

# Congenital Anomalies of the Kidney and Urinary Tract

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## Editors:

Micah A. Jacobs, MD, MPH

## Authors:

Vijaya Vemulakonda, MD, JD; Matthew D. Timberlake, MD

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## 1. Introduction

Approximately 30% of all congenital anomalies involve the kidney and urinary tract (CAKUT).<sup>1</sup> These anomalies account for nearly 50% of end stage renal disease (ESRD) in children.<sup>2</sup> The goal of this chapter is to discuss urinary tract embryology, as well as the most common CAKUT diagnoses and their management. While the embryology of the kidney and bladder are dealt with elsewhere (Core Curriculum section **Kidney, Adrenal, Ureter**), **Table 1** and **Table 2** summarize key concepts pertinent to this section.

Congenital anomalies of the kidney and urinary tract are classified as follows (**Table 3**):

- (i) Renal parenchymal anomalies
- (ii) Collecting system anomalies
- (iii) Renal ascent and fusion anomalies
- (iv) Lower urinary tract obstruction anomalies.

**Table 3** presents a summary of these abnormalities.

### 1.1 Key words

Renal dysplasia, multicystic dysplastic kidney, polycystic kidney disease, renal agenesis, hydronephrosis, ureteropelvic junction obstruction, megaureter, megacalycosis, ectopic ureter, ureterocele, duplex kidney, vesicoureteral reflux, ectopic kidney, horseshoe kidney, crossed fused renal ectopia, posterior urethral valves, anterior urethral valves, urethral atresia, prune belly syndrome.

**Table 1: Renal Development**

<i>Pronephros 4<sup>th</sup> week</i>	1) mature excretory structure in primitive vertebrates 2) no function in humans 3) in absence mesonephros will not develop
<i>Mesonephros 5<sup>th</sup>-12<sup>th</sup> weeks</i>	1) permanent kidney in amphibians 2) may function transiently in mammals 3) involutes by 12 weeks with exception of ducts in males and vestigial remnants in females ( <b>Table 2</b> )
<i>Metanephros 5<sup>th</sup> week onward</i>	1) forms when the ureteral bud appears on the lower end of the mesonephric duct 2) becomes the nephron-glomerulus, proximal convoluted tubule, loop of Henle and distal convoluted tubule 3) urine formation begins at 8 weeks
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**Table 2: Adult derivatives and vestigial remnants of embryonic urogenital structures**

Male	Embryonic Structure	Female
<i>Testis</i> <i>Seminiferous tubules</i> <i>Rete Testis</i>	<b>Indifferent Gonad</b> <b>Cortex</b> <b>Medulla</b>	<i>Ovary</i> <i>Ovarian follicles</i> <i>Medulla</i>
Ductuli Efferentes Paradidymis	<b>Mesonephric Tubules</b>	Epoophoron Paroophoron
<i>Appendix of epididymis</i>  <i>Ductus Epididymis</i>  <i>Ductus Deferens</i>  <i>Ureter, pelvis, calyces, and collecting tubules</i>  <i>Ejaculatory duct and seminal vesicle</i>	<b>Mesonephric Duct</b>	Duct of epoophoron Gartner's Duct <i>Ureter, pelvis, calyces, and collecting tubules</i>
Appendix of testis	<b>Paramesonephric Duct</b>	<i>Uterine tube</i> <i>Uterus</i> <i>Vagina</i>
<i>Urinary Bladder</i> <i>Urethra</i> <i>Prostatic utricle</i>	<b>Urogenital Sinus</b>	<i>Urinary Bladder</i> <i>Urethra</i> <i>Vagina</i>

<i>Prostate gland and bulbourethral glands</i>		<i>Urethral and paraurethral glands</i>
<i>Penis</i>	<b>Phallus</b>	<i>Clitoris</i>
<i>Ventral aspect of penis</i>	<b>Urogenital Folds</b>	<i>Labia Minora</i>
<i>Scrotum</i>	<b>Labioscrotal Swellings</b>	<i>Labia Majora</i>
<i>Italics denote functional derivatives of embryonic structure</i>		
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**Table 3: Summary of congenital anomalies of the kidney and urinary tract**

	Epidemiology	Characteristics	“Pearls” to remember
<b>Renal parenchyma anomalies</b>			
Renal dysplasia	3/1000 births	Many different appearances	Often associated with collecting system anomalies.
MCDK	~1/1000 births	Multiple non-communicating cystic structures. Most involute.	Confirm diagnosis with serial ultrasound or DMSA scan postnatal. Not associated with increased Wilm's risk.
ADPKD	1 in 400 to 1000 people	Large cysts. Progressive. Autosomal dominant. Renal failure in adulthood (50s – 60s) common.	In a child with 1-2 simple cysts, ADPKD is possible diagnosis.
ARPKD	1 in 10000 to 40000 births	Small cysts. Autosomal recessive. Often fatal.	Cysts arise from collecting ducts.

Renal agenesis	1 in 3000 to 5000 births	Lack of renal parenchyma. Failure of ureteral bud formation.	Often with ipsilateral Wolffian (males) or Mullerian (females) abnormalities.
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### Collecting system anomalies

Hydronephrosis	1 to 5% of all pregnancies	Dilation of renal collecting system. Can be associated with thinning of parenchyma.	Transient or physiologic hydronephrosis most common diagnosis. Severity correlates with outcomes.
UPJO	1 in 500 to 1000 births	Hydronephrosis. Delayed drainage on diuretic renography often with decreased differential function.	Indications for treatment include worsening split renal function, UTIs, pain, nephrolithiasis. Crossing vessels are more common in older children than in infants
Megaureter	0.4 in 1000 births	Large ureter > 7 mm. Classified according to presence or absence of reflux and/or obstruction.	Majority of primary megaureters resolve with time and differential function of 50% predicts resolution. Similar indications for surgery as

			UPJO.
Megacalycosis	Rare	Increased number of calyces that are dilated, medullary pyramid hypoplasia, and drainage on diuretic renography.	Consider diagnosis over UPJO when calyces are increased and dilated, there is drainage but it is delayed, and the renal pelvis is small in proportion to calyceal dilation.
Ectopic ureter	Less than 1 in 1000 births	Dilated ureter with varying degree of renal dysplasia in associated renal moiety. Often associated with upper pole of duplex kidney.	Management is quite variable but surgery is generally needed ( <b>Table 4</b> )
Ureterocele	1 in 1000 births	Cystic dilation of distal ureter in bladder. Often associated with upper pole of duplex kidney.	Consider function of renal segment and ipsilateral lower pole reflux in management decisions.
Duplex kidney	1-5% of population	Separate ureter draining upper and lower renal poles. Can be partial or complete.	The way the mesonephric duct and ureteral buds are incorporated into the bladder explains the

			Meyer-Weigert rule.
VUR	30% of children with febrile UTI	Retrograde flow of urine from bladder into upper tracts due to failure of adequate mucosal coaptation and closure of UO during voiding.	Voiding dysfunction should be treated prior to surgical intervention. Most low grade VUR resolves.

### Renal migration or fusion anomalies

Ectopic kidneys	1 in 500 to 1200 births	Kidney in ectopic location along route of ascent. Pelvic kidney is most common.	Increased risk of collecting system anomalies such as VUR and UPJO. Anomalous blood supply.
Horseshoe kidney	1 in 600 births	Inferior poles of kidneys fused prior to renal ascent and ascent limited by inferior mesenteric artery.	Increased risk of collecting system anomalies such as VUR and UPJO. Anteriorly oriented pelvis with often medial facing calyces. Anomalous blood supply.
Crossed fused ectopia	1 in 1000 to 2000 births	Both kidneys on same side and fused.	Ureter from ectopic kidney crosses back to other side. Anomalous blood supply.

Crossed unfused ectopia	Rare	Only 5% of crossed renal ectopia.	Ureter from ectopic kidney crosses back to other side. Anomalous blood supply.
Bilateral crossed renal ectopia	Rare	Both kidneys are on wrong side.	Ureters from each kidney cross to other side.

### Lower urinary tract obstruction anomalies

Posterior urethral valves	1 in 5000 to 8000 births	Obstruction at posterior urethra by membranous folds. Upper tract damage and bladder dysfunction.	These patients need long term monitoring of bladder and renal function. High incidence of renal failure.
Anterior urethral valves	Less than 300 reported cases	Characteristic web of tissue from ventral anterior urethra. Often associated with urethral diverticulum.	Endoscopic ablation is usually initial treatment. Occasionally urethroplasty needed if diverticulum. Has similar long term concerns as PUV.
Urethral atresia	Very rare	Failure of urethral cannulation.	Fetal survival associated with patent urachus or other method of urine drainage.

Prune belly syndrome	3-4 in 100,000 births	Abdominal wall muscle deficiency with “prune” appearance, bilateral undescended testis, GU tract dilation.	GU tract dilation generally worse distally. Need for CIC because of urinary retention with UTIs is not uncommon. Abdominoplasty controversial.
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## 2. Renal Parenchymal Anomalies

The term renal dysplasia refers to anomalies of renal parenchymal development, characterized histologically by fibrosis, immature tubules, and metaplastic tissue. Often there are associated collecting system abnormalities. A spectrum of clinical diagnoses may result, including renal dysplasia or hypoplasia, multicystic dysplastic kidney, polycystic kidney disease, and renal agenesis.

### 2.1 Renal Dysplasia



Figure 1: Typical appearance of MCKD on renal ultrasound (left) and renal scan (right). Note multiple non-communicating cysts of varying sizes and lack of radiotracer uptake indicating no functional renal tissue.

Renal dysplasia affects **3/1000** infants with a male predominance.<sup>3</sup> Collecting system anomalies such as vesicoureteral reflux (AUA guideline on **Management and Screening of Primary Vesicoureteral Reflux in Children**) and ectopic ureters are seen at a higher frequency when renal dysplasia is present. The presence of dysplasia is related to the magnitude of ureteral orifice deviation from the orthotopic location. As a result, kidneys with ectopic ureters or high-grade reflux are more likely to have dysplasia, and consequently, diminished function. A **multicystic dysplastic kidney (MCDK)** is the consequence of severe renal dysplasia and is characterized by a non-reniform shaped kidney composed of non-communicating cysts, no functional renal tissue, and an atretic or absent ureter (**Figure 1**). **MCDK occurs with a frequency of 0.3-1/1000 births.**<sup>4</sup> Unilateral renal dysplasia or MCDK can result in compensatory hypertrophy of the contralateral kidney. **Most MCDK kidneys involute with time and do not require surgical intervention.** Confirmation of diagnosis postnatally with characteristic appearance and involution on serial ultrasounds and no appreciable renal function on DMSA scan is often recommended. **Historical concerns about an increased risk of malignancy in MCDK are not currently supported.**<sup>5,6</sup>

### 2.2 Autosomal Dominant Polycystic Kidney Disease (ADPKD)

ADPKD is the most common cystic kidney disease with an **incidence of 1/400-1000 infants**. The

**PKD1 or PKD2 genes** encode the polycytin-1 and polycytin-2 proteins, respectively. A mutation in PKD1 (~85% of cases) or PKD2 leads to ADPKD.<sup>7</sup> Clinical features in children with ADPKD ranges from normal appearing kidneys to numerous bilateral renal cysts, and potentially, **hypertension, hematuria, and infection**.

## 2.3 Autosomal Recessive Polycystic Kidney Disease (ARPKD)

ARPKD is less common than ADPKD, with **an incidence of 1/10,000-40,000** infants, and is classically associated with renal cystic disease which is detectable early in life. **Hepatic cysts are often present.** Mutations in the **PKHD1 gene** that encode the protein fibrocystin thought to be causative. ARPKD can cause in utero or perinatal demise due to oligohydramnios and pulmonary hypoplasia, end stage renal disease in early life requiring dialysis, or, relatively normal renal function with delayed diagnosis in adulthood.<sup>8,9</sup> **Cysts in ARPKD tend to be smaller (millimeters), arise from the collecting duct, and appear early in life.** On the other hand, cysts in autosomal dominant disease tend to be larger (centimeters), involve the entire nephron, and appear later in life.

## 2.4 Renal Agenesis

Renal agenesis describes the absence of renal parenchymal tissue unilaterally, occurring at a frequency of **1/3000-5000 births**.<sup>1,10</sup> It is thought to result from a **failure of ureteral bud formation**. Males often have associated abnormalities of the vas deferens, seminal vesicles, and epididymis (but a normal ipsilateral testis). Females have an increased risk of ipsilateral Mullerian abnormalities such as uterine didelphys, duplicated vagina, and potentially obstructed ipsilateral hemivagina.<sup>11</sup> **In both males and females, the presence of genital duct abnormalities should prompt renal ultrasound to look for ipsilateral renal agenesis.** Bilateral renal agenesis has been traditionally considered a lethal anomaly; however, studies to assess the effect of in utero amnio-infusion on survival to renal transplantation in patients with prenatally diagnosed bilateral renal agenesis or functional renal agenesis are ongoing.

# 3. Collecting System Anomalies

Anomalies of the renal pelvis and ureter are often detected during routine prenatal care with the finding of hydronephrosis on ultrasound. **We begin by addressing the etiologies of hydronephrosis without hydroureter including ureteropelvic junction obstruction (UPJO) and megalocalycosis.** We then address etiologies of hydroureteronephrosis primary megaureter, ectopic ureter, pyeloureteral duplication, ureterocele, and vesicoureteral reflux.

## 3.1 Hydronephrosis

See Core Curriculum **Pediatrics: Hydronephrosis**

Hydronephrosis is detected prenatally during routine ultrasound screening in about **1% of pregnancies (Figure 2)**.<sup>12,13</sup> Most cases of antenatal hydronephrosis are mild and either resolve with time (transient hydronephrosis) or stabilize (non-obstructive hydronephrosis) and do not require any intervention.<sup>14</sup> The Society of Fetal Urology (SFU) grading system is a way to report the severity of

hydronephrosis.<sup>15</sup> In general, SFU grade 1-2 is considered mild and SFU grade 3-4 is considered severe. Severe antenatal hydronephrosis is more likely to be associated with need for intervention. Hydronephrosis may be obstructive or non-obstructive. Obstructive etiologies of hydronephrosis include, UPJO, ectopic ureter, and ureterovesical junction obstruction (also referred to as primary obstructed megaureter). Non-obstructive etiologies include transient nonpathologic dilation, vesicoureteral reflux (VUR), adynamic ureterovesical junction, and prune belly syndrome.

### 3.2 UPJ Obstruction

UPJO is the most common pathologic etiology of prenatal hydronephrosis, occurring in 1/500-1000 birth (**AUA Update Series: Current Management of Ureteropelvic Obstruction in Adults and Children**).<sup>14,16</sup> It is thought to be caused by either an **intrinsic functional or anatomical stenosis of the UPJ** (most often seen in infants) or by **extrinsic compression by a lower pole renal vessel** that crosses the pelvis and obstructs drainage (seen more frequently in older children). Diuretic renography is used to confirm the diagnosis of UPJO and can also be predictive of the need for surgical management with pyeloplasty (**Figure 3**). If the differential renal function is near 50%-50% on diuretic renography (indicating lack of renal functional loss due to obstruction), an observational approach may be indicated.<sup>17</sup> Indications for pyeloplasty include **worsening differential renal function, worsening hydronephrosis** especially with development of thinning parenchyma, **urinary tract infections, pain, or nephrolithiasis**.

### 3.3 Megacalycosis

Megacalycosis is a rare diagnosis of unknown etiology characterized by increased number of calyces, dilation of calyces without obstruction, and medullary pyramid hypoplasia (**Figure 4**).<sup>18</sup> It is usually detected on prenatal ultrasound with postnatal evaluation revealing increased number of dilated renal calyces on ultrasound and lack of obstruction on diuretic renography.

### 3.4 Megaureter or Hydroureter

See Core Curriculum **Pediatrics: Hydroureteronephrosis**

Megaureter is a descriptive term for a dilated ureter measuring **> 7 mm in diameter**. This finding occurs in **0.4/1000 births**.<sup>19,20</sup> Causes of megaureter or **hydroureteronephrosis** are discussed in more detail in the AUA core curriculum section on hydroureteronephrosis. The presence of VUR and/or obstruction are used to classify megaureters into 4 types (**Table 4**). The most common type is the **non-refluxing non-obstructed megaureter (primary megaureter)**. A **refluxing non-obstructed megaureter is often seen with Grade 5 VUR** and the other two types of megaureters (**non-refluxing obstructed** and **refluxing obstructed**) are relatively rare.

Non-refluxing non-obstructed megaureter, or primary megaureter, is thought to be caused by a defect in the collagen and muscle development in the distal ureter where there is an aperistaltic segment.<sup>21</sup> Diuretic renography is used to assess for obstruction and renal function (**Figure 5**). If the split function is equal, there is a high likelihood that the megaureter will improve or resolve with time.<sup>22,23</sup>

**Table 4: Classification of Megaureters**

<b>Refluxing, Nonobstructed Megaureter</b>	<b>Nonrefluxing, Obstructed Megaureter</b>
Primary (congenital reflux)	Primary (adynamic segment)
Secondary (urethral valves, neurogenic bladder)	Secondary (urethral obstruction, extrinsic mass, or tumor)
<b>Nonrefluxing, Nonobstructed Megaureter</b>	<b>Refluxing, Obstructed Megaureter</b>
Primary (idiopathic, physiologically insignificant adynamic segment)	Occurs with ectopic ureter to urethral sphincter. Reflux during voiding.
Secondary (polyuria, infection, postoperative residual dilation)	
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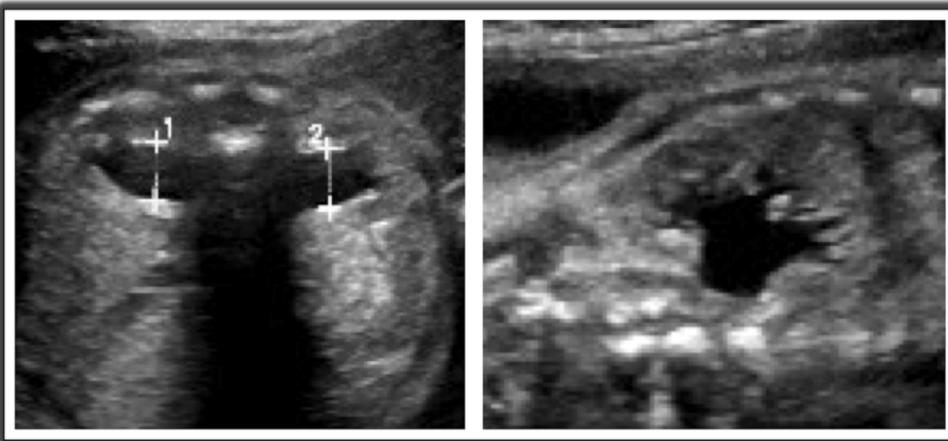


Figure 2: Images of bilateral antenatal hydronephrosis in a 28 week old fetus. The anterior posterior renal pelvis diameter (10 mm, left) and SFU grade (Grade 3, right) can be assessed.

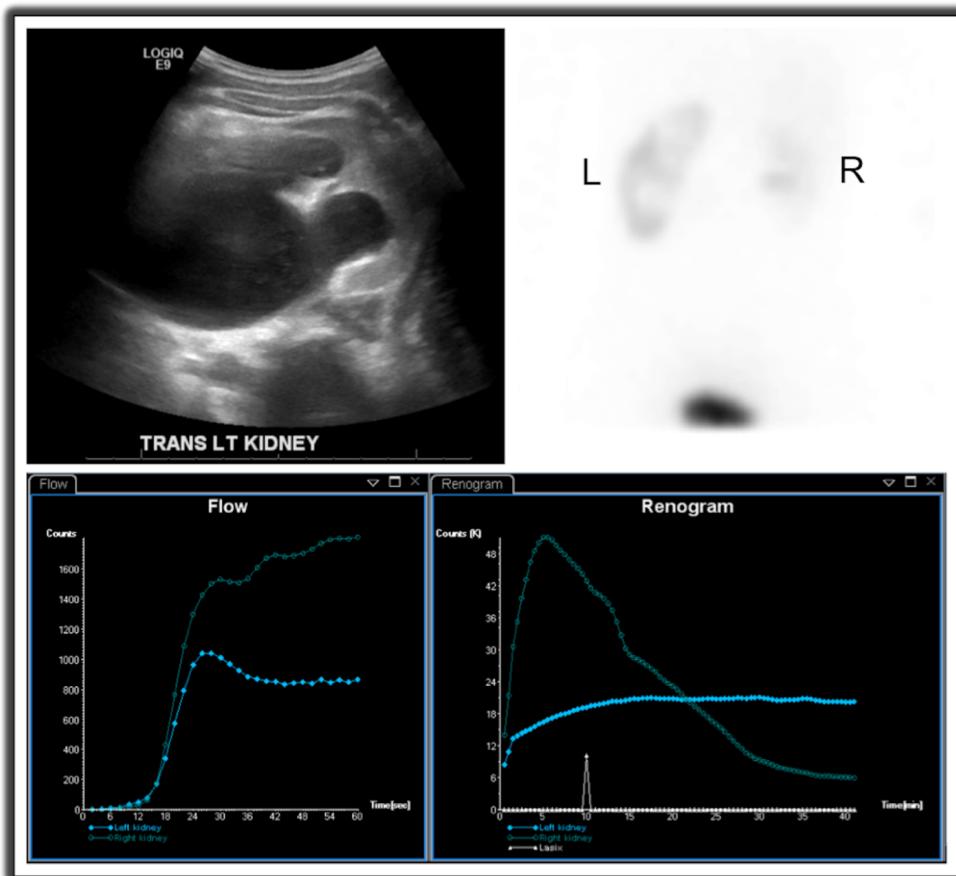


Figure 3: Images of left UPJO. SFU grade 4 hydronephrosis on ultrasound (upper left). Right kidney almost completely drained and left kidney with minimal excretion of radiotracer at 30 minutes (upper right). Decreased function and poor drainage of radiotracer on graphical representation of diuretic renography results (lower).

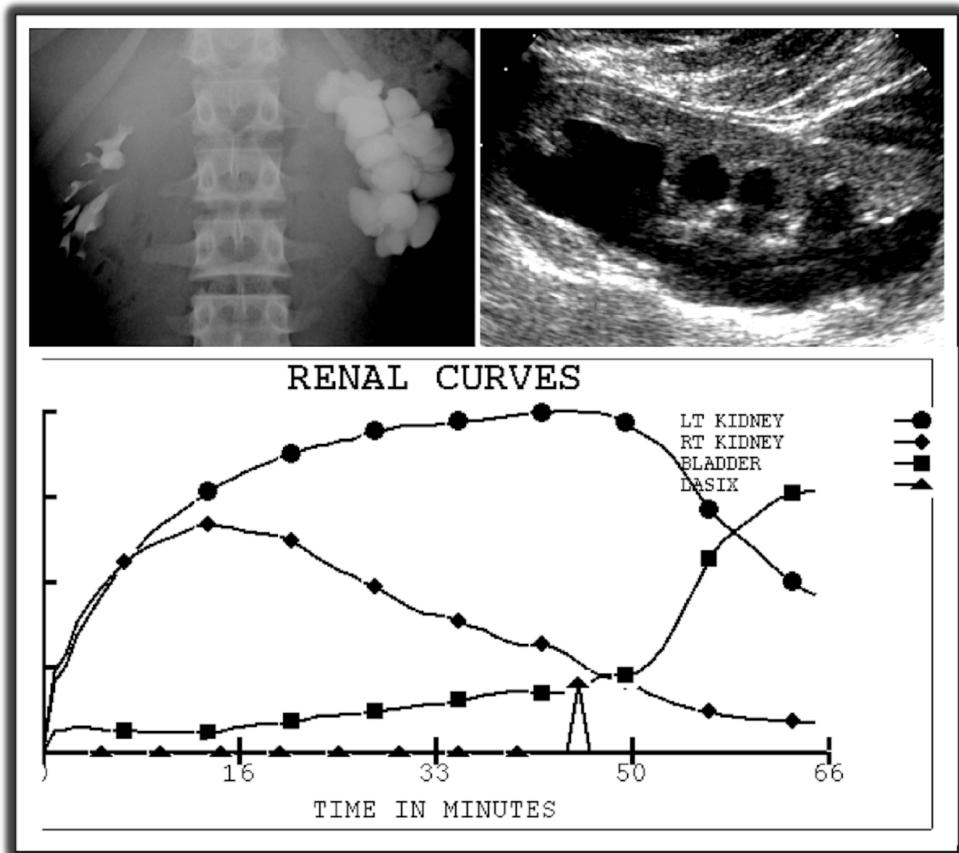


Figure 4: Megacalycosis. Increased number of dilated calyces on IVP (upper left) and renal ultrasound (upper right) without renal pelvis dilation. Preserved renal function with good drainage after lasix administration on diuretic renography (lower).

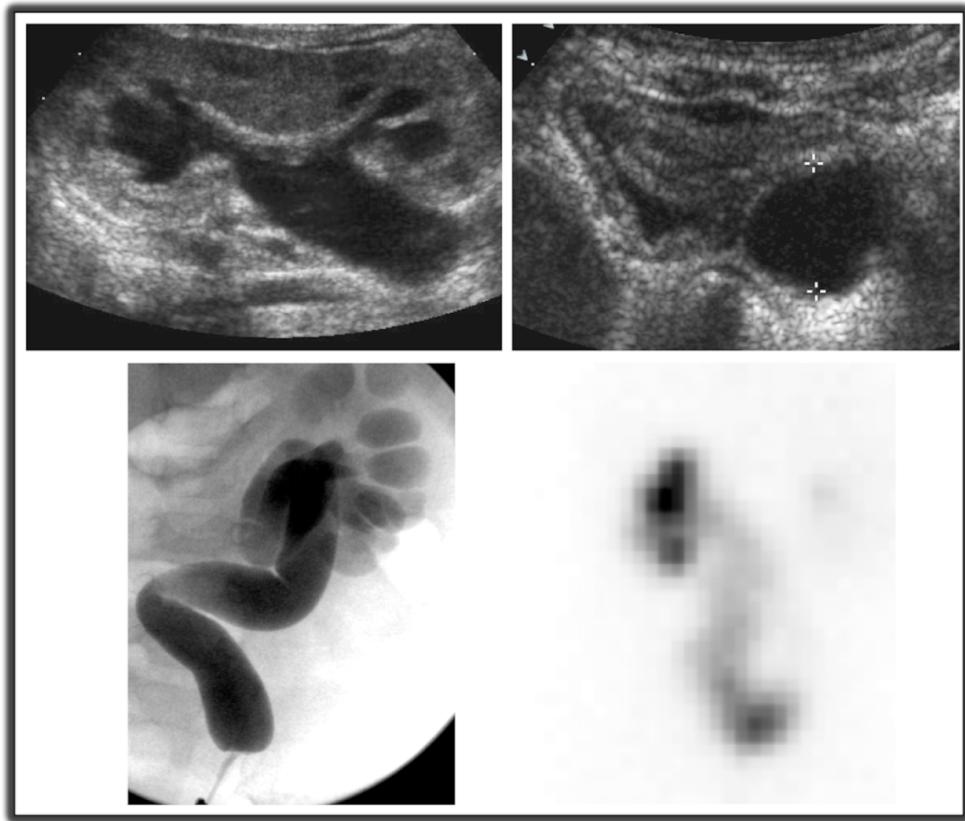
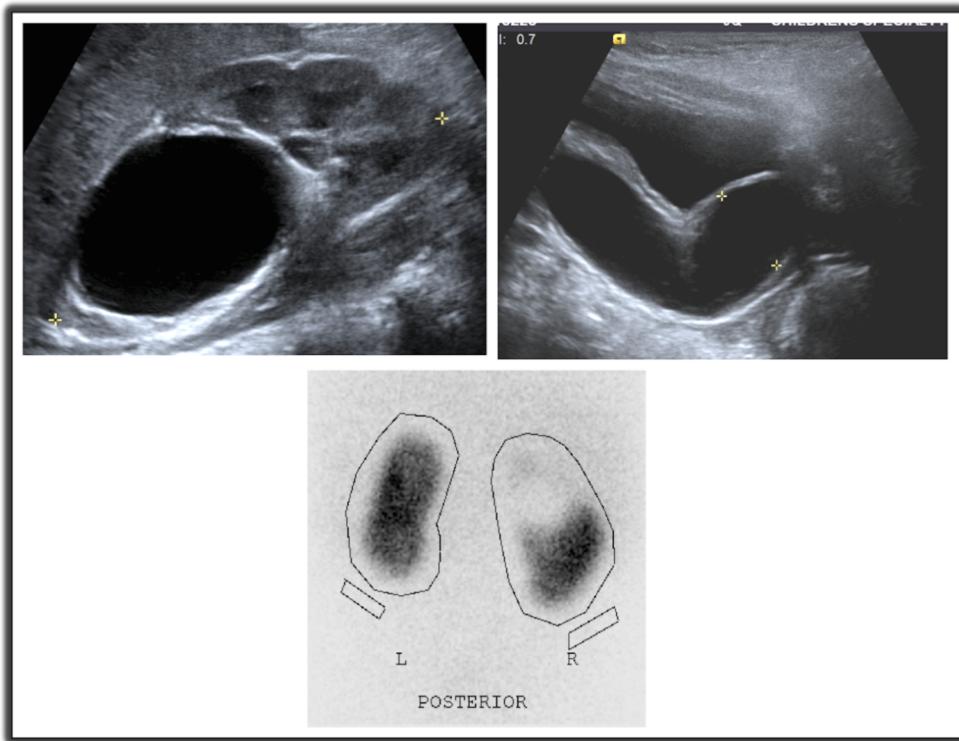


Figure 5: Megaureter. Hydronephrosis in kidney (upper left) and distal ureter (upper right) on ultrasound. Appearance of possible distal aperistaltic segment on retrograde pyelogram (lower left). Delayed drainage on diuretic renography in primary megaureter (lower right).

### 3.5 Ectopic Ureters



**Figure 6:** Appearance of duplex kidney (upper left) with upper pole ectopic ureter on ultrasound as well as distal ureter (upper right). Note lack of radiotracer uptake in upper pole consistent with associated renal dysplasia (lower).

Ectopic ureters drain to an abnormally caudal location (**Figure 6**). The estimated incidence is 1/1000 births with a female predominance. In girls, the ureteral orifice of an ectopic ureter may insert into the bladder neck, urethra, mullerian structures such as vagina or uterus, or the perineum. In boys, ectopic ureters may insert to the bladder neck, prostatic urethra, seminal vesical or vas deferens. In boys, the ectopic ureteral insertion is always proximal to the external sphincter, so urinary incontinence does not occur. By contrast, a vaginal ectopic ureter in a female may present clinically with continuous urinary incontinence.

In general, ureters with a more ectopic location—those inserting into the genital ducts, for example, tend to have greater associated renal dysplasia. Treatment options for ectopic ureter depend on the clinical picture and function of associated renal unit. Options may include observation, temporizing cutaneous ureterostomy, ipsilateral upper to lower ureteroureterostomy, ureteral reimplantation, among others (**Table 5**).

**Table 5: Upper pole renal duplication anomalies and management options**

	Management options	Pros	Cons
<i>Ectopic ureter</i>	Observation	Asymptomatic, may involute	1) UTI 2) urinary incontinence in females noted at toilet training
	Upper pole partial nephrectomy	Removes the abnormal tissue/definitively eliminates stasis	1) Risk of injury/loss of lower pole of kidney 2) removal of nephrons-typically little associated function
	Ipsilateral ureteroureterostomy	Avoids renal level and bladder level surgery	1) may have some persistent upper tract dilation 2) injury to lower pole ureter 3) theoretical creation of 'yo-yo' reflux
	Ureteral reimplantation	Directly addresses ectopic insertion	1) increased chance of obstruction/reflux with tapered

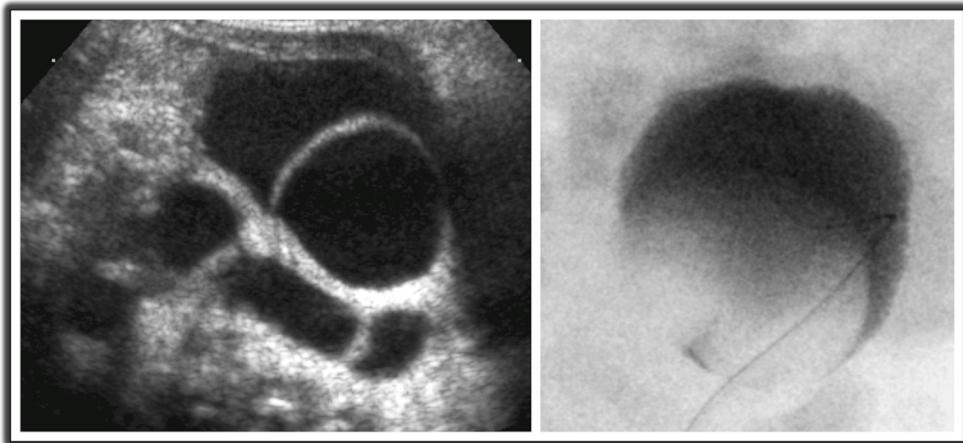
			ureteral reimplantation 2) injury to lower pole ureter 3) morbidity of bladder level surgery
<b>Ureterocele</b>	Observation	1) Asymptomatic 2) Definitive if upper pole is multicystic dysplastic and no bladder outlet obstruction	1) significant risk of UTI
	Ureterocele incision	1) minimally invasive 2) effectively relieves obstruction in >95% of cases decompression facilitates later reconstruction	Risk of iatrogenic upper pole reflux after incision of ectopic ureterocele is >50%
	Upper pole partial nephrectomy	1) removes poorly functioning hydronephrotic moiety 2) definitive procedure in >95% in absence of coexisting reflux	30-50% have coexisting reflux and recurrent infections requiring a second bladder level procedure

	<p>into other moieties 3) avoids bladder level surgery</p>	
Ureterocele excision and reconstruction	<p>Definitive - eliminates obstruction, reflux and reconstructs the bladder neck</p>	<p>Technically complex procedure with risks of persistent reflux, obstruction and bladder neck incompetence</p>

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### 3.6 Ureterocele

A ureterocele is a cystic dilation of the distal ureter at the bladder insertion site (**Figure 7**), seen in approximately **1/1000 births**.<sup>24</sup> Ureteroceles may be associated with single or duplex systems. In the setting of complete duplication, the ureterocele is associated with the upper moiety ureter, while reflux is common in the lower moiety ureter. Ureteroceles can be classified as intravesical if they are located entirely in the bladder or as ectopic if part of the ureterocele extends into the urethra. Given increased risk for urinary tract infections, ureteroceles are often punctured or incised endoscopically to allow drainage of potentially obstructed system. Surgical management of ureterocele and pyeloureteral duplication is summarized in **Figure 8**.<sup>25</sup>



**Figure 7:** Appearance of ureterocele on bladder ultrasound (left) and early filling with contrast on VCUG (right).

**Figure 8: Options for Definitive Surgical Management, Ureterocele**

	<b>Ideal Indications</b>	<b>Advantages</b>	<b>Limitations</b>
<b>Transurethral incision</b>	-Small infant -Large ureterocele with VUR	-Outpatient procedure* -Effective decompression -Occasionally definitive	-De-novo reflux into ureterocele segment necessitating subsequent lower tract reconstruction
<b>Upper pole nephrectomy</b>	No lower moiety VUR Nonfunctioning upper moiety	-May be definitive -Removes pathology -Avoids bladder surgery	-May still require lower tract reconstruction -Risk to lower moiety
<b>UU/ureteropyelostomy</b>	No lower moiety VUR Functional upper moiety	Drains obstructed segment with little risk for obstruction or UTI	Leaves ureterocele in bladder May develop VUR
<b>Common sheath reimplant with ureterocele excision</b>	Associated lower moiety VUR Functional upper moiety without significant dilation	Eliminates obstruction and VUR Removes ureterocele No renal risk	Complex surgery Risk to vagina and BN May require ureteral tapering

\*unless patient is an infant requiring admission for oxygen monitoring

**Figure 8: Options for Definitive Surgical Management, Ureterocele**

### 3.7 Pyeloureteral duplication

Complete or partial pyeloureteral duplication is the most common collecting system anomaly with an incidence of **1-5%**. Complete duplication results in two distinct ipsilateral ureteral orifices, two ureters and two collecting systems. The **Meyer-Weigert rule** refers to the tendency for the upper moiety ureter to drain to the more caudal orifice (with a tendency towards obstruction), and the lower moiety

ureter to drain to the more cranial/lateral orifice (with a tendency towards reflux), with rare exceptions.<sup>26</sup> Lower moiety ureters have an increased risk of VUR given the more lateral location on the bladder wall with shorter muscular tunnel. **When VUR is present in the lower moiety ureter, there is a characteristic “drooping lily” appearance on the VCUG (Figure 9).** Upper moiety ureters have an increased risk of obstruction from either an ureterocele or an ectopic ureter. Incomplete pyeloureteral duplication results in a Y-shaped ureter or bifid renal pelvis.

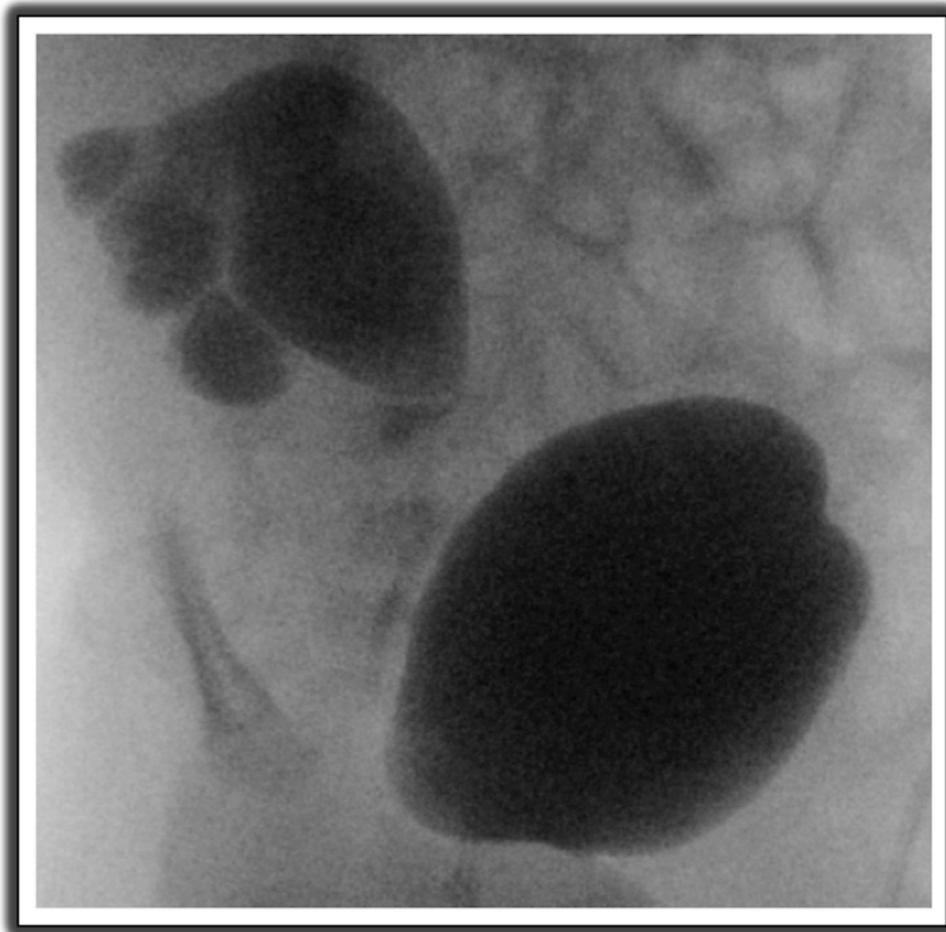


Figure 9: Lower pole reflux on VCUG in duplex kidney with “drooping lily” appearance

### 3.8 Vesicoureteral Reflux

See Core Curriculum **Vesicoureteral reflux**

VUR is the retrograde passage of urine from the bladder to the ureter that usually occurs during voiding when intravesical pressure rises (**Figure 10**) (**AUA guideline: Management and Screening of Primary Vesicoureteral Reflux** in children and **Urology Care Foundation: vesicoureteral reflux fact sheet**). VUR is detected in **30% of infants and young children presenting with a febrile UTI**.<sup>27</sup> Alternatively, VUR may be recognized during postnatal evaluation of prenatally-detected hydronephrosis.

VUR is classified as either primary or secondary. **Primary VUR** is thought to be caused by an

**incompetent ureterovesical junction (UVJ)** or a **short intravesical tunnel of the ureter** leading to incomplete mucosal coaptation during voiding. **Secondary VUR** is seen in children with high intravesical pressures due to **neurogenic bladder, dysfunctional voiding, or bladder outlet obstruction (e.g. posterior urethral valves)**. VUR is graded by the degree of retrograde flow on voiding cystourethrogram (VCUG) (**Table 6**) and outcomes are related to grade of VUR. Most low grade VUR (**Grade 1-2**) will spontaneously resolve without intervention while higher grades of VUR (**Grade 3-5**) have a lower likelihood of spontaneous resolution and higher likelihood of surgical intervention.<sup>28</sup> The AUA has published guidelines on management and screening of primary VUR in children (**AUA guidelines on Management and Screening of Primary Vesicoureteral Reflux in Children**) as well as a VUR patient guide (**Urology Care Foundation Vesicoureteral Reflux (VUR) Patient Guide**).

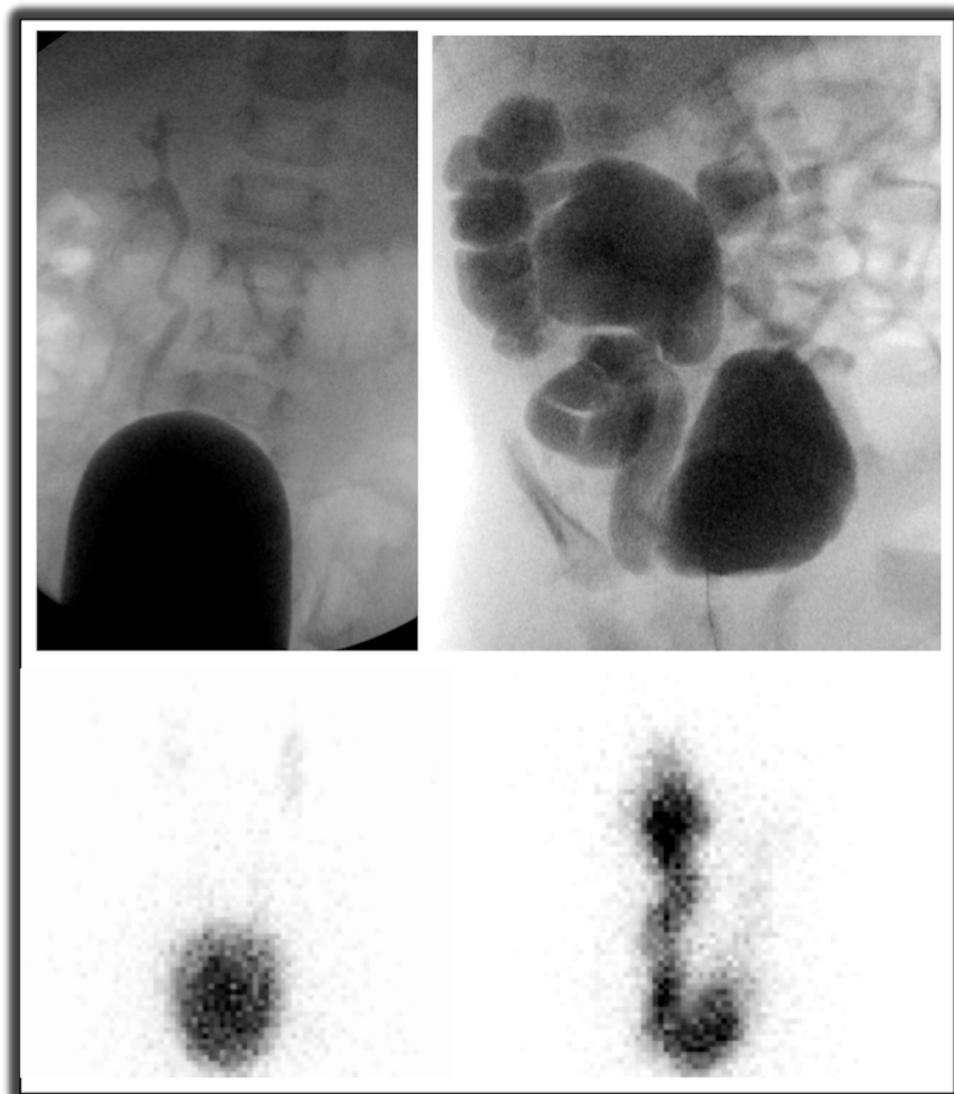


Figure 10: Examples of mild (left) and severe (right) VUR on VCUG (upper) and radionuclide cystogram (lower).

**Table 6: International Classification of Vesicoureteral Reflux**

Grade 1	Into a non-dilated ureter
Grade 2	Into non-dilated renal pelvis and calyces
Grade 3	Mild-moderate dilation of the ureter, renal pelvis and calyces with minimal blunting of calyces
Grade 4	Moderate ureteral tortuosity and dilates of pelvis and calyces
Grade 5	Grossly dilated and tortuous ureter, pelvis, and calyces with loss of papillary impressions
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## 4. Renal Migration and Fusion Anomalies

### 4.1 Ectopic Kidney

Disruptions in renal migration lead to ectopic kidneys, while renal fusion events result in the right and left renal units being abnormally paired together within the retroperitoneum. The kidneys normally ascend from a pelvic to lumbar location by 8 weeks of gestation. Failure of ascent leads to an ectopic kidney, with the bony pelvis representing the most common location (in 1/500-1200 births) (**Figure 11**).<sup>29</sup> Ectopic kidneys have an anomalous blood supply and tend to have anteriorly oriented renal pelvis. They also have an increased risk of renal dysplasia, vesicoureteral reflux (VUR), ureteropelvic junction obstruction (UPJO), nephrolithiasis, and urinary tract infection (UTI).<sup>30</sup> Rarely, an ectopic kidney can ascend too far and be located in the thoracic region.

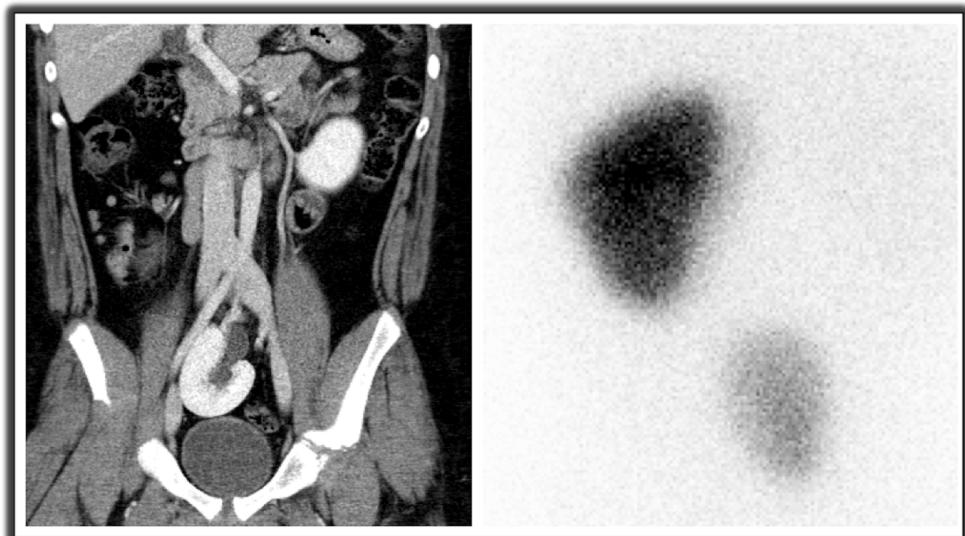


Figure 11: Ectopic pelvic kidney on CT (left) with evidence of decreased differential function on DMSA scan (right).

### 4.2 Renal Fusion

The most common fusion anomaly is a horseshoe kidney, occurring in **1/600** births (**Figure 12**).<sup>31</sup> The **inferior poles of the kidneys fuse** prior to ascent and the **inferior mesenteric artery limits further ascent** of the isthmus. Horseshoe kidneys do not rotate normally or ascend completely, so they have an anteriorly oriented pelvis as well as anomalous blood supply. As with ectopic kidneys, horseshoe kidneys have an increased risk of VUR, UPJO, nephrolithiasis, and UTI. Crossed renal ectopia occurs when both renal units are fused on the same side of the body, occurring in **1/1000 to 2000 births**. Crossed fused renal ectopia is a common example,<sup>29,32</sup> in which the ureter of the ectopic kidney crosses the body to insert on the contralateral trigone of the bladder. Other rare migration and fusion anomalies include crossed unfused renal ectopic and bilateral crossed renal ectopia.

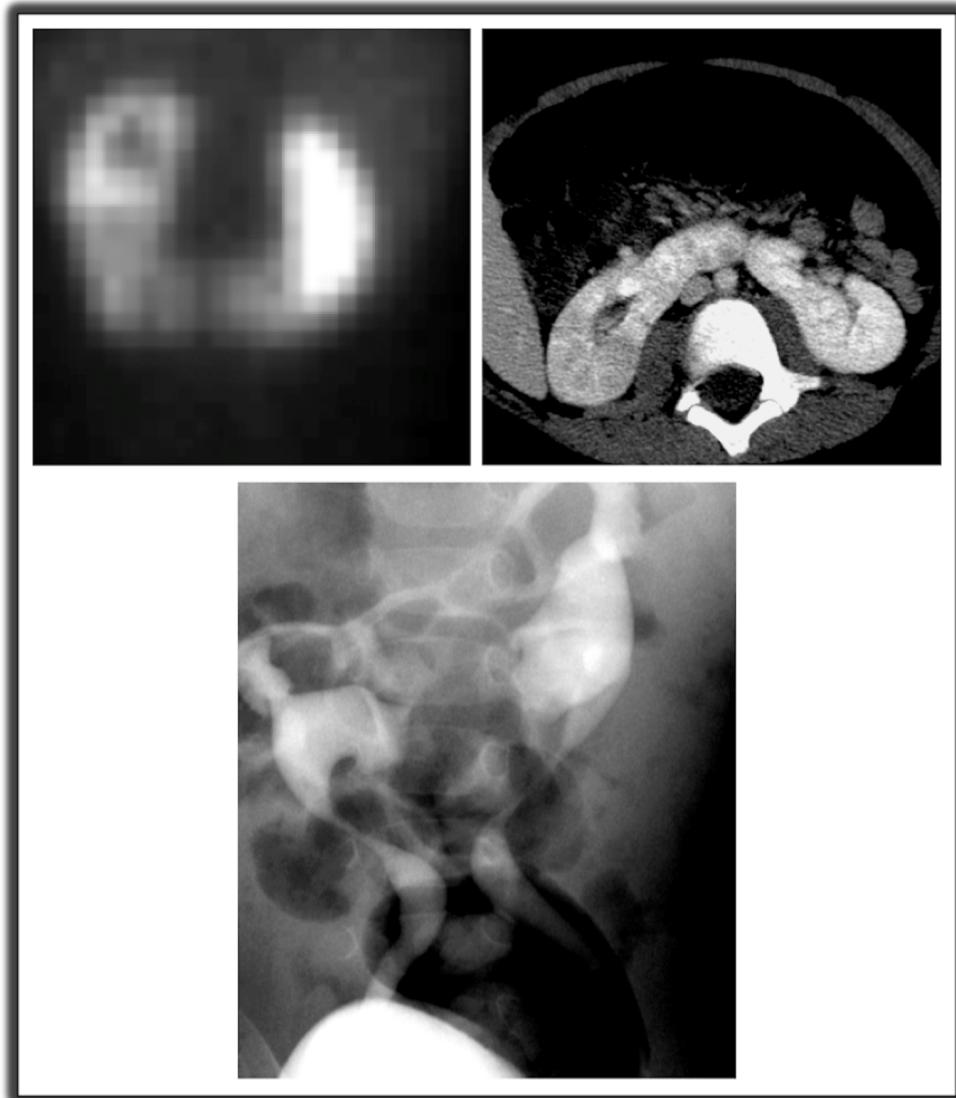


Figure 12: Images of horseshoe kidney on DMSA (upper left), CT (upper right), and VCUG (lower). Note the medially directed calyces on VCUG.

## 5. Lower Urinary Tract Obstruction Anomalies

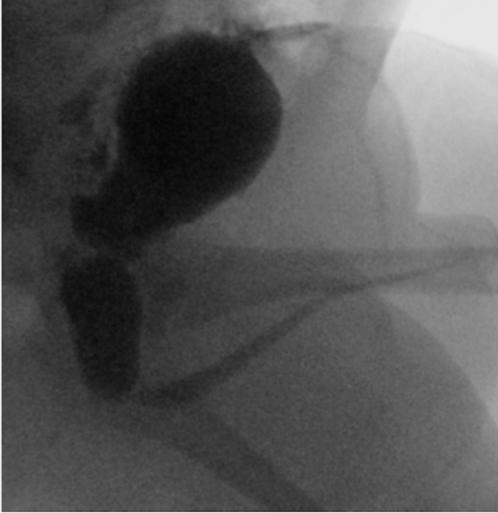
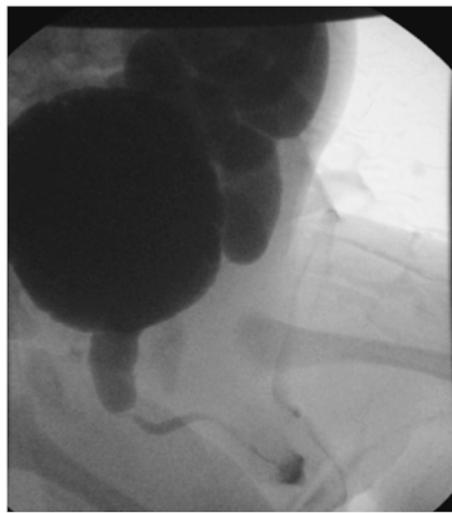
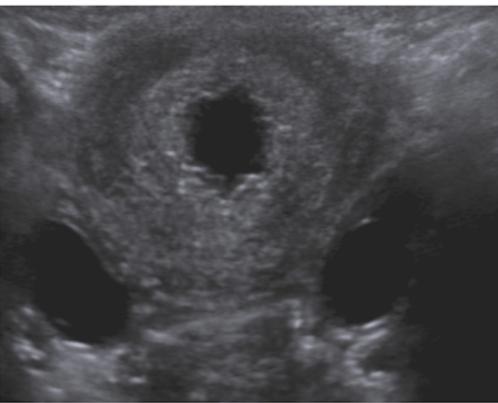
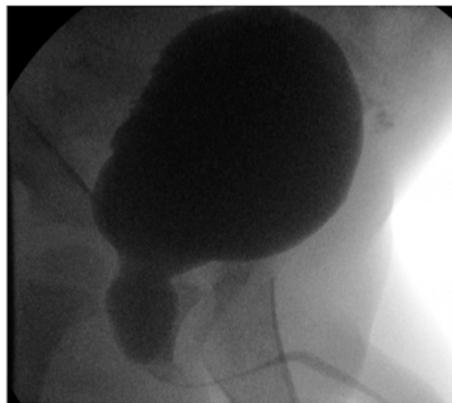


Figure 13: Examples of posterior urethral valves. Note possibility of high grade VUR (lower left) as well as small, trabeculated appearing bladder (lower right). The ultrasound (upper right) image demonstrates very thickened bladder with bilateral hydronephrosis that is evidence of bladder dysfunction.

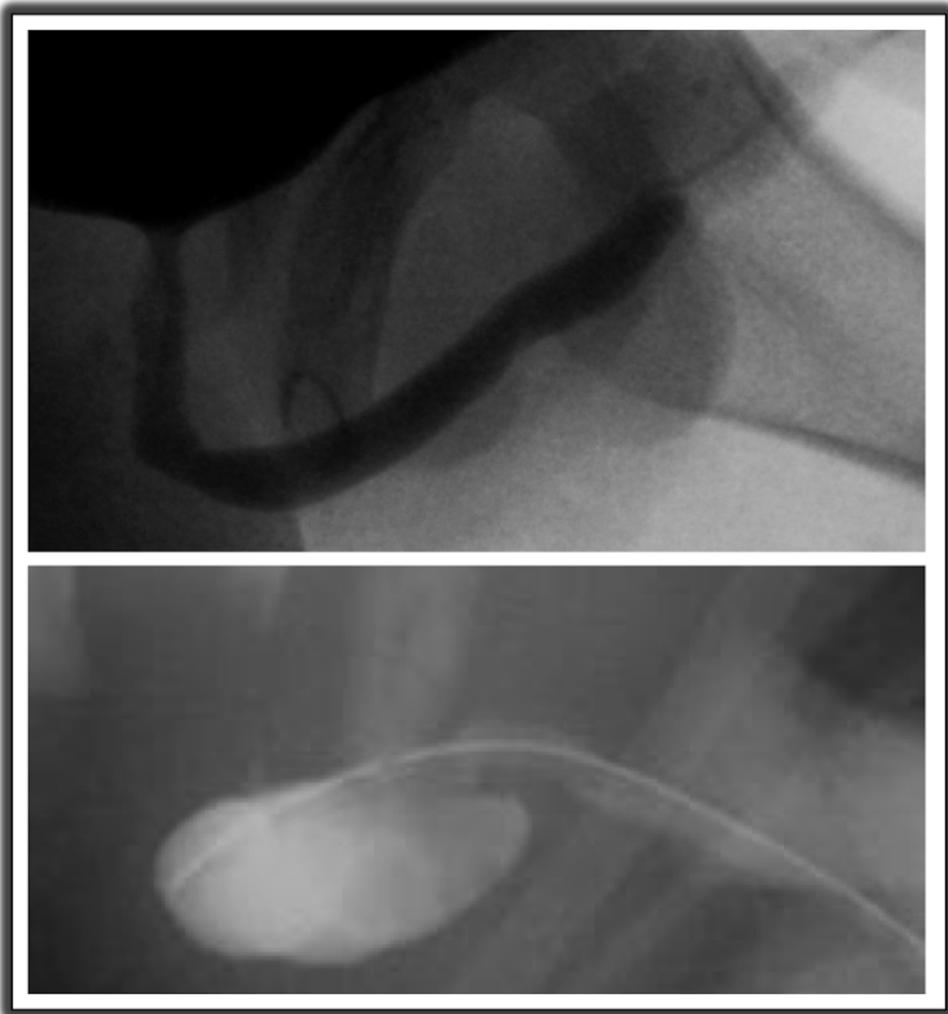


Figure 14: Anterior urethral valves on VCUG. Note the typical appearance of band of tissue in anterior urethra arising ventrally with proximal dilation on upper image in a child prior to treatment with endoscopic ablation. The lower image demonstrates a much larger urethral diverticulum.



Figure 15: Prune belly syndrome with typical appearance of abdominal wall laxity (upper images). IVP (lower left) and VCUG (lower right) demonstrating upper tract dilation and large bladder without obstruction respectively.

Congenital lower urinary tract obstruction can lead to poor clinical outcomes including severe bilateral hydroureteronephrosis with oligohydramnios or anhydramnios with fetal demise. The most common cause of congenital lower urinary tract obstruction is **posterior urethral valves (PUV)**. **Anterior urethral valves** and **urethral atresia** are much less common. **Prune Belly or Eagle Barrett syndrome (PBS)** is an additional congenital anomaly that can be confused with PUV, especially on antenatal ultrasound.

## 5.1 Posterior Urethral Valves (PUV)

See Core Curriculum **Posterior Urethral Valve**

PUV occurs in **1/5000-8000 pregnancies**,<sup>33</sup> thought to represent membranous folds in the posterior

urethra due to persistence of the urogenital membrane or improper cannulation of the urethra.<sup>34</sup> PUV results in a spectrum of obstructive severity, often with associated hydroureteronephrosis, secondary VUR, renal dysplasia, abnormal bladder function, and end stage renal disease (**Figure 13**). Clinical suspicion for PUV arises when significant bilateral hydroureteronephrosis and a dilated bladder / posterior urethra (“keyhole sign”) are seen on prenatal ultrasound. PUV are diagnosed via postnatal VCUG and are generally treated in the immediate post-natal period with **endoscopic posterior urethral valve ablation**. A syndrome of PUV, unilateral VUR with renal dysplasia, and relatively normal contralateral kidney at birth was previously thought to be associated with favorable long term renal outcomes but that is now controversial.<sup>35</sup> **Even with valve ablation, bladder dysfunction and progressive renal failure often occurs and therefore PUV patients need lifelong monitoring of renal and bladder function.**

## 5.2 Anterior Urethral Valves

Anterior urethral valves is a very rare entity (**less than 300 reported cases**)<sup>36</sup> caused by an obstructing web of ventrally-arising tissue in the anterior urethra. Anterior valves are often associated with a ventral urethral diverticulum. These lesions can also be associated with similar upper tract and bladder consequences as PUV and require close follow up. The treatment for anterior urethral valves is also **endoscopic ablation**. If there is a large associated diverticulum, excision of that diverticulum with urethroplasty and/or ureteral reimplantation may be needed (**Figure 14**).

## 5.3 Urethral Atresia

Urethral atresia is a very rare anomaly which may be associated with oligohydramnios, pulmonary hypoplasia, and fetal demise. A tragic outcome may be averted if the urine has an alternate route for drainage via a patent urachus or rectourethral fistula.<sup>37</sup> **Urethral atresia is most commonly identified in patients with prune belly syndrome**, though rare even in this group. In surviving patients, urethral atresia is typically managed with cutaneous vesicostomy until formal urethral reconstruction or urinary diversion can be undertaken.<sup>37</sup>

## 5.4 Prune Belly Syndrome (PBS)

PBS is a constellation of findings including abdominal wall muscle deficiency, genitourinary tract malformation characterized by dilation, and bilateral undescended testis (**Figure 15**). Most (95%) cases occur in males and occurs in **3-4/100,000 births**.<sup>38</sup> Collecting system anomalies seen with PBS include **large bladders that are not obstructed, dilated ureters, prostatic hypoplasia, and variable amounts of renal dysplasia**.<sup>39,40</sup> Interestingly, the hydroureter is often more severe distally, with relatively less dilation in the proximal ureter and kidney. Given the characteristic finding of dilated bladder and bilateral hydroureteronephrosis, PBS may be confused with PUV on prenatal ultrasound.

The cause of PBS is unknown but is thought to potentially be a defect in the development of the lateral plate mesoderm affecting the abdominal wall. Management of PBS generally includes bilateral

orchiopeoxies, and possible use of clean intermittent catheterization depending of presence of UTIs and bladder function, and abdominoplasty in select cases. There is varying degree of renal dysplasia in patients with PBS and chronic kidney disease is common.

## Presentations

### Urinary Tract Anomalies Presentation 1

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