1/2 cases died

Tuberous Sclerosis

- Polycystic or unilateral cystic disease
- Cysts identical to cysts of ADPKD
  - TS linked to marker gene near locus for PKD on chromosome 16
- Need to look for ash leaf nevi under UV light
- May develop end stage renal failure

Hydronephrosis

Evaluation Hydronephrosis

- Ultrasound
  - After birth and about 4-6 weeks of age
- VCUG
  - If abnormal US, may not be needed in mild hydronephrosis
- BUN/Cr
  - If severe prenatal or single kidney noted
- Prophylactic antibiotics
  - dependent on institutional preference and severity of hydronephrosis
# Obstructive Uropathy

- Ureteropelvic Junction Obstruction
- Multicystic Dysplastic Kidney
- Ureterovesical Junction Obstruction
- Posterior Urethral Valves
- Ureterocele
- Ectopic Ureters
- Prune Belly Syndrome

# UPJ Obstruction

- Most common location for obstruction
- U/S dilated renal pelvis, no dilated distal ureter
- Appearance does not predict function
- DTPA renal scan to show function of kidney

## UPJ Obstruction

- Prenatal suspicion
  - 1/3 obstructed and require immediate intervention
    - Pyeloplasty
  - 1/3 normal postnatal evaluation
  - 1/3 observed with serial U/S and renal scans
    - Up to 1/3 of this group may require surgical correction

## UPJ obstruction

- Pyeloplasty

## Ureterovesical Junction Obstruction

- 15% of all cases of prenatal hydronephrosis
- Peviectasis, caliectasis, and ureteral dilatation
- VCUG to r/o reflux
- DTPA scan for kidney function
- Does not always require surgery

## Multicystic Dysplastic Kidney

- 1/4300 live births
- Sporadic, nonsyndromal, congenital anomaly
- Multiple cysts, roughly equal size
  - “bunch of grapes” pattern
  - Unilateral
**Multicystic Dysplastic Kidney**

- Hypertrophy of contralateral kidney
- Affected kidney no function on DTPA scan
- If one large, centrally located cyst think of UPJ obstruction
- Cysts involute over time
- Occasional reports of malignancy

**Posterior Urethral Valves**

- Wide spectrum of presentation
- Renal U/S
  - Severe b/l hydroureteronephrosis
  - Thick walled bladder
  - Dilated posterior urethra

**Posterior Urethral Valves**

- VCUG
  - Dilated posterior urethra
  - Valve cusps at the distal aspect of the prostatic fossa
  - Heavily trabeculated bladder
  - High-grade vesicoureteral reflux

**Posterior Urethral Valves**

- Treatment
  - Decompression with urinary catheter
    - May result in post-obstructive diuresis
  - Antibiotic prophylaxis
  - Ablation of valves
  - Occasional vesicostomy
  - Up to 30% of pt require renal replacement therapy during lifetime
    - Poor urinary concentrating ability
**Ureterocele**

- Cystic dilatation of distal ureter and protrudes into bladder
- Found primarily in duplex systems
  - Always associated with upper pole
- U/S
  - Hydronephrosis with upper pole of duplex system
  - Ureteral dilatation down to level of the bladder
  - Cystic lesion within the lumen of the bladder

**Ectopic Ureter**

- Most detected on prenatal U/S
- Male never incontinent
  - Ureter always enters above level of external sphincter
- Female
  - Ectopic ureter to urethra or vaginal vault
- Most found in duplex systems

**Ureterocele**

- VCU
- Can prolapse into urethra
  - Most common form of bladder outlet obstruction in female neonates
- Surgical options
  - Endoscopic puncture
  - Upper pole partial nephrectomy
  - Excision of ureterocele with combined reimplantation

**Ectopic Ureter**

- U/S
  - Hydronephrosis - usually upper pole of duplex system
  - Dilated distal ureter that lies behind the bladder
  - Extension of dilated distal ureter past the bladder neck
- VCUG ~70% show reflux into ectopic ureter
Ectopic Ureter

• Surgical options
  – Partial nephrectomy
  – Complete nephrectomy
  – Cutaneous ureterostomy
  – Complete reconstruction with reimplantation

Prune-Belly Syndrome

• Massively dilated upper urinary tract and bladder
• Floppy, thin abdominal wall
• Bilateral undescended testes
• Flattened diaphragm with flaring of ribs
• Often with significant renal dysplasia

Prune-Belly Syndrome

• U/S
• No VCGU – if voiding on own, otherwise just increases urosepsis
• Antibiotic prophylaxis
• May require vesicostomy
• Likely to develop renal failure and require dialysis/transplant

Bladder Exstrophy

• Bladder plate protruding in the suprapubic area
• Males have associated complete epispadias
• Lack of formation of bladder neck
• Not associated with other anomalies
• Staged v. primary repair
**Hypospadias**

- Urethral meatus is not present in the normal glandular position
- Asymmetrical or hooded foreskin
- Tethered penis creating a chordee
- Foreskin is used in reconstruction
- Up to 10% of pt. may have an intact foreskin, only noticing after circumcision

**Hypospadias**

- Always check to see if testes are present to r/o intersex state
- Undesended teste or inguinal hernia occur in about 9% of patients
- Outpatient evaluation within 1 month
- Surgical correction after 6 months of age

**Hypospadias**

- First degree
  - Glandular or coronal
  - 50-75%
- Second degree
  - Shaft
  - 20%
- Third degree
  - Perineal
  - 30%

**Renal Failure**

- May be caused by
  - Renal dysplasia
  - Bilateral renal agenesis
  - Polycystic kidneys
  - Posterior urethral valves
  - Vesicoureteral reflux
  - Ureterocele
  - UPJ obstruction

**Renal Failure**

- Dialysis- hemo or peritoneal
- Transplant
  - No minimum age
  - At least 6kg, prefer 10kg
  - Graft 5 year survival rate about 80%
  - Younger infants have more vascular thrombotic events